Ocular Syndromes and Systemic Diseases

Fourth Edition

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Advice and suggestions given in this book are not meant to replace professional medical care. The reader is advised to consult his or her physician before undertaking any diet or exercise regimen and in order to gain answers about or treatment for any medical problems. The authors and publisher have made every effort to ensure that drug selection and dosage set forth in this text are in accord with current recommendations and practice at the time of publication. However, because the practice of medicine may change with ongoing research, changes in government regulations, and developments in medicine, the reader is encouraged to read the package insert for each drug for any change in indications and dosage and for added warnings and precautions. This is particularly important when the recommended agent is a new or infrequently employed drug.

Published in The United States of America.

To Mary Michelle, to Frederick, Kimberly, Robert,
Charles, Nichols, and Helena, and to Dr. Bruce Herndon,
who daily, throughout our joint eye residencies, asked me
about another "weird" syndrome

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Preface

This book has three main purposes: (1) to aid the busy ophthalmologist in identifying all ocular manifestations of a given syndrome, disease, or inherited disorder; (2) to provide references so that the ophthalmologist will know where to look for additional information; and (3) to promote greater observation of the characteristics of ocular syndromes, systemic diseases, and inherited disorders. This book is designed for quick reference while the patient's eyes are being dilated or immediately following an examination. Practicality is the objective; conciseness, logical arrangement, and authenticity are the main features.

I have endeavored to include all systemic syndromes, diseases, and inherited disorders that have ocular manifestations. The titles are arranged alphabetically and are numbered. Other names of the same disorder are also given. A general definition, clinical ocular findings, clinical systemic findings, and several references are included for each entry. Each entry is dealt with as completely as space and practicality allow. Laboratory findings have not been considered; the book is designed to serve as the ophthalmologist's primary source of information. If any syndromes, systemic diseases, or inherited disorders with ocular manifestations have been omitted, I urge you to inform me of this so that any future editions of the book may include them.

The *Ocular Differential Diagnosis* book provides comprehensive lists of causes for symptoms or findings. Frequently more information is needed, and it is hoped that, *Ocular Syndromes and Systemic Diseases* will furnish the physician with additional information to make a better diagnosis.

I am indebted to many individuals who assisted with the preparation of this manuscript, including: Renee Tindall, Dr. Fernando Murillo, Dr. Scott Lowery, Dr. Kae Chatman, Angie Brown, and Louise Geer.

A

A Esotropia Syndrome 1

Esotropia greater looking up by 15 prism diopters than looking down; an overaction of superior oblique muscles or underaction of inferior rectus muscles; fusion may be obtained by chin elevation; mongoloid (upward) slant of lid fissures; may be accommodative, nonaccommodative, or paralytic esotropia components.

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Hwang J, Wright KW. Strabismus syndromes. In: Wright KW, ed. *Pediatric Ophthalmology and Strabismus*. St. Louis: Mosby, 1995:223.

Roy FH. *Practical management of eye problems: glaucoma, strabismus, visual fields*. Philadelphia: Lea & Febiger, 1975:32-134.

A Exotropia Syndrome 2

Exotropia greater looking down by 15 prism diopters than looking up; mongoloid (upward) slant of lid fissures; alternating sursumduction and associated vertical divergence; overaction of superior oblique muscles or underaction of inferior oblique or inferior rectus muscles; fusion obtained by chin depression.

Hardesty HH. Superior oblique tenotomy. *Arch Ophthalmol* 1972; 88:181-184.

Hwang J, Wright KW. Strabismus syndromes. In: Wright KW, ed. *Pediatric Ophthalmology and Strabismus*. St. Louis: Mosby, 1995:223.

Roy FH. *Practical management of eye problems: glaucoma, strabismus, visual fields*. Philadelphia: Lea & Febiger, 1975:144-145.

Aarskog Syndrome (Facial-Digital-Genital Syndrome) 3

General: X-linked recessive; males fully affected; females exhibit partial features; normal birth weight and length.

Ocular: Telecanthus; hypertelorism; unilateral or bilateral blepharoptosis; strabismus; hyperopic astigmatism; large cornea.

Clinical: Short stature; triangular facies; deformity of hands and feet; anomalies of external genitalia; inguinal hernia; protruding umbilicus; abnormal cervical vertebrae; cryptorchidism.

Kirham TH, et al. Ophthalmic manifestations of Aarskog (facial-digital-genital) syndrome. *Am J Ophthalmol 1975;* 79:441-445.

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Porteous MEM, Goudie DR. Aarkog syndrome. *J Med Genet* 1991; 28:44-47.

Aarskog-Scott Syndrome (Faciogenital Dysplasia) 4

General: Sex-linked; characterized by ocular hypertelorism, anteverted nostrils, broad upper lip, and saddlebag scrotum.

Ocular: Ptosis; hypertelorism.

Clinical: Hyperextensibility of fingers; genu recurvatum; flat feet; hypermobility in cervical spine with neurologic deficit; cleft lip and palate; anteverted nostrils; broad upper lip; abnormal penoscrotal relations; "saddlebag scrotum."

Bowls E, et al. Aarskog-Scott syndrome: full male and female expression associated with an X autosome translocation. *Am J Med Genet* 1984; 17:595-602.

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Scott EI. Unusual facies, joint hypermobility, genital anomaly and short stature: a new dysmorphic syndrome. *Birth Defects* 1971; 6:240-246.

Abdominal Typhus (Enteric Fever; Typhoid Fever) 5

General: Causative agent, *Salmonella typhi.*

Ocular: Conjunctivitis; chemosis; corneal ulcer; tenonitis; paralysis of extraocular muscles; endophthalmitis; panophthalmitis; optic neuritis; retinal detachment; central scotoma; central retinal artery emboli; iritis with or without hypopyon; choroiditis; retinal hemorrhages; bilateral optic neuritis; abnormal ocular motility (likely secondary to thrombotic infarcts affecting the ocular motor nerve nuclei, fascicles, brainstem, or cerebral hemispheres).

Clinical: Fever; headache; bradycardia; splenomegaly; maculopapular rash; leukopenia; encephalitis. Salmonella may produce an illness characterized by fever and bacteremia without any other manifestations of enterocolitis or enteric fever, which is particularly common in patients with acquired immunodeficiency syndrome (AIDS).

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Imperato PJ. *The treatment and control of infectious diseases in man.* Springfield, IL: Charles C. Thomas, 1974: 154-161.

Jacobs JL, et al. Salmonella infections in patients with the acquired immunodeficiency syndrome. *Ann Intern Med* 1985; 102:186-188.

Mathur JS, et al. Post typhoid retinal detachment. *J All India Ophthalmol Soc* 1970; 18:135-137.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995:3100.

Aberfeld Syndrome (Schwartz-Jampel Syndrome; CongenitalBlepharophimosis Associated with Generalized Myopathy Syndrome; Ocular and Facial Abnormalities Syndrome) 6

General: Etiology not known; autosomal recessive inheritance, although there are reports of dominant inheritance; progressive disorder.

Ocular: Blepharophimosis; exotropia; myopia; congenital cataracts; microcornea.

Clinical: Myopathy; bone deformities; arachnodactyly; dwarfism; hypoplastic facial bones; hypertrichosis; kyphoscoliosis.

Aberfeld DC, et al. Myotonia dwarfism, diffuse bone disease and unusual ocular and facial abnormalities (a new syndrome). *Brain* 1965; 88:313.

Keating PD, Hepler RS. Blepharophimosis and acquired somato-facial dysmorphism associated with congenital cataracts. *Arch Ophthalmol* 1969; 82:1.

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Pascuzzi RM. Schwartz-Jampel syndrome with dominant inheritance. *Muscle Nerve* 1990; 13:1152-1163.

Acanthamoeba 7

General: Caused by *Acanthamoeba polyphaga* and *Acanthamoeba cartel* (see Herpes Simplex Masquerade Syndrome); all types of contact lenses have been associated with acanthamoeba keratitis, particularly daily-wear soft contact lenses.

Ocular: Hypopyon; uveitis; conjunctivitis and chemosis; keratitis; pannus; corneal ring abscess; papillitis; vitreitis; retinal perivasculitis; secondary glaucoma; postkeratoplasty acanthamoeba keratitis may present as an infectious crystalline keratopathy in the periphery of the graft.

Clinical: Meningoencephalitis; meningitis; hemorrhagic encephalitis.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

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Nagington J, et al. Amoebic infection of the eye. *Lancet* 1974;2:1537-1540.

Pavan-Langston D. Amoeba. In: Pavan-Langston D, ed. *Manual of ocular diagnosis and therapy*, 4th ed. Boston: Little, Brown and Company, 1995:95-96.

Samples JR, et al. Acanthamoeba keratitis possibly acquired from a hot tub. *Arch Ophthalmol* 1984; 102:707-710.

Watson PG. Amebic infection of the eye. *Trans Ophthalmol Soc U K* 1975; 95:204-206.

Wilhelmus KR. Parasitic keratitis and conjunctivitis. In: Smolin G, Thoft RA, eds. *The Cornea*, 3rd ed. Boston: Little, Brown and Company, 1994:262-266.

Acanthosis Nigricans 8

General: Rare skin disease of unknown etiology; occurs at any age; equal frequency in males and females; thickening and hyperpigmentation of the skin of the entire body, especially in flexural areas; autosomal dominant inheritance.

Ocular: Conjunctivitis; pigmentation of palpebral conjunctiva; tumors on lids and lid margins; madarosis; trichiasis.

Clinical: Hyperkeratotic lesions on face, neck, oral mucosa, axillae, groin, antecubital fossae, and umbilicus.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

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Tasjian D, et al. Familial acanthosis nigricans. *Arch Dermatol* 1984; 120:1351-1354.

Whitmore PV. Skin and mucous membrane disorders. In: Duane TD, ed. *Clinical Ophthalmology, vol. V.* Philadelphia: JB Lippincott, 1982:9-10.

Accommodative Insufficiency (Accommodative Effort Syndrome) 9

General: Male or female; 10 to 35 years of age.

Ocular: Asthenopia with near vision appearing within a few minutes after reading, sewing, or observing a near object; increased amplitude of accommodative adduction; abnormal relaxation of accommodation induced by relative divergence at close distances; latent convergence insufficiency; may result secondary to ciliary dysfunction associated with Adie pupil.

Clinical: It may be secondary to a systemic disorder such as Parkinson disease or oral lithium.

Brown B. The convergence insufficiency syndrome masquerade. *Am Orthoptic J* 1990; 40:94-97.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Wright KW. Exotropia. In: Wright KW, ed. *Pediatric Ophthalmology and Strabismus.* St. Louis: Mosby, 1995:200.

Aceruloplasminemia 10

General: Autosomal recessive, adult-onset

Ocular: Maculopathy which resembles aging macular degeneration

Clinical: Associated with increased levels of iron in the retina

Dunaief, JL, Richa C, Franks EP, et al. Macular degeneration in a patient with aceruloplasminemia, a disease associated with retinal iron overload. *Ophthalmology* 2005; 112: 1062-1065.

Achard Syndrome 11

General: All features of Marfan syndrome, with the addition of dysostosis mandibulofacialis; arachnodactyly; receding lower jaw; joint laxity limited to the hands and feet; differs from Marfan syndrome in that the skull is broad and brachycephalic with small mandible.

Ocular: Myopia; lens dislocation; spherophakia.

Clinical: Mandibulofacial dysostosis; skeletal anomalies; arachnodactyly; high arched palate; heart disease.

Achard D. Arachnodactylie. *Bull Mem Soc Med Hop Paris* 1902; 19:834.

Jackson LG. Genetic principles for the ophthalmologist. *Trans Am Acad Ophthalmol Otolaryngol* 1974; 27:20.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

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Achondroplasia 12

General: Dwarfism; etiology unknown; occurs in both sexes; inheritance is autosomal dominant with almost complete penetrance; characterized by rhizomelic dwarfism (reduction most marked in the proximal limbs); mid-face hypoplasia; exaggerated lumbar lordosis; limitation of hip and elbow expansion; location of achondroplastic gene reported to be in the short arm of chromosome 1.

Ocular: Strabismus; optic atrophy; hypermetropia.

Clinical: Osseous impingement upon cranial nerves; rhizomelic short stature; facial features include frontal bossing, depressed nasal bridge, relative mandibular prognathism; connective tissue dysplasia; hypotonic in infancy; paraplegia may develop in second or third decade.

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Griffin JR, et al. Optometric screening in achondroplasia, diastrophic dysplasia, and spondyloepiphyseal dysplasia congenita. *Am J Optom Physiol Opt* 1980; 57:118-123.

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Verloes A, et al. Neuroblastoma in a dwarfed newborn: possible clue to the chromosomal localization of the gene for achondroplasia? *Am Genet* 1991; 34:25-26.

ACHOO Syndrome (Autosomal Dominant Compelling Helio-Ophthalmic Outburst Syndrome; Photic Sneeze Reflex; Sneezing from Light Exposure; Peroutka Sneeze) 13

General: Autosomal dominant; sneezing in response to bright light, especially sunlight; association between photic sneeze and nephropathic cystinosis has been reported.

Ocular: Photic sneeze reflex by sudden exposure of dark-adapted subject to bright light.

Clinical: Successive sneezing, as many as 43 in a row.

Collie WR, et al. ACHOO syndrome. *Birth Defects* 1978; 14:361-363.

Katz B. et al. Photic sneeze reflex in nephropathic cystinosis. *Br J Ophthalmol* 1990; 74:706-708.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Acinetobacter (Mima Polymorpha; Acinetobacter Iwoffi) 14

General: Gram-negative pleomorphic bacillus *Mima;* generally occurs in patient with lowered resistance.

Ocular: Conjunctivitis and chemosis; corneal ulcer; blepharitis; iris prolapse; endophthalmitis.

Clinical: Meningitis; pneumonitis; endocarditis; urethritis; vaginitis; arthritis; dermatitis; intracranial abscess; subdural empyema.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1. 4th ed. Baltimore: Williams & Wilkins, 1995:2993.

Pavan-Langston D. Cornea and external disease. Bacterial corneal ulcers. In: Pavan-Langston D, ed. *Manual of ocular diagnosis and therapy,* 4th ed. Boston: Little, Brown and Company, 1995:76-79.

Schechter RJ. Treatment of Mima polymorpha conjunctivitis. *Arch Ophthalmol* 1976; 94:338-339.

Wand M, et al. Corneal perforation and iris prolapse due to Mima polymorpha. *Arch Ophthalmol* 1974; 93:239-241.

Ackerman Syndrome 15

General: Autosomal recessive; characterized by pyramidal molar roots.

Ocular: Juvenile glaucoma.

Clinical: Unusual upper lip and dental roots; pyramidal molar roots.

Ackerman JL, et al. Taurodont, pyramidal and fused molar roots associated with other anomalies in a kindred. *Am J Phys Anthropol* 1973; 38:681-694.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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ACL Syndrome (Acromegaloid, Cutis Verticis Gyrata, Corneal Leukoma Syndrome) 16

General: Autosomal dominant; rare; three features include cutis verticis, associated with acromegaly and corneal leukoma; onset by age I year.

Ocular: Bilateral corneal leukoma; keratitis.

Clinical: Unusually tall; large hands, feet, and chin; skin of hands very soft; skin of scalp lies in folds; frontal bosses; ear calcification; pituitary tumors; abnormal dermal ridge patterns; enlargement of supraorbital arch of frontal bone.

Antine BE, et al. Fibroma of the cornea. *Arch Ophthalmol* 1974; 91:278.

Kenyon KR. Mesenchymal dysgenesis in Peter anomaly, sclerocornea and congenital endothelial dystrophy. *Exp Eye Res* 1975; 21:124.

Acne Rosacea (Acne Erythematosa; Ocular Rosacea) 17

General: Etiology unknown; usually occurs in women 30 to 50 years of age; pathogenetic mechanism remains unclear.

Ocular: Conjunctivitis; corneal neovascularization (wedge-shaped); keratitis; meibomianitis; blepharitis; recurrent chalazion; conjunctival hyperemia; superficial punctate keratopathy; corneal vascularization, thinning, perforation, and scarring; episcleritis; scleritis; iritis; nodular conjunctivitis.

Clinical: Symmetrical erythema; papules; pustules; telangiectasia; sebaceous gland hypertrophy of the forehead, malar eminences, and nose.

Browning DJ, et al. Ocular rosacea in blacks. *Am J Ophthalmol* 1986; 101:441-444.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:267-268.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hozng-Xuan T, et al. Ocular rosacea. *Ophthalmology* 1990; 97:1468.

Marmion VJ. Tetracycline in the treatment of ocular rosacea. *Proc Roy Soc Med* 1969; 62: 11-12.

Mondino BJ. Clinical immunologic diseases. Dermatologic disease. In: Smolin G, Thoft RA, eds. *The Cornea*, 3rd ed. Boston: Little, Brown and Company, 1994:428-431.

Pavan-Langston D. Rosacea (acne rosacea). In: Pavan-Langston D, ed. *Manual of ocular diagnosis and therapy.* Boston: Little, Brown and Company, 1995:104-105.

Roper-Hall MJ. The ocular aspects of rosacea. *Trans Ophthalmol Soc U K* 1966; 86:727-732.

Acosta Syndrome (Mountain Sickness; Mountain Climber Syndrome; Monge Syndrome; Soroche Syndrome) 18

General: Cause is cerebral hypoxia at high altitudes; Monge syndrome is the chronic form of mountain sickness.

Ocular: Acute blurred vision; difficulties in color discrimination; impaired light adaptation; retinal hemorrhage; chronic lid edema; bluish scleral injection; decreased visual acuity.

Clinical: Restlessness and irritability; headaches; impaired judgment at approximately 15,000 feet; confusion, cyanosis, muscular incoordination and possible loss of consciousness at approximately 18,000 to 20,000 feet; exertional dyspnea; epistaxis; gum bleeding; hemoptysis; anorexia; nausea; vomiting; tinnitus; cough; loss of libido; paresthesian extremities; coma; clubbing of fingers; hepatosplenomegaly.

Monge C. High altitude disease. *Arch Intern Med* 1937; 59:32.

Shults WT, Swan KC. High altitude retinopathy in mountain climbers. *Arch Ophthalmol* 1975; 93:404.

Wiedman M. High altitude retinal hemorrhage. *Arch Ophthalmol* 1975; 93:401.

Acquired Immunodeficiency Syndrome (AIDS; Acquired Cellular Immunodeficiency; Acquired Immunodeficiency) 19

General: Acquired breakdown of the immune system followed by disease that takes advantage of the body's collapsed defenses; acquired by shared drug needles or sexual intercourse; occurs most frequently in homosexually active men (75%), intravenous drug abusers (13%), and Haitian immigrants (6%).

Ocular: Retinal cotton-wool spots; cytomegalovirus retinitis; retinal periphlebitis; conjunctival Kaposi sarcoma; necrotizing retinitis; retinal hemorrhages; conjunctivitis sicca; orbital Burkitt lymphoma; peripheral retinochoroiditis; vitreitis; fungal corneal ulcer; hypopyon; acute glaucoma; third nerve palsy; anterior uveitis; atypical retinitis; orbital pseudotumor; herpes zoster ophthalmicus; herpes simplex keratitis; bacterial keratitis; molluscum contagiosum; cytomegalovirus retinitis; toxoplasma retinitis; acute retinal necrosis; human immunodeficiency virus (HIV) retinitis; syphilitic retinitis; *Pneumocystis carinii* choroiditis; fungal and bacterial endophthalmitis; fungal choroiditis; conjunctival microvasculopathy; keratitis sicca; subconjunctival hemorrhage.

Clinical: Because of lowered immunity, one third develop Kaposi sarcoma; pneumonia caused by *Pneumocystis carinii;* death.

Antworth MV, Beck RW. Third nerve palsy as a presenting sign of acquired immune deficiency syndrome. *J Clin Neural Ophthalmol* 1987; 7:125.

Benson WH, et al. Orbital pseudotumor in a patient with AIDS. *Am J Ophthalmol* 1988; 105:697-698.

Brooks HL, et al. Orbital Burkitt's lymphoma in a homosexual man with acquired immune deficiency. *Arch Ophthalmol* 1984; 102:1533-1537.

Fay MT, et al. Atypical retinitis in patients with the acquired immunodeficiency syndrome. *Am J Ophthalmol* 1988; 105:483-490.

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Khadem M, et al. Ophthalmologic findings in acquired immune deficiency syndrome. *Arch Ophthalmol* 1984; 102:201-206.

McMullen WW, D'Amico DJ. AIDS and its ophthalmic manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3102-3120.

Parrish CM, et al. Spontaneous fungal corneal ulcer as an ocular manifestation of AIDS. *Am J Ophthalmol* 1987; 104:302.

Pflugfelder SC, et al. Peripheral corneal ulceration in patient with AIDS-related complex. *Am J Ophthalmol* 1987; 104:542-543.

Sandor EV, et al. Herpes zoster ophthalmicus in patients at risk for the acquired immune deficiency syndrome (AIDS). *Am J Ophthalmol* 1986; 101:153-155.

Williams AS, et al. AIDS presenting as acute glaucoma. *Arch Ophthalmol* 1988; 106:311-312.

Acquired Lues (Syphilis; Acquired Syphilis; Lues Venerea; Malum Venereum) 20

General: Causative agent, *Treponema pallidum*, usually transmitted sexually.

Ocular: Conjunctival chancroid; conjunctivitis; keratitis; blepharitis; ptosis; iris atrophy; hippus; dacryocystitis; optic nerve atrophy; optic neuritis; periostitis; episcleritis; scleritis; nystagmus; uveitis; vitreous hemorrhages; paralysis of sixth nerve; papilledema; retinal hemorrhages; retinitis proliferans; oculogyric crisis; neuroretinitis; papilledema (associated with aseptic meningitis); diffuse or multifocal chorioretinitis; vertical supranuclear gaze palsy; Benedikt syndrome.

Clinical: Primary lesion associated with regional lymphadenopathy; secondary bacteremic stage associated with generalized mucocutaneous lesions; tertiary stage characterized by destructive mucocutaneous, musculoskeletal, or parenchymal lesions, aortitis, or central nervous system disease; syphilis and HIV infection often coexist in the same patient who experiences a higher incidence and greater severity of neurologic and ocular manifestations; a significant percentage of patients infected with HIV-I and *T pallidum* become seronegative to syphilis testing.

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Halperin LS. Neuroretinitis due to seronegative syphilis associated with human immunodeficiency virus. *J Clin Neuro-Ophthalmol* 1992;12:171-172.

Ho AC, et al. Ocular syphilis: *Clinical* manifestations and recent observations. *Semin Ophthalmol* 1993;8:53-60.

Lukehart SA, Holmes KK. Spirochetal diseases, syphilis. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:726-37.

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Spektor FE, et al. Granulomatous conjunctivitis secondary to Treponema pallidum. *Ophthalmology* 1981; 88: 863-865.

Acrodermatitis Chronica Atrophicans 21

General: Rare familial skin disorder; autosomal recessive; both sexes equally affected; occurs in infants; not present at birth but develops during first few weeks; zinc deficiency; there are reports of improvement following treatment with zinc suggesting an abnormality in the zinc-binding factor.

Ocular: Scarring of the conjunctiva; recurrent iridocyclitis; keratomalacia; cataracts; photophobia; blepharitis; punctal stenosis; corneal opacification.

Clinical: Vesiculobullous eruption around body orifices, skin of knees, elbows, and paronychial areas; complete alopecia; erythematous psoriasiform plaques.

Feldberg R, et al. Keratomalacia in acrodermatitis enteropathica (AE). *Metab Pediatr Ophthalmol* 1981; 5:207-211.

Matta CS, et al. Eye manifestations in acrodermatitis enteropathica. *Arch Ophthalmol* 1975; 93:140-142.

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Racz P, et al. Bilateral cataract in acrodermatitis enteropathica. *J Pediatr Ophthalmol Strabismus* 1979; 16:180-182.

Whitmore PV. Skin and mucous membrane disorders. In: Duane TD, ed. *Clinical Ophthalmology, vol. V.* Philadelphia: JB Lippincott, 1982:6-7.

Acropachy (Hypertrophic Pulmonary Osteoarthropathy) 22

General: Three separate components: clubbing of fingers, periosteal proliferation of distal ends of long bones, and arthritis; symptoms disappear with control of disease.

Ocular: Exophthalmos.

Clinical: Finger edema; fibrous overgrowth to fingertips; nail deformity; elevated hormone levels; periosteal bone changes affecting distal radius, ulna, tibia, fibula, metacarpals, and phalanges.

Duke-Elder S, MacFaul PA. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1971.

Robbins SL. *Pathologic Basis of Disease*. Philadelphia: WB Saunders, 1974.

Acrorenoocular Syndrome 23

General: Autosomal dominant; Duane syndrome with radial defects.

Ocular: Complete coloboma; coloboma of optic nerve; ptosis and Duane anomaly.

Clinical: Renal anomalies; hypoplasia of distal part of thumb with lack of motion at phalangeal joint; renal ectopia without fusion; bladder diverticula; malrotation of both kidneys; absence of kidney; clubhand or absence of thumb.

Halal F, et al. Acro-renal-ocular syndrome: autosomal dominant thumb hypoplasia; renal ectopia and eye defect. *Am J Med Genet* 1984; 17:753-762.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Actinomycosis 24

General: Gram-positive *Actinomyces israelii.*

Ocular: Hypopyon; conjunctivitis; keratitis; corneal ulcer; proptosis; uveitis; dacryocystitis; yellow nodules on conjunctiva and eyelids; occlusion of nasolacrimal canaliculi; canaliculitis; orbital abscess; endophthalmitis (rare).

Clinical: Chronic inflammatory induration and sinus formation.

Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine*, 11th ed. New York, McGraw-Hill, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hussain I, et al. Canalicular infection caused by Actinomyces. *Eye* 1993; 7[Pt 4]:542-544.

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Sullivan TJ, et al. Actinomycosis of the orbit. *Br J Ophthalmol* 1992; 76:505-506.

Acute Follicular Conjunctivitis (Adenoviral Conjunctivitis; Pharyngoconjunctival Fever; Syndrome of Beal) 25

General: Infectious disease produced by adenovirus; serotypes 3, 4, 7, 8, 19, 37, and several others may cause acute conjunctivitis with or without upper respiratory tract involvement; epidemic keratoconjunctivitis has been reported worldwide associated with 11 virus serotypes, with serotypes 8, 11, and 19 being the most commonly responsible.

Ocular: Conjunctivitis; chemosis; keratitis; blepharitis; blepharospasm.

Clinical: Fever; pharyngitis; lymph node enlargement; malaise; myalgia; headache; diarrhea.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Thygeson P. Follicular conjunctivitis. In: Allen RF, et al., eds. *Infectious diseases of the conjunctiva and cornea.* St. Louis: Mosby, 1963: 103-111.

Acute Frosted Retinal Periphlebitis 26

General: Etiology unknown, virus suspected; involvement of veins and arteries; veins more severely affected.

Ocular: Vascular sheathing; retinal hemorrhages; exudative detachment; retinal neovascularization; thick, inflammatory infiltrates around retinal veins; macular detachment; retinal tears associated with posterior vitreous detachment; peripheral uveitis; retinal periphlebitis; associated cytomegalovirus retinitis in HIV-seropositive patients; observed as an idiopathic finding in a child.

Clinical: Associated with tuberculosis; syphilis; sarcoidosis; multiple sclerosis; HIV infection.

Kleiner RC, et al. Acute frosted retinal periphlebitis. *Am J Ophthalmol* 1988; 106:27-34.

Mansour AM, Li HK. Frosted retinal periphlebitis in the acquired immunodeficiency syndrome. *Ophthalmologica* 1993; 207: 182-186.

Nakai A, Saika S. A case of frosted-branch retinal angiitis in a child. *Ann Ophthalmol* 1992; 24:415-417.

Secci AG, et al. Acute frosted retinal periphlebitis associated with cytomegalovirus retinitis. *Retina* 1992; 12: 245-247.

Watanabe Y, et al. Case of frosted branch angiitis. *Br J Ophthalmol* 1987; 71:553.

Acute Hemorrhagic Conjunctivitis (AHC; Epidemic Hemorrhagic Keratoconjunctivitis) 27

General: First reported in 1969, first epidemic in United States in 1981; enterovirus; explosive onset; usually bilateral; coxsackievirus A24 and enterovirus 70 have been implicated in the most recent outbreaks.

Ocular: Chemosis; follicular conjunctivitis; petechial bulbar hemorrhages; seromucous discharge; keratitis; lacrimation; lid edema; photophobia; preauricular lymphadenopathy.

Clinical: Systemic symptoms are rare, although several cases of lumbosacral radiculomyelitis have occurred late in the course of the disease; polio-like paralysis (associated with enterovirus 70).

Bern C, et al. Acute hemorrhagic conjunctivitis due to enterovirus 70 in American Samoa: serum-neutralizing antibodies and sex-specific protection. *Am J Epidemiol* 1992; 136: 1502-1506.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Minami K, et al. Seroepidemiologic studies of acute hemorrhagic conjunctivitis virus (enterovirus type 70) in West Africa. I. Studies with human sera from Ghana collected eight years after the first outbreak. *Am J Epidemiol* 1981; 114:267-273.

Whitcher JP, et al. Acute hemorrhagic conjunctivitis in Tunisia. Report of viral isolations. *Arch Ophthalmol* 1976; 94: 51-55.

Wright PW, et al. Acute hemorrhagic conjunctivitis. *Am Fam Physician* 1992; 45: 173-178.

Yin-Murphy M, et al. A recent epidemic of acute hemorrhagic conjunctivitis. *Am J Ophthalmol* 1993; 116:212-217.

Acute Retinal Necrosis Syndrome (ARN Syndrome; Bilateral Acute Retinal Necrosis; Barn Syndrome) 28

General: Evidence of association with herpes-type deoxyribonucleic acid (DNA) virus; occurs both unilaterally and bilaterally; includes varicella-zoster virus and herpes simplex virus type 1.

Ocular: Uveitis; vasculitis; vitreitis; retinal detachment; vitreous opacification; retinal periarteritis; exudates of peripheral retina; retinal necrosis; optic nerve enlargement; papillitis; arcuate neuroretinitis; arteritis and phlebitis (affecting the retinal vasculature); necrotizing retinitis; moderate to severe vitreitis; anterior segment inflammation; optic neuritis; late retinal detachment.

Clinical: None.

Culbertson WW, et al. Chickenpox-associated acute retinal necrosis syndrome. *Ophthalmology* 1991; 98:1641-1645.

Culbertson WW, et al. Varicella zoster virus is a cause of the acute retinal necrosis syndrome. *Ophthalmology* 1986; 93:559-567.

Duker JS, Blumenkranz MS. Diagnosis and management of the acute retinal necrosis syndrome (ARN) syndrome. *Surv Ophthalmol* 1991; 35:327-343.

Falcone PM, Brockhurst RJ. Delayed onset of bilateral acute retinal necrosis syndrome: a 34-year interval. *Ann Ophthalmol* 1993; 25:373-374.

Fisher JP, et al. The acute retinal necrosis syndrome. Part 1: *Clinical* manifestations. *Ophthalmology* 1982; 89: 1309-1316.

Margolis T, et al. Acute retinal necrosis syndrome with papillitis and arcuate neuroretinitis. *Ophthalmology* 1988; 95: 937-940.

Peyman GA, et al. Vitrectomy and intravitreal antiviral drug therapy in acute retinal necrosis syndrome. *Arch Ophthalmol* 1984; 102:1618-1621.

Thompson WS, et al. Acute retinal necrosis caused by reactivation of herpes simplex virus type 2. *Am J Ophthalmol* 1994; 118:205-211.

Addison Pernicious Anemia Syndrome (Pernicious Anemia Syndrome; Vitamin B12 Deficiency Anemia; Macrocytic Anemia; Biermer Syndrome) 29

General: Autosomal dominant; female preponderance; onset between ages 30 and 50 years; lack of intrinsic factor normally produced in the fundus of stomach and important for absorption of vitamin B_{12} in the intestinal tract; infrequent ocular involvement.

Ocular: Central scotoma, centrocecal scotomata, and field contractions in a few cases; retinal hemorrhages (round with white center) at the posterior pole; both retina and disk may have a whitish, hazy appearance; optic neuritis (ischemic); optic atrophy; palsies of extraocular muscles; ocular hypotony; cataract; bilateral, slowly progressive optic neuropathy, unclear etiology.

Clinical: Megaloblastic anemia (chronic and progressive); hypochlorhydria; glossitis; stomatitis; constipation or diarrhea; paresthesias and numbness; incoordination; ataxia; sphincter malfunction.

Biermer A. Form von Progressiver Pernizioser Anamie. *Korresp Bl Schweiz Arzt* 1872; 2:15.

Fisher JM, Taylor KB. A comparison of autoimmune phenomena in pernicious anemia and chronic atrophic gastritis. *N Engl J Med* 1965; 272:499.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 1, 4th ed. Baltimore: Williams & Wilkins, 1982.

Addison Syndrome (Addison Disease; Idiopathic Hypoparathyroidism; Adrenal Cortical Insufficiency; Moniliasis-Idiopathic Hypoparathyroidism) 30

General: Familial occurrence; association with moniliasis; onset during end of first and beginning of second decade of life; atrophy of adrenal cortex; prognosis for life is poor, with death in adrenal crisis.

Ocular: Ptosis; blepharitis; blepharospasm; keratoconjunctivitis with extreme photophobia; corneal ulcers; episcleritis; keratitic moniliasis; cataracts; papilledema.

Clinical: Moniliasis; tetany; progressive weakness; anorexia; progressive skin pigmentation; dry skin; brittle fingernails and toenails; sparse pubic and axillary hair or total alopecia; impotence.

Addison T. On the constitutional and local effects of disease of the suprarenal capsules. London S. Highley: Lend. Med. Gaz., 1949; 43:517.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:229.

Gass JD. The syndrome of keratoconjunctivitis, superficial moniliasis, idiopathic hypoparathyroidism and Addison's disease. Am J Ophthalmol 1962; 54:660.

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Williams GH, Dluhy RG. Diseases of the adrenal cortex. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:1953-1975.

Adie Syndrome (Holmes-Adie Syndrome; Markus Syndrome; Saenger Syndrome; Tonic Pupil; Iridoplegia Interna; Myotonic Pupil; Pseudotonic Pupillotonia; Weill-Reys Syndrome; Ross Syndrome) 31

General: Cause unknown; more frequent in females; manifested in second and third decades; abnormal sensitivity to 2.5% solution of methacholine; segmental compensatory hyperhidrosis; tonic pupil constricts, whereas normal pupils are unaffected; tonic pupil, hyporeflexia, and segmental hypohidrosis are manifestations of Ross syndrome.

Ocular: Slightly enlarged pupils; delayed or diminished direct and consensual reaction to light; usually unilateral; consensual reflex is abolished on the affected side but normal on the other; amblyopia.

Clinical: Loss of tendon reflexes, particularly ankle and knee jerk (partial or total).

Adie W. Complete and incomplete forms of the benign disorder characterized by tonic pupils and absent tendon reflexes. *Br J Ophthalmol* 1932; 16:449.

Agbeja AM, Dutton GN. Adie's syndrome as a cause of amblyopia. *J Pediatr Ophthalmol Strabismus* 1987; 24:176.

Fenichel GM. Tonic pupil syndrome (Adie syndrome). In: Fenichel GM, ed. *Clinical pediatric neurology,* 2nd ed. Philadelphia: WB Saunders, 1993:336.

Loewenfe1d IE, Thompson HS. The tonic pupil: a reevaluation. *Am J Ophthalmol* 1967; 63:46.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Reinauer S, et al. Ross syndrome: treatment of segmental compensatory hyperhidrosis by a modified iontophoretic device. *J Am Acad Dermatol* 1993; 28[2 Pt 2]:308-312.

Adrenoleukodystrophy (Melanodermic Leukodystrophy; Sudanophilic Leukodystrophy) 32

General: Degenerative metabolic disease in which cholesterol with long-chain fatty acids accumulates in affected cells; symptoms usually begin between ages 3 and 12 years but may have their onset in adulthood; X-linked recessive with predominantly central nervous system and adrenal dysfunction.

Ocular: Optic atrophy; retinal ganglion cell degeneration; exotropia; esotropia; cataracts; optic pallor; optic nerve hypoplasia; visual field defects; macular pigmentary changes; progressive visual loss.

Clinical: Central nervous system manifestations consisting of behavioral changes, disturbance of gait, dysarthria, and dysphagia; seizures; spastic quadriparesis; decorticate posturing; one third of patients show adrenal insufficiency.

Adams RD. Sudanophilic leukodystrophy with bronzing of skin and adrenal atrophy (adrenoleukodystrophy). In: Raymond D, Adams MV, eds. *Principles of Neurology,* 5th ed. New York: McGraw-Hill, 1993:840-841.

Cohen SMZ, et al. Ocular histopathologic studies of neonatal and childhood adrenoleukodystrophy. *Am J Ophthalmol* 1983; 95:82-96.

Fenichel GM. Adrenoleukodystrophy. In: Fenichel GM, ed. *Clinical pediatric neurology,* 2nd ed. Philadelphia: WB Saunders, 1993:143.

Folz SJ, Trobe JD. The peroxisome and the eye. *Surv Ophthalmol* 1991; 35:353-368.

Glasgow BJ, et al. Ocular pathologic findings in neonatal adrenoleukodystrophy. *Ophthalmology* 1987; 94: 1054-1060.

MacCollin M, De Vivo DC. Adrenoleukodystrophy. In: Rowland LP, ed. *Merritt's Textbook of Neurology,* 9th ed. Baltimore: Williams & Wilkins, 1995:579-580.

Sack GH Jr, Morrell JC. Visual pigment gene changes in adrenoleukodystrophy. *Invest Ophthalmol Vis Sci* 1993; 34: 2634-2637.

Traboulsi EI, Maumenee IH. Ophthalmologic manifestations of X-linked childhood adrenoleukodystrophy. *Ophthalmology* 1987; 94:47-52.

African Eye Worm Disease (Loiasis) 33

General: Caused by filarial nematode *Loa loa;* transmitted to humans by diurnally biting flies (deerflies) of the *Chrysops* species that live in the rain forests of West and Central Africa.

Ocular: Parasites of anterior chamber, conjunctiva, eyelid, vitreous, and choroid; conjunctivitis; keratitis; optic nerve atrophy; white, cottony mass of vitreous; central retinal artery occlusion; macular hemorrhages; paralysis of extraocular muscles; nystagmus; uveitis.

Clinical: Transient erythematous swelling; pruritus; eosinophilia; fever; urticaria; rarely neurologic involvement.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Malouf AR, et al. Loiasis in Maryland. *Arch Ophthalmol* 1992; 110:1010.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995.

Osuntokun O, Olurin O. Filarial worm (Loa loa) in the anterior chamber. Report of two cases. *Br J Ophthalmol* 1975; 59:166-167.

Sacks HN, et al. Loiasis. Report of a case and review of the literature. *Arch Intern Med* 1976; 136:914-915.

Agranulocytosis Syndrome (Pernicious Leukopenia Syndrome; Schultz Syndrome; Agranulocytic Angina Syndrome; Malignant Neutropenia Syndrome) 34

General: Caused by hypersensitivity reaction to chemicals, drugs, and ionizing radiation; may be idiopathic; more frequent in adults; female (3:1) preponderance; acute onset.

Ocular: Scleral and conjunctival icterus; conjunctival hemorrhages; retinal hemorrhages.

Clinical: Swollen, painful joints; malaise; sore throat with mucous membrane ulceration; sepsis.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McCluskey HB. Corticotropin (ACTH) in treatment of agranulocytosis following sulfisoxazole therapy. *J Am Med Assoc* 1953; 152:232.

Schultz W. Uber Eigenartige Halserkrankungen. *Dtsch Med Wochenschr* 1922; 48: 1495.

Aicardi Syndrome 35

General: All symptoms present at birth; cause unknown; all findings progress with age; shows X-linked dominant inheritance.

Ocular: Microphthalmia; lid twitching; absent pupillary reflexes; round retinal lacunae up to disk size look like holes with retinal vessels crossing over them; funnel-shaped disk; chorioretinitis.

Clinical: Infantile spasms (tonic seizures in flexion); epileptic seizures; cyanosis; mental anomaly; vertebral anomalies; telangiectasia; hypotonia; head deformities with biparietal bossing, occipital flattening, and plagiocephaly; defects of corpus callosum; cortical heterotopia; characteristic electroencephalogram; dilated intracranial ventricle with leukomalacia.

Aicardi J, et al. Le Syndrome Spasmes en Flexion, Agenesie Calleuse, Anomalies Chorio-Retiniennes. *Arch Fr Pediatr* 1969; 26:1103.

Carney SH, et al. Aicardi syndrome: more than meets the eye. *Surv Ophthalmol* 1993; 37:419-424.

Font RL. Aicardi syndrome. A clinicopathologic case report including electron microscopic observations. *Ophthalmology* 1991; 98:1727-1731.

Gedik Y, et al. Aicardi syndrome: a variant example with new *Clinical* findings. *Genet Couns* 1993; 4:281-283.

Geeraets WJ *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Alagille Syndrome (AGS; Alagille-Watson Syndrome, AWS; Cholestasis with Peripheral Pulmonary Stenosis; Arteriohepatic Dysplasia, AHD; Hepatic Ductular Hypoplasia, Syndromatic) 36

General: May be associated with 20p 11.2 deletion and four distinct coding mutations in *Jag 1* gene.

Ocular: Posterior embryotoxon and retinal pigmentary changes; anterior chamber anomalies, associated with eccentric or ectopic pupils.

Clinical: Neonatal jaundice; prominent forehead and chin; pulmonic valvular stenosis as well as peripheral arterial stenosis; abnormal vertebrae ("butterfly" vertebrae) and decrease in interpediculate distance in the lumbar spine; absent deep tendon reflexes and poor school performance; in the facies, broad forehead, pointed mandible, and bulbous tip of the nose; and in the fingers, varying degrees of foreshortening.

Alagille D, et al. Hepatic ductular hypoplasia associated with charactenstic facies, vertebral malformations, retarded physical, mental and sexual development, and cardiac murmur. *J Pediatr* 1975; 86:63-71.

LaBrecque DR, Mitros FA. Autosomal dominant transmission of arteriohepatic dysplasia to four generations of a single kindred. *Clin Res* 1982; 30:285A.

Li L, et al. Alagille syndrome is caused by mutations in human Jagged 1, which encodes a ligand for Notch 1. *Nat Genet* 1997; 16:243-251.

Raymond WR, Kearney JJ, Parmley VC. Ocular findings in arteriohepatic dysplasia (Alagille's syndrome). *Arch Ophthalmol* 1989; 107:1077.

Romanchuk KG, Judisch GF, LaBrecque DR. Ocular findings in arteriohepatic dysplasia (Alagille's syndrome). *Can J Ophthalmol* 1981; 16:94-99.

Schnittger S, et al. Alagille-Watson syndrome is assigned to 20 (p1.1-p1.2) and provisionally to the region p1 1.23- P12.1. *Cytogenet Cell Genet* 1989; 51:1074.

Yuan Z, et al. Mutational analysis of the Jagged 1 gene in Alagille syndrome families. *Hum Mol Genet* 1998; 7: 363-1369.

Albers-Schonberg Disease (Marble Bone Disease; Osteosclerosis Fragilis Generalisata; Osteopetrosis; Osteopoikilosis; Osteosclerosis Congenita Diffusa) 37

General: Simple recessive inheritance, also dominant transmission; benign form is asymptomatic in about 50% of cases and known under the synonym Henck-Assmann syndrome; prognosis is poor for malignant form, with death usually in infancy.

Ocular: Oculomotor paralysis; cranial nerve VII palsy; optic atrophy; ptosis; exophthalmos; papilledema; nystagmus; anisocoria; congenital cataracts; hypertelorism; visual loss in infancy; nasolacrimal duct obstruction; keratoconus.

Clinical: Cartilage and bone thickening; multiple fractures; hyperchromic anemia; osteomyelitis; *severe forms:* jaundice, hepatosplenomegaly, skeleton sclerosis, lymphadenopathy, and hydrocephalus in infants; *mild forms:* nerve compression, fractures, and milder form of anemia; pancytopenia from marrow obliteration; low serum calcium; elevated phosphorus.

Aasved H. Osteopetrosis from the ophthalmological point of view. A report of two cases. *Acta Ophthalmol* 1970; 48: 771-778.

Ainsworth JR, et al. Visual loss in infantile osteopetrosis. *J Pediatr Ophthalmol Strabismus* 1993; 30:201-203.

Albers-Schonberg H. Rontgenbilder einer Seltenen Knochenerkrankung. *Munch Med Wochenschr* 1904; 51:365.

Ellis PP, Jackson WE. Osteopetrosis: a *Clinical* study of optic nerve involvement. *Am J Ophthalmol* 1962; 53: 943-953.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Orengo SD, Patrinely JR. Dacryocystorhinostomy in osteopetrosis. *Ophthalmic Surg* 1991; 22:396-398.

Albinism (Brown Oculocutaneous Albinism; Nettleship Falls Syndrome) 38

General: Congenital hypopigmentation.

1. Complete.

Ocular: Iris thin, pale blue; prominent choroidal vessels with poorly defined fovea; nystagmus; head nodding; frequently myopic astigmatism and strabismus; marked photophobia; eyelashes and eyebrows are white; optic atrophy; cataract; abnormal decussation of retinogeniculate axons at the chiasm.

Clinical: White hair, eyebrows, and skin; autosomal recessive.

2. Modified complete.

Ocular: Slight pigmentation at pupillary border; may be nystagmus, photophobia, and myopia; choroidal vessels prominent.

Clinical: Negroes; slight pigmentation; golden hair; tendency to hyperkeratoses; freckling in exposed areas of skin; autosomal recessive.

3. Ocular.

Ocular: Marked deficiency of pigment in iris and choroid; nystagmus and myopic astigmatism; iris of female carrier frequently is translucent; macular hypoplasia; photophobia; pigmentation of retinal pigment epithelium.

Clinical: Normal pigmentation elsewhere; autosomal recessive.

4. Amish.

Ocular: At birth, complete albinism with blue translucent irides and albinotic fundal reflex; nystagmus; photophobia; increasing pigmentation with age; abnormal decussation of retinogeniculate axons at the chiasm.

Clinical: White hair and skin at birth; increasing pigmentation with yellow hair and normal skin that tans; autosomal recessive.

Brodsky MC, et al. Magnetic resonance imaging of the visual pathways in human albinos. *J Pediatr Ophthalmol Strabismus 1993; 30:382-385.*

Fitzgerald K, Cibis GW: The value of flash visual evoked potentials in albinism. *J Pediatr Ophthalmol Strabismus* 1994; 31:18-25.

Flander M, Young D. Atypical sensory nystagmus and its surgical management. *Can J Ophthalmol* 1983; 18:349-351.

Francois J. Albinism. *Ophthalmologica* 1979; 178: 19-31.

Fulton AB, et al. Human albinism. Light and electron microscopy study. *Arch Ophthalmol* 1978; 96:305-310.

Harley RD. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Hoeft WW, Hughes MK. A comparative study of low-vision patients: their ocular disease and preference for one specific series of light transmission filters. *Am J Optom Physiol Opt* 1981; 58:841-845.

King RA, et al. Brown oculocutaneous albinism. *Ophthalmology* 1985; 92:1496-1504.

Taylor WOG. Visual disabilities of oculocutaneous albinism and their alleviation. *Trans Ophthalmol Soc U K* 1978; 98:423-445.

Witkop CJ. Albinism: hematologic-storage disease, susceptibility to skin cancer, and optic neuronal defects shared in all types of oculocutaneous and ocular albinism. *Ala J Med* Sci 1979; 16:327-330.

Albright Syndrome (Fuller Albright Syndrome; Jaffe-Lichtenstein Syndrome; McCune-Albright Syndrome; Osteitis Fibrosa Disseminata; Osteodystrophia Fibrosa; Polyostotic Fibrous Dysplasia; Fibrous Dysplasia) 39

General: Etiology unknown; disease rare; manifested in children and young adults; found predominantly in females.

Ocular: Unilateral proptosis; papilledema; optic atrophy; lacrimal fossa mass; acute or chronic monocular visual loss.

Clinical: Medullary structures replaced by fibrous dysplasia; pelvic bones and lower extremities most frequently involved (spontaneous fractures); brown pigmented areas of skin, from small, freckle-like dots to large, flat patches on thighs, sacrum, upper spine, neck, and scalp; endocrine dysfunction (precocious puberty in females) with early menarche, adolescent external genitalia, and breast enlargement; loss of hearing; convulsions; mental retardation.

Albright F, et al. Syndrome characterized by osteitis fibrosa disseminata, areas of pigmentation and endocrine dysfunction, with precocious puberty in females. *N Engl J Med* 1937; 16:727.

Arroyo JG, et al. Steroid induced visual recovery in fibrous dysplasia. *J Clin Neuro-Ophthalmol* 1991; 11:259-261.

Bland LI, et al. Acute monocular blindness secondary to fibrous dysplasia of the skull: a case report. *Ann Ophthalmol* 1992; 24:263-266.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:260.

Lods F. Ophthalmologic localization of an unusual disease: Jaffe-Lichtenstein syndrome or an approximate form of Albright's syndrome. *Bull Soc Ophthalmol Fr* 1970; 70:1220.

McCluskey P, et al. Monostotic fibrous dysplasia of the orbit: an unusual lacrimal fossa mass. *Br J Ophthalmol* 1993; 77:54-56.

Alcoholism 40

General: Classified into three groups; symptoms of mental disease, physiologic poison, or result of social drinking; addiction compounds other health disorders.

Ocular: Congestion of conjunctiva; amblyopia; diplopia; night blindness; nystagmus; cataracts; paralysis of accommodation; paralysis of extraocular muscles; esophoria for distance fixation; acute visual loss; cotton-wool spots; cherry-red spot (associated with pancreatitis).

Clinical: Tremors; seizures; delirium; alcoholic hepatitis; cirrhosis; gastritis; pancreatitis; cancer of mouth and esophagus; peripheral neuropathy; organic brain disease; hypertension; cardiomyopathy; hypoglycemia; anemia; hyperuricemia; susceptibility to infections; skeletal myopathies.

Duane TD, et al. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987:2.

Steel JR, et al. Blind drunk: alcoholic pancreatitis and loss of vision. *Postgraduate Med J* 1993; 69:151-152.

Alexander Disease 41

General: Rare degenerative neurologic disorder characterized by diffuse demyelination in the presence of Rosenthal fibers; cases may resemble multiple sclerosis; neuroradiologic findings include increased cerebellar white matter hyperintensity and diffuse periventricular signal hyperintensities.

Ocular: Impaired smooth pursuit; gaze-evoked horizontal nystagmus; slowed saccades; ocular myoclonus.

Clinical: In infants, hydrocephalus, spasticity, and seizures; in juveniles and adults, bulbar palsy and hyperreflexia, intermittent neurologic dysfunction.

Reichard EAP, Ball WS, Bove KE. Alexander disease: a case report and review of the literature. *Pediatr Pathol Lab Med* 1996; 16:327-343.

Russo LS, Aron A, Anderson PJ. Alexander's disease: a report and reappraisal. *Neurology* 1976; 26:607-614.

Alkaptonuria (Ochronosis; Garrod Syndrome) 42

General: Rare autosomal recessive metabolic disease; enzyme homogentisic acid oxidase missing; both sexes affected; onset in first few days of life; manifestations more severe in males.

Ocular: Pigmentation of cornea, sclera, and conjunctiva; ochronosis of sclera; oil globulation within Bowman membrane.

Clinical: Black-colored urine on standing; osteoarthritis; valvular heart disease; atherosclerosis (homogentisic acid oxidase deficiency); pigmentation of cartilage and other connective tissues.

Carlson DM, et al. Ocular ochronosis from alkaptonuria. *J Optom Assoc* 1991; 62:854-856.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:246.

Garrod AF. About alkaptonuria. Lancet 1901; 2:1484-1486.

Kampik A, et al. Ocular ochronosis. Clinicopathological, histochemical, and ultrastructural studies. *Arch Ophthalmol* 1980; 98:1441-1447.

Allergic to Everything Syndrome 43

General: "Environmentally ill" or allergic to unusual or common substances in the environment; females affected most frequently.

Ocular: Transient visual loss.

Clinical: Stupor; lethargy; memory loss; depression; gastrointestinal dysfunction; rashes; migraine headache; hearing loss.

Brodsky CM. Allergic to everything: a medical subculture. *Psychosomatics* 1983; 24:7731-7741.

Grieco MH. Controversial practices in allergy. *JAMA* 1982; 247:3106-3111.

Alopecia Areata 44

General: Unknown etiology; increased incidence of autoimmune disease, Addison disease, diabetes mellitus, and vitiligo; initially occurs episodically; most commonly in patients between the ages of 5 and 40 years; most cases repopulate with normal hair in 6 to 12 months without any specific treatment.

Ocular: Loss of eyelashes and eyebrows and cataract.

Clinical: Circumscribed patches of hair loss on scalp and all body hair areas; fingernail changes.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Rubin P. Disorders of the eyelashes and eyebrows. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:1853.

Alport Syndrome (Hereditary Familial Congenital Hemorrhagic Nephritis; Hereditary Nephritis; Familial Nephritis) 45

General: Autosomal dominant inheritance; early death in males; normal life span in females.

Ocular: Anterior lenticonus (bilateral progressive); subcapsular cataracts; thinning of lens capsule; fundus albipunctatus; retinopathy similar to juvenile macular degeneration; hyaline bodies of optic nerve head; vesicles in Descemet membrane affecting basement membrane collagen; anterior and polar cataracts.

Clinical: Hemorrhagic nephritis; progressive nerve deafness; deafness (high tone, sensorineural); most often transmitted as an X-linked dominant trait, although dominant and recessive transmission has been reported.

Alport AC. Hereditary familial congenital hemorrhagic nephritis. *Br Med J* 1927; 1:504.

Jacobs M, et al. Ophthalmologic assessment of young patients with Alport syndrome. *Ophthalmology* 1992; 99: 1039-1044.

Polak BCP, Hogewind BL. Macular lesions in Alport's diseases. *Am J Ophthalmol* 1977; 84:532.

Streeteen BW Pathology of the lens. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV*. Philadelphia: WB Saunders, 1994:2189.

Streeteen BW, et al. Lens capsule abnormalities in Alport's syndrome. *Arch Ophthalmol* 1987; 105:1693-1697.

Sukhija J, Jagjit MS, Saini MD, et al. Phacoemulsification and intraocular lens implantation in an Alport's syndrome patient with bilateral anterior and posterior lenticonus. *J Cataract Refract Surg* 2003; 29: 1834-1836.

Teekhasaenee C, et al. Posterior polymorphous dystrophy and Alport syndrome. *Ophthalmology* 1991; 98: 1207-1215.

Allgrove Syndrome 46

General: Autosomal dominant

Ocular: Distichiasis, conjuncitivits, keratitis, congenital alacrima

Clinical: ACTH insensitivitiy, achialasia

Moore PS, Couch RM, Perry YS, et al. Allgrove syndrome: An autosomal recessive syndrome of ACTH insensitivity, achialasia and alacrima. *Clin Endocrinol*. 1991; 34: 107-114.

Davidoff E, Friedman AH: Congenital alacrima. *Surv Ophthalmol* 1997; 22: 113-118.

Alstrom Disease (Cataract and Retinitis Pigmentosa) 47

General: Retinal lesion associated with deafness; severe visual loss in the first decade. **Ocular:** Cataract; retinitis pigmentosa; optic atrophy; salt and pepper pigment epithelial abnormalities. Electroretinogram pathognomonic findings include initially normal rod component, which can become undetectable as early as 5 years of age; undetectable cone activity at 18 months.

Clinical: Nerve deafness; diabetes mellitus in childhood; obesity; renal disease; baldness; hyperuricemia; hypogenitalism; acanthosis nigricans; skeletal anomalies; diabetes mellitus; deafness.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Harley RD, ed. *Pediatric Ophthalmology,* 4th ed. Philadelphia: WB Saunders, 1998.

Konigsmark BW, et al. Dominant congenital deafness and progressive optic nerve Atrophy. *Arch Ophthalmol* 1974; 91:99-103.

Millay RH, et al. Ophthalmologic and systemic manifestations of Alstrom's disease. *Am J Ophthalmol* 1986; 102: 482-490.

Tremblay F, et al. Longitudinal study of the early electroretinographic changes in Alstrom's disease. *Am J Ophthalmol* 1993; 115:657-665.

Alzheimer Disease (Dementia) 48

General: Diffuse brain atrophy coming on well before the senile period of life; progressive; etiology currently unknown but hereditary disorder suspected; terminally, nearly decorticate, with loss of all ability to think, perceive, speak, or move.

Ocular: Fixed dilated pupil; optic atrophy; decreased contrast sensitivity, color vision, and stereo vision; abnormalities of the optic nerve head and nerve fiber layer; controversy exists regarding the ability to diagnose patients with Alzheimer disease by their marked hypersensitivity in pupil dilation response to tropicamide.

Clinical: Emotional disturbances; depression; anxiety; antisocial behavior; aphasia; apraxic disturbances; abnormalities of space perception; shuffling gait; generalized shuffling gait with short steps; disturbances in thought process.

Bassi CJ, et al. Vision in aging and dementia. *Optom Vis Sci* 1993; 70:809-813.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Roy FH. *Ocular Differential Diagnosis*. 7th ed. Philadelphia: Lea & Febiger, 2002.

Tsai CS, et al. Optic nerve head and nerve fiber layer in Alzheimer's disease. *Arch Ophthalmol* 1991; 109:199-204.

Wolin MJ. Alzheimer's disease and the eye. *J South Carolina Med Assoc* 1994; 90:534-538.

Amaurosis Fugax Syndrome 49

General: Caused by malignant hypertension; often occurs in association with heavy smoking; may indicate vascular insufficiency of the vertebrobasilar arterial system; may precede a cerebrovascular accident and not infrequently seen in vascular insufficiency problems of the carotid arterial system; the cause, if found, is commonly an abnormality in the ipsilateral carotid artery or a cardiac source of embolism.

Ocular: Partial blindness in short attacks to permanent complete blindness; scintillating scotoma; teichopsia; retinal arteriolar spasm; signs of arteriolar sclerosis.

Clinical: Malignant hypertension; atherosclerosis; expanding lesions of the frontal or temporal lobe; vascular insufficiency.

Guerry D III, Wiesinger H. Ocular complications in carotid angiography. *Am J Ophthalmol* 1953; 55:241.

Haney WP, Preston RE. Ocular complications of carotid arteriography in carotid occlusive disease. *Arch Ophthalmol* 1983; 67:127.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Muller M, et al. Carotid artery disease in vascular ocular syndromes. *J Clin Neuro-Ophthalmol* 1993; 13:175-180.

Smit RL, et al. The source of embolism in amaurosis fugax and retinal artery occlusion. *Int Ophthalmol* 1994; 18: 83-86.

Amblyopic Schoolgirl Syndrome 50

General: Etiology unknown.

Ocular: Amblyopia; changes in visual fields; abnormal dark adaptation curve; visual field defects usually are tubular or spiral but also include central, paracentral, and ring scotomas; hemianopsias and superior and inferior field defects reported.

Clinical: Psychogenic disorder: affective and hysterical.

Mantyjarvi MI. The amblyopic schoolgirl syndrome. *J Pediatr Ophthalmol Strabismus* 1981; 18:30-33.

Schlaegel TF Jr, Quilala FV. Hysterical amblyopia. *Arch Ophthalmol* 1955; 54:875-884.

Amebiasis (Amebic Dysentery, Entamoeba Histolytica) 51

General: Caused by *Entamoeba histolytica; E. histolytica* cysts in stools are diagnostic.

Ocular: Conjunctivitis; iridocyclitis; hypopyon; central choroiditis; retinal hemorrhages; retinal perivasculitis; macular edema; corneal ulceration; granulomatous and nongranulomatous uveitis; vitreous hemorrhage.

Clinical: Chronic dysentery; abscesses of liver and brain; toxic megacolon.

Ashton N, Stamm W. Amoebic infection of the eye. A pathological report. *Trans Ophthalmol Soc U K* 1975; 95: 214-220.

Lund OE, et al. Amoebic keratitis: a clinicopathological case report. *Br J Ophthalmol* 1978; 62:373-375.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995.

Naginton J, et al. Amoebic infection of the eye. *Lancet* 1974; 2:1537-1540.

Watson PG. Amoebic infection of the eye. *Trans Ophthalmol Soc U K* 1975; 95:204-206.

Amendola Syndrome 52

General: Observed in Sao Paulo, Brazil; all ethnic groups are affected; endemic form of pemphigus foliaceus; possibly caused by environmental agents; autoimmune disease mediated by autoantibodies of the immunoglobulin G (IgG) class, IgG4 subclass.

Ocular: Blisters around eyebrows; entropion; ectropion; trichiasis; iritis.

Clinical: Brazilian pemphigus (fogo selvagem, "wild fire"), which resembles, because of its appearance, pemphigus foliaceus; fever; chills.

Amendola F. Cataracta No Pemfigo Foliaceo (nota previa). *Rev Paul Med* 1945; 26:286.

Korting GW. *The skin and the eye: a dermatologic correlation of diseases of the periorbital region*. Philadelphia: WB Saunders, 1973:82.

Magalini SI, Scrascia, E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Sampaio SA, et al. Brazilian pemphigus foliaceus, endemic pemphigus foliaceus or fogo selvagem (wild fire). *Dermatol Clin* 1994; 12:765-776.

American Mucocutaneous Leishmaniasis (Cutaneous Leishmaniasis; Oriental Sore) 53

General: Causative agent is protozoal parasite *Leishmania brasiliensis.*

Ocular: Keratitis; eyelid edema; conjunctival ulcer; vascular sclerosis of choroid; granulomata of eyelid and conjunctiva; blepharoconjunctivitis.

Clinical: Ulcerating granulomas of the skin, nasal septum, nasopharynx, lips, soft palate, larynx, and genitals.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Nandy A, et al. Leishmanial blepharoconjunctivitis. *Trop Geographical Med* 1991; 43:303-306.

Wilcocks C, Manson-Bahr PEC, eds. *Manson's Tropical Diseases,* 17th ed. Baltimore: Williams & Wilkins, 1972: 140-147.

Aminopterin-Induced Syndrome 54

General: Teratogenic effect of aminopterin and derivatives on fetus; present at birth; usually fetal or postnatal death.

Ocular: Hypertelorism.

Clinical: Small body; microcephaly; hypoplasia of cranial bones; broad nasal bridge; micrognathia; cleft palate; low-set ears; mesomelic; hypodactyly; talipes equinovarus.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Shaw EB, Steinbach HL. Aminopterin-induced fetal malformation. *Am J Dis Child* 1968; 115:477-482.

Amniogenic Band Syndrome (Ring Constriction; Streeter Dysplasia) 55

General: Caused by fetus swallowing one or more of the free-floating strands that result from amniotic rupture; the tension of these strands intraorally and extraorally produces secondary tears and deformations; no hereditary factor known.

Ocular: Upward slant of palpebral fissures; bilateral upper and lower lid colobomas; telecanthus; bilateral corneal opacities; microphthalmos; strabismus; hypertelorism; epibulbar choristoma; unilateral chorioretinal defects or lacuna (rare).

Clinical: Craniofacial and limb abnormalities.

Braude LL, et al. Ocular abnormalities in the anmiogenic band syndrome. *Br J Ophthalmol* 1981; 65:299-303.

Hashemi K, et al. Chorioretinal lacuna in the anmiotic band syndrome. *J Pediatr Ophthalmol Strabismus* 1991; 28: 238-239.

Miller MT, et al. Amniotic bands as a cause of ocular anomalies. *Am J Ophthalmol* 1987; 104:270-279.

Murata T, et al. A case of anmiotic band syndrome with bilateral epibulbar choristoma. *Br J Ophthalmol* 1992; 76: 685-687.

Streeter GL. Focal deficiencies in fetal tissues and their relation to intrauterine amputation. *Contrib Embryol Carney Inst* 1930; 22:1-44.

Amyloidosis of Gingiva and Conjunctiva, with Mental Retardation (Primary Systemic Amyloidosis) 56

General: Autosomal recessive; primary amyloidosis differs from secondary by the mesodermal tissues being affected and nodular form of deposits; no preexisting medical condition; preferential involvement of mesenchymal tissues; variable staining of deposits.

Ocular: Conjunctivitis with deposits; corneal leukoma; waxy eyelid papules with purpura; proptosis; diplopia; decreased vision; ptosis; keratitis sicca; upper lid mass; tonic pupil; accommodative paresis; diffuse yellow conjunctival mass.

Clinical: Hyperplastic gingivitis, tongue, skin, and muscles; lungs with icing-like coating; mental retardation; peripheral neuropathy; congestive heart failure; polyarthropathy; spontaneous, incidental purpura; macroglossia; bleeding diathesis; idiopathic carpal tunnel syndrome.

Hornova J, Dlurosova O. Primary amyloidosis of gingiva and conjunctiva and mental disorder in a brother and sister. *Oral Surg* 1968; 25:457-464.

Lamkin JC. Amyloidosis and the eye. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:2956-2975.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Andersen-Warburg Syndrome (Whitnall-Norman Syndrome; Oligophrenia Microphthalmos Syndrome; Norrie Disease; Atrophia Oculi Congenital Fetal Iritis Syndrome; Congenital Progressive Oculo-Acoustico-Cerebral Dysplasia) 57

General: Sex-linked inheritance; gross deformation of both eyes; only males affected; onset at birth; putative gene for Norrie disease has been isolated and mapped to Xp11.3.

Ocular: Bilateral microphthalmos with extensive destruction of all ocular structures often resembling a pseudotumor; blindness at birth; iris atrophy; iritis; corneal opacification and lenticular destruction with a mass visible behind the lens as long as the lens is still clear; malformed retina and choroid with retinal pseudotumors; retinal detachment; retrolental vascular mass.

Clinical: Mental retardation ranging from imbecility to idiocy (may begin at any age) in about two thirds of cases; deafness of differing severity with onset between ages 9 and 45 years.

Andersen SR, Warburg M. Norrie's disease: congenital bilateral pseudotumor of retina with recessive X-chromosomal inheritance. *Arch Ophthalmol* 1961; 66:614-618.

Black G, Redomond RM. The molecular biology of Norrie's disease. *Eye* 1994; 8[Pt 5]:491-496,

Enyedi LB, et al. Ultrastructural study of Norrie's disease. *Am J Ophthalmol* l991*;* 111:439-445,

Liberfarb RM, et al. Norrie's disease: a study of two families. *Ophthalmology* 1985; 92:1445-1451.

Norrie G. Causes of blindness in children; twenty-five years' experience of Danish institutes for the blind. *Acta Ophthalmol* 1927; 5:357-386,

Warburg M. Norrie's disease: differential diagnosis and treatment. *Acta Ophthalmol* 1975; 53:217-236.

Wong F, et al. Identification of a nonsense mutation at codon 128 of the Norrie's disease gene in a male infant. *Arch Ophthalmol* 1993; 111:1553-1557,

Andogsky Syndrome (Atopic Cataract Syndrome; Dermatogenous Cataract) 58

General: Inherited abnormality involving the skin and lens with an altered reactivity to antigen.

Ocular: Atopic keratoconjunctivitis; keratoconus; uveitis; dense subcapsular cataract developing to a complete dense opacification.

Clinical: Atopic dermatitis as erythematous thickening of the skin with papular hyperpigmented and scaly changes, most frequently found in regions of the wrist, popliteal fossa, neck, and sometimes forehead.

Andogsky N. Cataracts Dermatogenes. Ein Beitrag zur Aetiologieder Linsentrubung. *Klin Monatsbl Augennheilk* 1914; 52:824.

Coles RS, Laval J. Retinal detachments occurring in cataract associated with neurodermatitis. *Arch Ophthalmol* 1952; 48:30.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Angiolymphoid Hyperplasia with Eosinophilia (Kimura Disease) 59

General: Benign, slow-growing tumor involving primarily the face and scalp; etiology unknown but inflammatory reaction suspected; occurring primarily in young Oriental males.

Ocular: Proptosis; orbital tumor; upper lid swelling (may be bilateral).

Clinical: Single or multiple subcutaneous or dermal nodules; regional lymphadenopathy; peripheral blood eosinophilia; nephrotic syndrome.

Archer KF, et al. Orbital angiolymphoid hyperplasia with eosinophilia. Presentation as chalazion. *Ophthal Plastic Reconstr Surg* 1991; 7:208-221.

Kennedy SM, et al. Bilateral Kimura's disease of the eyelids. *Br J Ophthalmol* 1992; 76:755-757.

Kimura T, et al. On the unusual granulation combined with hyperplastic changes of lymphatic tissues. *Trans Soc Pathol Jpn* 1948; 37:179-180.

Smith DL, et al. Angiolymphoid hyperplasia with eosinophilia (Kimura's disease) of the orbit. *Arch Ophthalmol* 1988; 106:793-795.

Anemia 60

General: Ocular complications generally only seen in severe anemia.

Ocular: Palpebral conjunctival pallor; retinal hemorrhages; cotton-wool spots; retinal vein dilation; papilledema; ischemic optic neuropathy.

Clinical: Blood loss; excessive red blood cell destruction; inadequate red blood cell production; thrombocytopenia; leukemia.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. Clinical *Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Angelucci Syndrome (Critical Allergic conjunctivitis Syndrome) 61

General: Etiology unknown; pruriginous cutaneous and mucous reactions that appear and cease rather suddenly.

Ocular: Chemosis; conjunctivitis (papillary type); severe itching and burning; photophobia.

Clinical: Tachycardia; vasomotor lability; excitability; allergies (asthma, urticaria, edema); dystrophic conditions and endocrine disorders are frequently associated findings.

Angelucci A. Di una Sindrome Sconoscita Negli Infermi di Cattarro Primaverile, *Arch Ottal Palermo* 1897-1898:4: 270.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Angioedema (Angioneurotic Edema; Giant Edema; Giant Urticaria; Hives; Nettle Rash; Quincke Disease) 62

General: Vascular reaction involving subcutaneous tissues or submucosa; both sexes affected; allergy to various agents, including medications; emotional factor may be involved; recurrent.

Ocular: Optic neuritis; papilledema; central serous retinopathy; corneal edema; exophthalmos; nystagmus; secondary glaucoma; uveitis; periorbital and lid edema.

Clinical: Transient erythema; angioneurotic edema of loose subcutaneous tissue; sporadic urticaria; nausea; vomiting; diarrhea; cephalalgia; severe respiratory distress; polyuria.

Katz Y, et al. Localized periorbital edema induced by aspirin. *Allergy* 1993; 48:366-369.

Magalini SI, Scrascia E. *Dictionary o/Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Margo CE, et al. Ophthalmic manifestations of chronic angioedema with necrotizing vasculitis. *Am J Ophthalmol* 1992; 113:691-696.

Quincke HI. Uber Akutes Unbeschriebenes Hautderm. *Mhefte Prakt Dermatol* 1882; 1:129-131.

Angular Conjunctivitis (Morax-Axenfeld Bacillus) 63

General: Caused by *Moraxella lacunata*, which frequently inhabits the nose.

Ocular: Conjunctivitis; hypopyon; keratitis; uveitis; corneal marginal ulcer.

Jones DB. Early diagnosis and therapy of bacterial corneal ulcers. *Int Ophthalmol Clin* 1973; 13:1-29.

Marioneaux SJ, et al. Moraxella keratitis. *Cornea* 1991; 10:21-24.

Van Bijsterveld OP. Bacterial proteases in Moraxella angular conjunctivitis. *Am J Ophthalmol* 1971; 74:181-184.

Aniridia 64

General: Hereditary, recessive (two thirds of cases), can be dominant, sporadic, or traumatic; absence of the iris; rare; usually bilateral unless due to trauma.

Ocular: Absence of iris; subluxed lens; iridodialysis; cataract; glaucoma; corneal scarring, vascularization, and edema; iris colobomata; round eccentric pupils; keratoconus.

Clinical: Cerebellar ataxia; mental retardation; Wilms tumor.

Francois J, et al. Aniridia Wilms' tumor syndrome. *Ophthalmologica* 1977; 174:35-39.

Johns KJ, O'Day DM. Posterior chamber intraocular lenses after extracapsular extraction in patients with aniridia. *Ophthalmology* 1991; 98:1698-1702.

Kremer I, et al. Results of penetrating keratoplasty in aniridia. *Am J Ophthalmol* 1993; 115:317-320.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Mintz-Hittner HA, et al. Criteria to detect minimal expressivity within families with autosomal dominant aniridia. *Am J Ophthalmol* 1992; 114:700-707.

Nelson LB, Spaeth GL, Nowinski TS, et al. Aniridia. A review. *Surv Ophthalmol* 1984; 28:621-642.

Aniridia and Absent Patella 65

General: Autosomal dominant; rare.

Ocular: Absence of iris; cataracts; glaucoma.

Clinical: Absence of knee cap, hypoplastic or aplastic.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Murkinson AE, Murkinson NK. A familial syndrome of aniridia and absence of patella. *Birth Dejects* 1975; 11: 129-131.

Aniridia, Cerebellar Ataxia, and Mental Deficiency (Gillespie Syndrome) 66

General: Autosomal recessive; onset at birth.

Ocular: Congenital cataracts; incomplete formation of iris; bilateral congenital mydriasis.

Clinical: Cerebellar ataxia; mental deficiency; delayed developmental milestones; persistent hypotonia of muscles; gross incoordination; attention tremor; scanning speech.

Gillespie FD. Aniridia, cerebellar ataxia and oligophrenia in siblings. *Arch Ophthalmol* 1965; 73:338-341.

Glaser T, et al. Absence of PAX6 gene mutations in Gillespie syndrome (partial aniridia, cerebellar ataxia and mental retardation). *Genomics* 1994; 19:145-148.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Quarrell O. Gillespie syndrome reported as bilateral congenital mydriasis. *Br J Ophthalmol* 1993; 77:827-828.

Sarsfield JK. The syndrome of congenital cerebellar ataxia, aniridia, and mental retardation. *Dev Med Child Neurol* 1971; 13:508-511.

Aniridia, Partial with Unilateral Renal Agenesis and Psychomotor Retardation 67

General: Autosomal recessive.

Ocular: Congenital glaucoma; telecanthus; absence of iris; hypertelorism.

Clinical: One kidney absent or in failure; motor effects of cerebral or psychic activity retarded or slowed.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Sommer A, et al. A syndrome of partial aniridia, unilateral renal agenesis, and mild psychomotor retardation in siblings. *J Pediatr* 1974; 85:870-872.

Ankyloblepharon Filiforme Adnatum and Cleft Palate (Hay-Wells Syndrome; AEC Syndrome) 68

General: Autosomal dominant; cleft palate and/or cleft lip; congenital filiform fusion of eyelids.

Ocular: Filiform fusion of eyelids; pterygium; keratoconus.

Clinical: Cleft lip and palate; paramedian mucous pits of lower lip; ectodermal dysplasia; infrequent association with trisomy 18; partial thickness fusion of central portion of lid margins.

Akkermans CH, et al. Ankyloblepharon filiforme adnatum. *Br J Ophthalmol* 1979; 63:129-131.

Bacal DA, et al. Ankyloblepharon filiforme adnatum in trisomy 18. *J Pediatr Ophthalmol Strabismus* 1993; 30: 337-339.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Vanderhofft SL, et al. Severe skin erosions and scalp infections in AEC syndrome. *Pediatr Dermatol 1*993; 10: 334-340.

Weiss AH, et al. Ankyloblepharon filiforme ad natum. *Am J Med Genet* 1992; 42:369-373.

Ankylostomiasis (Hookworm Disease) 69

General: Causative agents include *Necator americanus*, *Ancylostoma duodenale*, *Ancylostoma braziliense*, and *Ancylostoma caninum;* final diagnosis depends upon finding eggs in feces.

Ocular: Retinal hemorrhages around optic disk; diplopia; conjunctival xerosis; visual field defects; cataract.

Clinical: Maculopapules; localized erythema; microcytic hypochromic anemia.

Brown HW *Basic Clinical parasitology.* Norwalk, CT: Appleton-Century-Crofts, 1969:121-122.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Annette Von Droste-Hulshoff Syndrome 70

General: Premature birth; pseudo strabismus due to macular ectopia in retinopathy of prematurity.

Ocular: Myopia; retinal detachment; negative and positive angle kappa; esotropia; temporal macular ectopia; chorioretinal colobomata; falciform folds; persistent hyaloids artery; abnormal position of blind spot; epicanthus; telecanthus; blepharophimosis; hypertelorism; asymmetrical orbits; exophthalmos; enophthalmos.

Clinical: Face turn; angioma; neoplasia.

Alfieri MC, et al. The Annette Von Droste-Hulshoff syndrome, pseudostrabismus due to macular ectopia in retinopathy of prematurity. *Ophthalmic Paediatr Genet* 1988; 9:13-16.

Myer-Schweckerath G. Dos Annette Von Droste-Hulshoff syndrome. *Klin Monatsbl Augenheilk* 1984; 184:574.

Anorexia Nervosa (Apepsia Hystericci) 71

General: Compulsive neurosis; refusal to eat; occurs in adolescent to young adult females; symptomatic recovery or chronic course.

Ocular: Cataract; central retinal vein occlusion; myopathy of orbicularis oculi (weakness of eye closure).

Clinical: Severe cachexia; loss of hair; nausea; constipation; diarrhea; depression; vigorous activity; weight loss; menstrual disturbance.

Browning CH, Miller SI. Anorexia nervosa: a study in prognosis and management. *Am J Psychiatry* 1968; 124: 1128-1132.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Shibuya Y, Hayasaka S. Central retinal vein occlusion in a patient with anorexia nervosa. *Am J Ophthalmol* 1995; 119:109-110.

Woodruff PW, et al. Neuromyopathic complications in a patient with anorexia nervosa and vitamin C deficiency. *Int J Eating Disord* 1994; 16:205-209.

Anoxic Overwear Syndrome 72

General: Caused by a reduction in oxygen supply due to continuously worn hydrogel lenses; allergic or toxic reactions to preservatives used in the cleaning process.

Ocular: Refractive error changes; endothelial cell changes; physical trauma to the anterior surface of the cornea; corneal neovascularization; giant papillary conjunctivitis; contact lens deposits; acute red eye syndrome.

Binder PS. The physiologic effects of extended wear soft contact lenses. *Ophthalmology* 1980; 87:745-749.

Sarver MD, et al. Corneal edema with hydrogel lenses and eye closure: effect of oxygen transmissibility. *Am J Optom Physiol Opt* 1981; 58:386.

Anterior Capsule Contraction Syndrome 73

General: Associated with diabetes mellitus, myotonic muscular dystrophy, uveitis, shrinkage of anterior capsular opening after anterior continuous curvilinear capsularhexis

Ocular: Uveitis, retinitis pigmentosa, high myopia, pseudoexfoliation syndrome

Reyntizens B, Marie-Jose, T, Von March E: Capsular peeling in anterior capsule contraction syndrome. *J Cataract Refract Surg* 2004; 30: 906-910.

Rosen E, Gore C: Posterior chamber phakic intraocular lens to correct myopia and hyperopia. *J Cataract Refract Surg* 1998; 24:596-606.

Anterior Chamber Cleavage Syndrome (Reese-Ellsworth Syndrome; Peters-Plus Syndrome) 74

General: Abnormalities in the embryologic development of the anterior chamber due to failure of normal migration of mesodermal cells across the anterior segment of the eye or failure of later differentiation of the mesodermal elements; various conditions described as congenital: central anterior synechiae, persistent mesenchymal tissue in the chamber angle, posterior embryotoxon, congenital corneal hyaline membrane, posterior marginal dysplasia, prominent Schwalbe line, mesodermal dysgenesis, and internal corneal ulcer seem all to fall in this same category of the anterior chamber cleavage syndrome; condition is present at birth; about 80% are bilateral; autosomal dominant inheritance; may be associated with congenital sensory neuropathy and ichthyosis.

Ocular: Increased intraocular pressure; adhesions between the iris and cornea; persistence of mesenchymal tissue in the chamber angle; usually shallow anterior chamber; iris coloboma and hypoplasia; prominent Schwalbe ring; contiguous hyaloid membrane; corneal opacities of various density with or without edema, usually at the site of iris adhesion; anterior pole cataract; remains of hyaloid artery.

Clinical: Dental anomalies; mental retardation; cleft palate; syndactyly; craniofacial dysostosis; myotonic dystrophy.

Kolbert GS, Seelenfreund M. Sclerocornea, anterior cleavage syndrome and trisomy 18. *Ann Ophthalmol* 1970; 2:26.

Merin S. Anterior chamber cleavage syndromes. In: Merin S, ed. *Inherited Eye Diseases, Diagnosis and Clinical Management*. New York: Marcel Dekker, 1991:74-78.

Quinlivan R, et al. Congenital sensory neuropathy in association with ichthyosis and anterior chamber cleavage syndrome. *Neuromuscul Disord* 1993; 3:217-221.

Reese AB, Ellsworth RM. The anterior chamber cleavage syndrome. *Arch Ophthalmol* 1966; 75:307.

Anterior Segment Ischemia Syndrome 75

General: Occasional complication of strabismus surgery; usually occurs in adult patients who have paretic strabismus after extensive transposition procedures; also may be secondary to giant cell arteritis or develop following trabeculectomy or strabismus surgery.

Ocular: Corneal edema; corneal ulceration; uveitis; iris atrophy; ectopic pupil; posterior synechiae; cataract; hypotony; phthisis bulbi.

Birt CM, et al. Anterior segment ischemia in giant cell arteritis. *Can J Ophthalmol* 1994; 29:93-94.

Hiatt RL. Production of anterior segment ischemia. *J Pediatr Ophthalmol Strabismus* 1978; 15:197.

Saunders RA, Sandall GS. Anterior segment ischemia syndrome following rectus muscle transposition. *Am J Ophthalmol* 1982; 93:34-38.

Saunders RA, et al. Anterior segment ischemia after strabismus surgery. *Surv Ophthalmol* 1994; 38:456-466.

Watson NJ. Anterior segment ischemia. *Ophthalmic Surg* 1992; 23:429-431.

Anterior Spinal Artery Syndrome (Ventral Medullary Syndrome; Myelomalacia Syndrome; Beck Syndrome; Medullary Syndrome) 76

General: Impaired blood supply to the anterior two thirds of the spinal cord, which includes the corticospinal and lateral spinothalamic tracts and anterior horns of the gray matter; caused by thrombosis, aneurysms, or extramedullary tumors; onset is sudden, and symptoms are preceded by pain and paresthesias.

Ocular: Nystagmus.

Clinical: Quadriplegia (usually sudden with cervical lesions) with loss of sense of position; arms may be unimpaired with thoracic location of the lesion; anesthesia for determination of sensations; disturbed intestinal and bladder function with incontinence; disturbed sense for temperature and pain.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Steegmann AT. Syndrome of the anterior spinal artery. *Neurology* 1952; 2:15.

Anthrax 77

General: Disease of wild and domestic animals; transmitted to humans by contact with animals or their products; causative agent is *Bacillus anthracis.*

Ocular: Pustules and edema of lids; phlebitis of ophthalmic veins; optic neuritis; optic atrophy; panophthalmitis; itchy, erythematous papule of the eyelids.

Clinical: Necrotic cutaneous ulcer.

Amraoui A, et al. Anthrax of the eyelids. *Br J Ophthalmol* 1992; 76:753-754.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Tasman IS. *The eye manifestations of internal diseases*. St. Louis: CV Mosby, 1951:315-317.

Antimongolism Syndrome (Chromosome 21 Partial Deletion Syndrome; Monosomy 21 Partial Syndrome; G-Deletion Syndrome) 78

General: Partial monosomy of chromosome 21 with absence of the short arm and part of the long arm of this chromosome.

Ocular: Antimongoloid slant of lid fissures; blepharochalasis; sclerocornea.

Clinical: Hypertony; large ear lobes; prominent nasal bridge; mental retardation; pyloric stenosis; dystrophic nails; retarded growth; heart disease; hemivertebrae; micrognathia.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Doane JF, et al. Bilateral penetrating keratoplasty for sclerocornea in an infant with monosomy 21. Case report and review of the literature. *Cornea* 1994; 13:454-458.

Lejeune J, et al. Monosomie Partielle Pour un Petit Acrocentrique. *C R Acad Sci (Paris)* 1964; 259:4187.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Penrose LS. Antimongolism [Letter]. *Lancet* 1966; 1:497.

Antiphospholipid Antibody Syndrome (Hughes syndrome) 79

General: Recurrent arterial and venous thrombosis

Ocular: Subconjunctival hemorrhage, hyphema, vitreous hemorrhage, central retinal vein and artery occlusions

Behbehani R, Sergott RC, Sabino PJ: Antiphospholipid antibody syndrome: diagnostic aspects. *Current Opin Ophthalmol* 2004: 6: 483-485.

Gerkowicz M, Lutalska M, Kosior-Jarecka E, et al. Antiphospholipid syndrome and changes in the eye. *Klin Oczna* 2005; 107: 143-6.

Anton Syndrome (Denial-Visual Hallucination Syndrome) 80

General: Cause unknown, but isolation of diencephalon from occipital lobe would be necessary to result in the features of the syndrome; lesions of the calcarine-thalamic connections or bilateral destruction of the occipital regions have been claimed to cause denial of blindness; the disease is rare and little understood; has been reported in association with blindness from a peripheral lesion such as bifrontal contusions and optic nerve damage.

Ocular: Denial of blindness; patients may persistently deny having any loss of visual perception; the objects the patient describes and claims to see are regarded as visual hallucinations; visual field hemianopsia.

Clinical: Confabulation; allocheiria (reference of a sensation is made to the opposite side to which the stimulus is applied).

Anton G. Ueber die Selbstwahrnehmung der Herderkrankung des Gehirns durch den Kranken bei Rindentaubheit. *Arch Psychiatr Nervenkr* 1899; 32:86.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McDaniel KD, McDaniel LD. Anton's syndrome in a patient with posttraumatic optic neuropathy and bifrontal contusions. *Arch Neurol* 1991; 48:101-105.

Tyler HR. Cerebral disorders of vision. In: Smith JL, ed. *Neuro-Ophthalmology, vol. IV.* St. Louis: CV Mosby, 1968.

Apert Syndrome (Acrocephalosyndactylism Syndrome; Acrocranio-dysphalangia; Acrodysplasia; Sphenoacrocranio-syndactyly; Absent-Digits-Cranial-Defects Syndrome) 81

General: Inherited; most often recessive, sometimes dominant; an extreme form of Apert syndrome has been described as Carpenter syndrome, with the latter being familial and transmitted as an autosomal recessive.

Ocular: Shallow orbit; exophthalmos; hypertelorism; ptosis; strabismus; nystagmus; ophthalmoplegia; hyperopia; exposure keratitis; cataracts; ectopia lentis; medullated nerve fibers; retinal detachment; papilledema with subsequent optic atrophy; keratoconus.

Clinical: Oxycephaly ("tower skull"); syndactyly (symmetrically); synostoses and synarthroses of shoulder and elbows frequent; agenesis of spinal bones and limbs; headaches; hypertelorism; hypoplastic maxilla; acrocephaly; abnormality of sutures.

Apert E. Acrocephalosyndactylia. *Bull Soc Med Hop Paris* 1906; 23: 1310.

Buncic JR. Ocular aspects of Apert syndrome. *Clin Plast Surg* 1991; 18:315-319.

Ferraro NF. Dental, orthodontic, and oral/maxillofacial evaluation and treatment in Apert syndrome. *Clin Plast Surg* 1991; 18:291-307.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Heeckt P, et al. Early treatment of pan craniofacial synostosis. *Ann Plast Surg* 1993; 30:312-319.

Kaplan LC. Clinical assessment and multispecialty management of Apert syndrome. *Clin Plast Surg* 1991; 18: 217-225.

Krueger JL, Ide CH. Acrocephalosyndactyly (Apert's syndrome). *Ann Ophthalmol* 1974; 6:787.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Mah J, Kasser J, Upton J. The foot in Apert syndrome. *Clin Plast Surg* 1991; 18:391-397.

Marchac D, Renier D. Craniosynostosis. *World J Surg* 1989; 13:358-365.

Marchac D, Renier D, Broumand S. Timing of treatment for craniosynostosis and facio-craniosynostosis: a 20 year experience. *Br J Plast Surg* 1994; 47:211-222.

Margolis S, et al. Oculocutaneous albinism associated with Apert's syndrome. *Am J Ophthalmol* 1977; 84:830-839.

Margolis S. Structural alterations of extraocular muscle associated with Apert's syndrome. *Br J Ophthalmol* 1977; 61:683-689.

McGill T. Otolaryngologic aspects of Apert syndrome. *Clin Plast Surg* 1991; 18:309-313.

Parker TL, Roth JC, Esterly NB. Isotretinoin for acne in Apert syndrome. *Pediatr Dermatol* 1992; 9:298-300.

Suparta N, Hartono, Sunartini. Apert syndrome. *Paediatr Indones* 1991; 31:319-324.

Thompson D, et al. Assessment and treatment of craniosynostosis. *Br J Hosp Med* 1994; 52: 17-24.

Thompson DN, et al. Subdural intracranial pressure monitoring in craniosynostosis: its role in surgical management. *Childs Nerv Syst* 1995; 11:269-275.

Arcus Cornea (Arcus Senilis) 82

General: Autosomal dominant; occurs earlier in blacks; frequently associated with abnormal serum lipid levels, but may occur without any predisposing factors; more commonly associated with familial hypercholesterolemia.

Ocular: Arcus of cornea.

Clinical: Can be associated with Tangier disease, Norum disease, and homozygotes type II hyperlipoproteinemia; osteogenesis imperfecta ring resembling arcus seen.

Barchiesi BJ, et al. The cornea and disorders of lipid metabolism. *Surv Ophthalmol* 1991; 36:1-22.

MacAraez PVJ, et al. Arcus not so senilis. *Ann Intern Med* 1968; 68:345-354.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Argininosuccinic Aciduria (Trichorrhexis Nodosa) 83

General: Deficient argininosuccinase (A Sase) deficiency; both sexes affected; prevalent in females; autosomal recessive inheritance.

Ocular: Friable tufted eyelashes and eyebrows; visual field defects; cataract.

Clinical: Clinical findings vary widely; mental retardation; seizures; ataxia; hepatomegaly; friable hair (trichorrhexis nodosa); may have citrullinemia; hyperammonemia; increased argininosuccinic acid (most pronounced in the cerebrospinal fluid).

Cecil RL. *Cecil textbook of medicine*, 16th ed. Philadelphia: WB Saunders, 1982: 1105.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Gerrits GP, et al. Argininosuccinic aciduria: Clinical and biochemical findings in three children with the late onset form, with special emphasis on cerebrospinal fluid findings of amino acids and pyrimidines. *Neuropediatrics* 1993; 24:15-18.

Widhalm K, et al. Long-term follow-up of 12 patients with the late-onset variant of argininosuccinic acid lyase deficiency: no impairment of intellectual and psychomotor development during therapy. *Pediatrics* 1992; 89[6 Pt 2]: 1182-1184.

Argyll Robertson Syndrome (Spinal Miosis) 84

General: Caused by syphilis or, rarely, epidemic encephalitis; disseminated sclerosis; diabetes; brain tumor; syringomyelia; syringobulbia; chronic alcoholism; injury; encephalitis lethargic a, Guillain-Barré syndrome, Lyme disease, multiple sclerosis, polyarteritis nodosa, and sarcoidosis have been associated with this condition.

Ocular: No direct or consensual pupil reaction to light but to normal accommodation (except in terminal stages, when pupil is fixed to all stimuli); pupil contraction with eserine but poor dilation with atropine; miosis (generally); irregular pupil; occurs unilaterally and bilaterally; anisocoria or discoria frequent.

Clinical: Syphilis of central nervous system; general paresis; tabes dorsalis.

Lebensohn JD. The Argyll Robertson pupil. *Am J Ophthalmol* 1969; 68:1119.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995.

Poole CJM. Argyll Robertson pupils due to neurosarcoidosis: evidence for site of lesion. *Br Med J* 1984; 289:356.

Robertson DA. Four cases of spinal miosis: with remarks on the action of light on the pupil. *Edinburgh Med J* 1869; 15:487.

Arndt-Gottron Syndrome (Scleromyxedema) 85

General: Etiology unknown; rare cutaneous disease of hyaluronic acid deposits in dermis; variant of lichen myxedematosus (papular mucinosis); progressive disease commonly involving the face, neck, upper trunk, forearms, and hands, producing thickening of the skin with overlying fine papules.

Ocular: Corneal opacities of amyloid deposits; thickening of eyelids; lagophthalmos; ectropion; thickened eyebrow or eyelid skin; corneal opacities.

Clinical: Exaggerated facial folds impair opening of the mouth; flexion contractures from poor joint mobility; erythema; scaling of skin; phimosis; urethral stenosis.

Davis ML, et al. Ophthalmic findings in scleromyxedema. *Ophthalmology* 1994; 101:252-255.

Goldin HM, et al. Scleromyxedema with corneal deposits. *Ophthalmology* 1987; 94:1334-1337.

Pusateri TJ, et al. Corneal manifestations of scleromyxedema. *Ophthalmology* 1987; 94:510-513.

Arnold Pick Syndrome (Aphasia-Agnosia-Apraxia Syndrome; Pick Syndrome [2]; Pick Disease of the Brain) 86

General: Widespread cortical atrophy; manifested between 40 and 70 years; pathogenesis remains unknown; cannot be consistently differentiated from Alzheimer disease on clinical grounds alone.

Ocular: Apperceptive blindness (inability of patient to fix upon objects within his or her gaze); visual agnosia (inability to recognize familiar objects by sight); visual field defects due to atrophy in occipital lobe.

Clinical: Presenile or progressive dementia; patient is unaware of his or her surroundings; poor insight; loss of words and utterance of stereotyped phrases; aphasia (motor type); apathy and indifference.

Magalini SI, Scrasia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Morris JC, et al. Hereditary dysphasic dementia and the Pick-Alzheimer spectrum. *Ann Neurol* 1984; 16:455-466.

Pick A. Apperzeptive Blindheit der Senilen. *Arb Dtsch Psychiatr Klin Prag* 1908:43.

Arnold-Chiari Syndrome (Platybasia Syndrome; Cerebellomedullary Malformation Syndrome; Basilar Impressions) 87

General: Malformation of the hindbrain; developmental deformity of the occipital bone and upper cervical spine; recognized in children or adults; clinical picture may be indistinguishable from that of Dandy-Walker syndrome in infants.

Ocular: Horizontal, vertical, and rotary forms of nystagmus; vertical nystagmus in both up gaze and down gaze is most common; papilledema; esotropia; Duane retraction syndrome (association); oscillopsia.

Clinical: Hydrocephalus; cerebellar ataxia; bilateral pyramidal tract signs.

Arnold J. Myelocyste, Transposition von Gewebskeimen und Sympodie. *Beitr Path Anat Allgem Path Jena* 1894; 16: 1.

Bixenman WW, Laguna JF. Acquired esotropia as initial manifestation of Arnold-Chiari malformation. *J Pediatr Ophthalmol Strabismus* 1987; 24:83.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Yamanouchi H, et al. Duane retraction syndrome associated with Chiari I malformation. *Pediatr Neural* 1993; 9: 327-329.

Arterial Occlusive Retinopathy and Encephalopathy Syndrome 88

General: Rare; etiology unknown but may be virally induced, immune-mediated disease; most frequent in women; mechanism could be related to microangiopathy secondary to immunologically mediated vasculitis, although an abnormal coagulation system or microembolisms have been proposed to explain this condition.

Ocular: Multiple branch retinal arterial occlusions; rotary nystagmus; retinal hemorrhage; visual field defects; bilateral gaze palsy.

Clinical: Encephalopathy; behavior and memory disturbances; hearing loss; paranoid psychosis; neurologic dysfunction; seizures; headache; spasticity and hyperreflexia.

Bonamo J, et al. Anencephaly. *J Med Soc N J* 1980; 77:439.

Cappeto JR, et al. A syndrome of arterial occlusive retinopathy and encephalopathy. *Am J Ophthalmol* 1984; 98: 189-202.

Gordon DL. Microangiopathy of the brain, retina and ear: improvement without immunosuppressive therapy. *Stroke* 1991; 22:933-937.

Nicolle MW, McLachlan RS. Microangiopathy with retinopathy, encephalopathy, and deafness (RED-M) and systemic features. *Semin Arthritis Rheum* 1991; 21:123-128.

Arteriosclerosis 89

General: Thickening and induration of the arterial wall; prominent in the elderly.

Ocular: Increased arterial light reflex, copper/silver wire arteries; arteriovenous crossing changes; arterial caliber variation/irregularity; arterial straightening or tortuosity; intimal hyperplasia, medial atrophy, atherosclerotic fibrous plaques and calcifications of the internal elastic lamina observed in aged human orbital arteries.

Clinical: Increased collagen deposition in small- and medium-sized arteries with progressive replacement of the smooth muscle in the vessel walls; arterial wall changes at arteriovenous crossings.

Buchi ER, et al. Common histopathological changes in aged human orbital arteries. *Int Ophthalmol* 1993; 17:37-42.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:269.

Arteriovenous Fistula (Arteriovenous Aneurysm; Arteriovenous Angioma; Arteriovenous Malformation; Cirsoid Aneurysm; Racemose Hemangioma; Varicose Aneurysm) 90

General: Abnormal communications between arteries and veins that allow arterial blood to enter the vein directly without traversing a capillary network; may be congenital or secondary to penetrating trauma or blunt trauma.

Ocular: Uveitis; chemosis and neovascularization of conjunctiva; bullous keratopathy; eyelid edema; ptosis; exophthalmos; iris atrophy; papilledema; retinal hemorrhages; cataract; paresis of third or sixth nerves; glaucoma; upper lid tumor; total choroidal detachment; leaking retinal macroaneurysms; central retinal vein occlusion; iris neovascularization.

Clinical: Cerebral hemorrhage; death; substernal pain; dyspnea; varicose veins.

Barke RM, et al. Spontaneous dural carotid-cavernous fistula with central retinal vein occlusion and iris neovascularization. *Ann Ophthalmol* 1991; 23: 11-17.

Busin M, et al. Arteriovenous fistula presenting as a tumor of the upper eyelid. *Ophthalmic Surg* 1994; 25:471-473.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Tilanus MD, et al. Congenital arteriovenous communications and the development of two types of leaking retinal macroaneurysms. *Am J Ophthalmol* 1991; 112:313.

Wang ML, et al. Total choroidal detachment complicating dural arteriovenous sinus fistula. *Ophthalmic Surg* 1993; 24:856-857.

Wolter JR. Arteriovenous fistula involving the eye region. *J Pediatr Ophthalmol* 1975; 12:22-39.

Wolter JR. Arteriovenous fistula of the eyelid: secondary to a chalazion. *J Pediatr Ophthalmol* 1977; 14:225-227.

Arthrogryposis Multiplex Congenita 91

General: Heterogeneous group of disorders of multiple proposed etiologies; often one manifestation of a complex of congenital anomalies; probable autosomal recessive transmission; found in Eskimos; affects more males than females; characterized by decreased fetal joint mobility secondary to neuropathic disease, myopathic disease, or some other cause.

Ocular: Congenital bilateral cataract; associated with ophthalmoplegia, retinopathy, goniodysgenesis, and infantile glaucoma, as well as Duane retraction syndrome.

Clinical: Multiple articular rigidities; hypoplasia of adjacent muscle groups; soft tissue shortening; ducklike waddle; muscle atrophy.

Beckerman RC, Buchino JJ. Arthrogryposis multiplex congenita as part of an inherited symptom complex: two case reports and a review of the literature. *Pediatrics* 1978; 61:417-422.

Brooks JG Jr, Coster DJ. Arthrogryposis multiplex congenita: a report of two cases. *Aust N* Z *J Ophthalmol* 1994; 22: 127-132.

Drachman DB. The syndrome of arthrogryposis multiplex congenita. *Birth Defects* 1971; 7:90-97.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller BA, Pollard ZF Duane's retraction syndrome and arthrogryposis multiplex congenita. *Surv Ophthalmol* 1994; 38:395-396.

Schrander-Stumpel CT, et al. Arthrogryposis, ophthalmoplegia, and retinopathy: confirmation of a new type of arthrogryposis. *J Med Genet* 1993; 30:78-80.

Arylsulfatase A Deficiency (Metachromatic Leukodystrophy; Sulfatide Lipoidosis Syndrome; Greenfield Disease; Scholz Syndrome; Scholz-Bielschowsky-Henneberg Syndrome; Van Bogaert-Nyssen Disease; Van Bogaert-Nyssen-Peiffer Disease; Familial Progressive Cerebral Sclerosis; Infantile Progressive Cerebral Sclerosis; Infantile Metachromatic Leukodystrophy; Leukodystrophia Cerebri Progressiva Metachromatica Diffusa; Opticochleodentate Degeneration) 92

General: Accumulation of sulfatide caused by deficient activity of arylsulfatase A; autosomal recessive; familial form of metachromatic leukodystrophy; Greenfield disease (late infantile form); van Bogaert-Nyssen-Peiffer syndrome (adult form); affects central and peripheral nervous systems by demyelination and by accumulation of metachromatic material.

Ocular: Visual loss in association with optic atrophy; strabismus; macular cherry-red spot; corneal opacification; oculomotor disorders (nystagmus, strabismus); optic nerve and retinal demyelination.

Clinical: Motor and mental deterioration with spasticity; paralysis; seizures; dementia; death in early childhood, although attenuated and adult forms of the disease occur; schizophrenia; temporooccipital demyelination; unreactive to visual and auditory stimuli; adult form: moodiness, withdrawal, megalomania, hallucinations, violent reactions, and dementia.

Evans OB. Inborn errors of metabolism of the nervous system: metachromatic leukodystrophy. In: Bradley WG, ed. *Neurology in Clinical practice, vol. II.* 2nd ed. Boston: Butterworth-Heinemann, 1995:1555-1556.

Fenichel GM. Metachromatic leukodystrophy. In: Fenichel GM, ed. *Clinical Pediatric Neurology,* 2nd ed. Philadelphia:WB Saunders, 1993:139-140.

Goebel HH, et al. The ultrastructure of the retina in adult metachromatic leukodystrophy. *Am J Ophthalmol* 1978; 85:841-849.

Goebel HH, et al. Ultrastructural study of the retina in late infantile metachromatic leukodystrophy. *Ophthalmic Res* 1992; 24:103-109.

Jablonski S. Illustrated dictionary of eponymic syndromes and diseases. Philadelphia: WB Saunders, 1969:128.

Kolodny EH, et al. Genetic heterogeneity in aryl sulfatase A deficiency. *Neurology* 1979; 29:576.

Quigley HA, Green WR. Clinical and ultrastructural ocular histopathologic studies of adult-onset metachromatic leukodystrophy. *Am J Ophthalmol* 1976; 82:472-479.

Scott IU, et al. New sites of ocular involvement in late-infantile metachromatic leukodystrophy revealed by histopathologic studies. *Graefes Arch Clin Exp Ophthalmol* 1993; 231:187-191.

Weiter JJ, et al. Retinal pigment epithelial degeneration associated with leukocytic arylsulfatase A deficiency. *Am J Ophthalmol* 1980; 90:768-772.

Ascariasis 93

General: Roundworm infection caused by *Ascaris lumbricoides.*

Ocular: Conjunctivitis; xerosis; periphlebitis; pigmentation of macular lesion; papilledema; uveitis; subluxation of lens; scotoma; secondary glaucoma; vitreal hemorrhages; possible association with phlyctenular eye disease.

Clinical: Occasional colicky abdominal pain; slight abdominal distention; pneumonitis; intestinal obstruction.

Bratton RL, Nesse RE. Ascariasis. An infection to watch for in immigrants. *Postgrad Med* 1994; 393:171-173, 177-178.

Donnelly JJ, et al. Intraocular IgE antibody induced in guinea pigs with Ascaris suum larvae. *Invest Ophthalmol* 1977; 16:976-981.

Esrey SA, et al. Effects of improved water supply and sanitation on ascariasis, diarrhea, dracunculiasis hookworm infection, schistosomiasis, and trachoma. *Bull World Health Organ* 1991; 69:609-621.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Most H. Treatment of common parasitic infections of man encountered in the United States. *N Engl J Med* 1972; 287: 495-498.

Schuster DI, et al. Ascariasis: its complications, unusual presentations and surgical approaches. *South Med J* 1977; 70: 176-178.

Ascher Syndrome (Blepharochalasis with Struma and Double Lip) 94

General: Rare occurrence; blepharochalasis transmitted as a simple dominant; related to development of thyroid gland; symptoms usually start around puberty.

Ocular: "Bulging" of orbital fat; blepharochalasis; protrusion of lacrimal gland; entropion (rare).

Clinical: Goiter; reduplication of upper lip; hypothyroidism; alopecia areata totalis.

Ascher KW. Blepharochalasis mit Struma und Doppellipe. *Klin Monatsbl Augenheilk* 1920; 65:86.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Mathew MS, et al. Ascher's syndrome: an unusual case with entropion. *Int J Dermatol* 1992; 31:710-712

Aspergillosis 95

General: Systemic infection common in poultry farmers, feeders or breeders of pigeons, and persons who work with grains; should be considered in immunocompromised patients.

Ocular: Corneal ulcer; blepharitis; keratitis; scleritis; endophthalmitis; exophthalmos; retinal hemorrhages; retinal detachment; vitreitis; cataract; conjunctivitis; orbital cellulitis; paresis of extraocular muscles; secondary glaucoma; scleromalacia perforans; endogenous endophthalmitis; anterior chamber mass; invasion of choroid and anterior optic nerve.

Clinical: Pulmonary infections; invasive fungal disease.

Clarkson JG, Green WR. Endogenous fungal endophthalmitis. In: Duane TD, ed. *Clinical Ophthalmology, vol. III, part II*. Philadelphia: JB Lippincott, 1982:14-15.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Jones BR. Principles in the management of oculomycosis. *Trans Am Acad Ophthalmol Otolaryngol* 1975; 79: 15-53.

Kalina PH, Campbell RJ. Aspergillus terreus endophthalmitis in a patient with chronic lymphocytic leukemia. *Arch Ophthalmol* 1991; 109:102-103.

Katz G, et al. Ocular aspergillosis isolated III the anterior chamber. *Ophthalmology* 1993; 100:1815-1818.

Wolter JR. Diagnosis and management of orbital aspergillosis. *Ann Ophthalmol* 1976;8: 17-20.

Asthma (Hayfever) 96

General: Asthma characterized by paroxysms of expiratory dyspnea and wheezing, overinflation of the lungs, cough, and rhonchi; causes include allergy to external inhaled allergens, respiratory infections, and psychophysiologic reaction to stress. Hayfever (allergic asthma) characterized by sneezing, rhinorrhea, swelling of nasal mucosa, and itchy eyes; caused by spread of pollens in air or exposure to antigens; seasonal; occurs most frequently in young persons.

Ocular: Lacrimation; allergic conjunctivitis; periocular xanthogranulomas.

Clinical: Rhinorrhea; sneezing; mucosal swelling with occlusion of airway; insomnia; nasal polyps; wheezing; cough; headache; rhinitis.

Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Jakobiec FA, et al. Periocular xanthogranulomas associated with severe adult-onset asthma. *Trans Am Ophthalmol Soc* 1993; 91:99-125, discussion 125-129.

Norman PS. Treatment of allergic rhinitis. *Johns Hopkins Med J* 1967; 121:49.

Ataxia, Spastic, with Congenital Miosis 97

General: Autosomal dominant.

Ocular: Congenital miosis; nystagmus; small, nonreacting pupils.

Clinical: Symmetrical ataxia of gait and limb movement; dysarthria; late in walking; slurred speech; increased deep tendon reflexes; extensor plantar reflexes.

Dick DJ. Hereditary spastic ataxia with congenital miosis; four cases in one family. *Br J Ophthalmol* 1983; 67: 97-10l.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Atherosclerosis 98

General: Etiologic importance of lipid infiltration; cholesterol; patchy nodular form of arteriosclerosis.

Ocular: Central retinal artery obstruction; branch retinal artery obstruction; most common cause is embolization from carotid plaques.

Clinical: Atheroma, which is a nodular, subendothelial lipid deposition in the intima of large- and medium-sized arteries.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:269.

Muller M, et al. Carotid artery disease in vascular ocular syndromes. *J Clin Neuro-Ophthalmol* 1993; 13:175-180.

Walsh JB. Cardiovascular disorders. In: Duane TD, ed. *Clinical Ophthalmology, vol. V*. Philadelphia: JB Lippincott, 1982:5-6.

Atopic Dermatitis (Atopic Eczema; Besnier Prurigo) 99

General: Highly specific disease resulting from a heredity determined lowered cutaneous threshold to pruritus and characterized by intense itching; elevated total and specific immunoglobulin E.

Ocular: Keratoconjunctivitis; keratoconus; cataract; atopic dermatitis of lid; secondary glaucoma; uveitis; possible association with retinal detachment; pannus; blepharoconjunctivitis; corneal scarring; suppurative keratitis.

Clinical: In infants it involves the face with dry or oozing erythematous patches; in children and adolescents itching localized in the neck, antecubital spaces, popliteal folds, and ears; seborrheic changes.

Akova YA, et al. Atypical ocular atopy. *Ophthalmology* 1993; 100:1367-137l.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hogan MJ. Atopic keratoconjunctivitis. *Am J Ophthalmol* 1953; 36:937-947.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Matsuo N, et al. Photoreceptor outer segments in the aqueous humor of patients with atopic dermatitis and retinal detachment. *Am J Ophthalmol* 1993; 115:21-25.

Oka C, et al. Retinal detachment with atopic dermatitis similar to traumatic retinal detachment. *Ophthalmology 1994;* 101:1050-1054.

Tuft SJ. Clinical features of atopic keratoconjunctivitis. *Ophthalmology* 1991; 98:150-158.

Autoimmune Corneal Endotheliopathy 100

General: Etiology unknown; associated with implantation of an intraocular lens (IOL), pars planitis, iritis, secondary herpetic keratitis, and corticosteroid use; rare.

Ocular: Stromal edema; migrating line of keratic precipitates; iritis; clouding of the cornea; lymphocytes and macrophages in anterior chamber; linear pigmented endothelial line; evidence supports that herpes simplex virus can be isolated in the aqueous humor of patients with this condition.

Clinical: Autoimmune disease, nonspecific.

Khodadoust AA, Attarzedah A. Presumed autoimmune corneal endotheliopathy. *Am J Ophthalmol* 1986; 102: 633-634.

O'Hashi Y, et al. Demonstration of herpes simplex virus DNA in idiopathic corneal endotheliopathy. *Am J Ophthalmol* 1991; 12:419-423.

O'Hashi Y, et al. Idiopathic corneal Endotheliopathy. *Arch Ophthalmol* 1985; 103:1666.

Paul RH. Presumed autoimmune corneal endotheliopathy. *Am J Ophthalmol* 1988; 105:519-522.

Autoimmunologically Mediated Syndrome (Lymphocytic Hypophysitis Associated with Dacryoadenitis Syndrome) 101

General: Lymphocytes infiltrate the hypophysis.

Ocular: Dacryoadenitis.

Clinical: Lymphocytic infiltration of the hypophysis by CD3 cells, T cells, and $CD20^+$ B cells is an autoimmune process that may rarely cause lacrimal gland swelling.

Hirowitz JJ, Rodgers KJA. Management of acquired dacryocystitis. *Can J Ophthalmol* 1983; 13:213-216.

Jakobiec FA, Iwanoto T, Knowles DM. Ocular adnexal lymphoid tumors: correlative ultrastructural and immunological markers studies. *Arch Ophthalmol* 1982; 100:84-98.

Karesh JW, Permank I, Rodriguez MM. Dacryocystitis associated with dacryoadenitis. *Arch Ophthalmol* 1999; 117: 959-962.

Lee J-H, et al. Lymphocytic hypophysitis: occurrence in two men. *Neurosurgery* 1994; 43:159-163.

Avitaminosis B2 (Ariboflavinosis; Pellagra) 102

General: Niacin deficiency.

Ocular: Conjunctivitis; corneal vascularization; keratitis; pupillary dilation; optic atrophy; optic neuritis; cataract; blepharitis; central scotoma; marked photophobia.

Clinical: Occasional cranial nerve palsies; dermatitis; glossitis; gastrointestinal and nervous system dysfunction; mental deterioration; diarrhea; stomatitis.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:272.

Harley RD, ed. *Pediatric Ophthalmology*. 4th ed. Philadelphia: WB Saunders, 1998.

Avitaminosis C (Scurvy; Vitamin C Deficiency) 103

General: Vitamin C deficiency.

Ocular: Hemorrhages of lids, anterior chamber, vitreous cavity, retina, subconjunctival space, and orbit (most prominent, with resulting exophthalmos); keratitis, corneal ulcer; cataract.

Clinical: Increased capillary fragility with a tendency to hemorrhage in tissues throughout the body; poor wound healing; loose teeth; purpuric rash.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:273.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Taylor A, et al. Relationship in humans between ascorbic acid consumption and levels of total and reduced ascorbic acid in lens, aqueous humor, and plasma. *Curr Eye Res* 1991; 10:751-759.

Wilson JD. Vitamin deficiency and excess. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:472-480.

Woodruff PW, et al. Neuromyopathic complications in a patient with anorexia nervosa and vitamin C deficiency. *Int J Eating Disord* 1994; 16:205-209.

Avulsed Retinal Vessel Syndrome 104

General: Visual prognosis good; reports of avulsed retinal vessels not associated with retinal breaks.

Ocular: Recurrent vitreous hemorrhages caused by an avulsed retinal vessel, during retinal tear formation; vitreous hemorrhages may recur until interruption of vessel occurs.

Bustros SD, Welch PB. The avulsed retinal vessel syndrome. *Ophthalmology* 1982; 89[Suppl]: 189.

Uto M, et al. Two cases of isolated avulsed retinal vessels. *Nippon Ganka Gakkai Zasshi Acta Soc Ophthalmol Jpn* 1992; 96:541-545.

Axenfeld-Rieger Syndrome (Posterior Embryotoxon; Axenfeld Syndrome) 105

General: Dominant inheritance; occasionally sporadic; variable in expression.

Ocular: Posterior embryotoxon: ringlike opacity of cornea; long trabecula; prominent Schwalbe line; iris adhesions to Schwalbe line and cornea with large abnormal iris processes or broad sheets of tissues of varying size and location; anterior layer of iris may appear hypoplastic; ectopia of the pupil not uncommon; polycoria occurs; ringlike opacity of the deep corneal layers extending several millimeters from the limbus in continuity with the sclera; keratoconus.

Alward WLM. Axenfeld-Rieger syndrome in the age of molecular genetics. *Am J Ophthalmol* 2000; 130: 107-115.

Axenfeld T. Embryotoxon corneae posterius. *Berl Dtsch Ophth Ges* 1920; 42:301.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Nemet P, et al. Spontaneous filtering blebs in Axenfeld syndrome. *Am J Ophthalmol* 1973; 76:590.

Axenfeld-Schurenberg Syndrome (Cyclic Oculomotor Paralysis) 106

General: Congenital manifestation; frequently unilateral.

Ocular: Cyclic oculomotor paralysis (paralysis alternating with spasm); during periods of paralysis, lid exhibits ptosis and affected eye is abducted; during spasm, lid is raised, deviation of affected eye is either inward or outward, and pupil is fixed and contracted.

Axenfeld T, Schurenberg L. Beitrage zur Kenntnis der Angeborenen Beweglichkeitsdefekte des Auges. *Klin Monastbl Augenheilkd* 1901; 39:64.

Hamed LM. Oculomotor palsy with cyclic spasm. In: Margo CE, Mames R, Hamed LM, eds. *Diagnostic Problems in Clinical Ophthalmology*. Philadelphia: WB Saunders, 1994:712.

Levy MR. Cyclic oculomotor paralysis with optic atrophy. *Am J Ophthalmol* 1968; 65:766.

Purvin V. Oculomotor palsy in children. In: Margo CE, Mames R, Hamed LM, eds. *Diagnostic Problems in Clinical Ophthalmology*. Philadelphia: WB Saunders, 1994:682-683.

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Babinski-Nageotte Syndrome (Medullary Tegmental Paralysis) 107

General: Lesion in pontobulbar transitional region (corpus restiforme, Deiters nucleus, sympathetic fibers); Horner triad is always part of this syndrome; the findings are similar to Cestan-Chenais syndrome and Wallenberg syndrome; rare condition caused by ischemic lesion of the medulla oblongata involving the unilateral and medial areas of the medulla.

Ocular: Enophthalmos; ptosis; nystagmus; miosis.

Clinical: Contralateral hemiparesis and disturbance of sensibility; ipsilateral cerebellar hemiataxia; perhaps ipsilateral analgesia of the face, vocal cord, and soft palate; adiadochokinesis; lateral pulsion; dysmetria.

Babinski JFF, Nageotte J. Hemiasynergie, Lateropulsion et Myosis Bulbaires Avec Hemianesthesie et Hemiplegie Croisees. *Rev Neurol (Paris)* 1902; 10:358.

Kistler JP, Ropper AH, Martin JB. Cerebrovascular diseases. In: Isselbacher KJ, et al. eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:2242-2243.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology*. 4th ed. Baltimore: Williams & Wilkins, 1987.

Mossuto-Agatiello L, Kniahynicki C. The syndrome of Babinski and Nageotte. *Stroke* 1992; 23:919-920.

Bacillus Cereus 108

General: Highly virulent pathogen; most common contaminant of drug injection paraphernalia; usually enters the body as a result of penetrating trauma with a contaminated metallic foreign object; cause of food poisoning is toxin induced.

Ocular: Hypopyon; ring abscess of cornea; panophthalmitis; phthisis bulbi; orbital cellulitis; proptosis; vitreous abscess; necrosis of retina; endophthalmitis; keratitis.

Clinical: Fever; leukocytosis; septicemia; meningitis; endocarditis; osteomyelitis; wound infection.

David DB, et al. Bacillus cereus endophthalmitis. *Br J Ophthalmol* 1994; 78:577-580.

Drobniewski FA. Bacillus cereus and related species. *Clin Microbiol Rev* 1993; 6:324-338.

O'Day DM, et al. The problem of Bacillus species infection with special emphasis on the virulence of Bacillus cereus. *Ophthalmology* 1981; 88:833-838.

Tuazon CU, et al. Serious infections with Bacillus sp. *JAMA* 1979; 241:1137-1140.

Bacillus Subtilis (Hay Bacillus) 109

General: Gram-positive rod found in air, soil, dust, water, milk, and hay; frequently seen in people who work near hay.

Ocular: Conjunctivitis; ring abscess of cornea; corneal ulcer; endophthalmitis; panophthalmitis; dacryocystitis; orbit abscess.

Clinical: Fever; leukocytosis.

Awan KJ. Bacillus endophthalmitis. *Ophthalmic Surg* 1992; 23:368.

Forester RK, et al. Management of infectious endophthalmitis. *Ophthalmology* 1980; 87:313-318.

O'Day DM, et al. The problem of Bacillus species infection with special emphasis on the virulence of Bacillus cereus. *Ophthalmology* 1981; 88:833-838.

Bacterial Endocarditis 110

General: Inflammation of the lining on the heart caused by an infective agent.

Ocular: Conjunctival petechial hemorrhages; retinal hemorrhages; Roth spots; cotton-wool spots; branch or central retinal arterial occlusion; metastatic endophthalmitis; iridocyclitis; optic disk edema; cranial nerve palsies; diplopia; nystagmus; choroidal abscess; choroidal neovascular membrane; anterior segment necrosis.

Clinical: Emboli of the central nervous system; fever; splenomegaly; heart murmur; embolic episodes.

Coil GE, Lewis H. Metastatic choroidal abscess and choroidal neovascular membrane associated with Staphylococcus aureus endocarditis in a heroin user. *Retina* 1994; 14:256-259.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:221.

Moffett DG, Edward DO. Anterior segment necrosis associated with endogenous endophthalmitis secondary to group C streptococcal septicemia. *Can J Ophthalmol* 1991; 26:283-287.

Walsh JB. Cardiovascular Disorders. In: Duane TD, ed. *Clinical Ophthalmology. vol. V.* Philadelphia: JB Lippincott, 1982:1-2.

Balint Syndrome (Psychic Paralysis of Visual Fixation Syndrome) 111

General: Bilateral lesion of parietooccipital region; rare occurrence; affected patients are unaware of objects otherwise familiar to them.

Ocular: Psychic paralysis of visual fixation; lack of full voluntary control of eye movements; unstable visual fixation.

Clinical: Tonic and motor phenomena of upper limbs; loss of body coordination (bilateral); optic ataxia; it has been reported to occur in association with human immunodeficiency virus (HIV) encephalitis and with presenile-onset cerebral adrenoleukodystrophy.

Balint R. Seelenlahmung des Schauens, optische Ataxie, Raumliche Storung der Aufmerksamkeit. *Monatsschr Psychiatr Neurol* 1909; 25:51.

Cogan DG, Adams RD. A type of paralysis of conjugate Gaze (ocular motor apraxia). *Arch Ophthalmol* 1964; 71:489.

Damasio AR. Disorders of complex visual processing: agnosias, achromatopsia, Balint's syndrome, and related difficulties of orientation and construction. In: Mesulan M-M, eds. *Principles of Behavioral Neurology*. Philadelphia: FA Davis, 1985:259-262.

Gottlieb D, et al. Reappearance of the visual percept after intentional blinking in a patient with Balint's syndrome. *J Clin Neuro-Ophthalmol* 1991; 11:62-65.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Schnider A, et al. Balint's syndrome in subacute HIV encephalitis. *J Neurol Neurosurg Psychiatry* 1991; 54:822-825.

Uyama E, et al. Presenile-onset cerebral adrenoleukodystrophy presenting as Balint's syndrome and dementia. *Neurology* 1992; 43:1249-1251.

Baller-Gerold Syndrome (Craniosynostosis Radial Aplasia) 112

General: Autosomal recessive inheritance.

Ocular: Ocular hypertelorism; epicanthic folds.

Clinical: High nasal bridge; low philtrum; dysplastic ears; radius hypoplastic or absent; ulna short and bowed; carpal bones missing or fused; thumb hypoplastic or missing; craniosynostosis; anal, urogenital, cardiac, central nervous system, and vertebral defects; agenesis of frontal and parietal bones; midline facial angioma; scrotally positioned anus; microcephaly; erythroblastosis of the liver; pancreatic islet cell hypertrophy.

Baller F. Radiuasplasie und Inzucht. *Z Mensch Vererb Konstitutionsl Lehre* 1950; 29:782-790.

Dallapiccola B, et al. Baller-Gerold syndrome: case report and *Clinical* and radiological review. *Am J Med Genet* 1992; 42:365-368.

Lin AE, et al. Further delineation of the Baller-Gerold syndrome. *Am J Med Genet* 1993; 45:519-524.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Van Maldergem L, et al. The Baller-Gerold syndrome. *J Med Genet* 1992; 29:266-268.

Bamatter Syndrome (Osteoplastic Geroderma; Walt Disney Dwarfism) 113

General: Hereditary X-linked; rare; onset in early childhood; precocious aging; osteoporosis; autosomal recessive inheritance.

Ocular: Glaucoma; microphthalmia; microcornea; corneal opacities.

Clinical: Senile changes in skin; stunted growth; articular hypertrophy; multiple fractures and bone malformations; osteodysplasia; osteoporosis; dwarfism.

Bamatter F. Gerodermic ostheodysplastique hereditaire. *Ann Pediatr* 1950; 174:126-127.

Hunter AGW. Is geroderma osteodysplastica underdiagnosed? *J Med Genet* 1988; 25:854-857.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Bang Disease (Brucellosis; Malta Fever; Mediterranean Fever; Pig Breeder Disease; Gibraltar Fever; Undulant Fever) 114

General: Transmitted to man from animals or animal products containing bacteria of the genus *Brucella;* human infection results from ingestion of infected animal tissue and milk products or through skin wounds directly bathed in freshly killed animal tissues.

Ocular: Conjunctivitis; punctate keratitis; optic neuritis; swollen optic nerves; chorioretinitis; extraocular muscle palsies; phlyctenules; dacryoadenitis; papilledema; episcleritis; macular edema; phthisis bulbi; uveitis; vitreous opacities; changes in intraocular pressure (early decrease or late increase).

Clinical: Fever; icterus; weakness; sweats; general malaise; mammary abscess.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:236.

Gasser, I, et al. Bilateral mammary abscess and uveitis caused by Brucella melitensis: report of a case. *Injections* 1991; 19:44-45.

Lyall M. Ocular brucellosis. *Trans Ophthalmol Soc U K* 1973; 93:689-697.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Walker J, et al. Brucellosis and uveitis. *Am J Ophthalmol* 1992; 114:374-375.

Banti Disease (Chronic Congestive Splenomegaly; Fibrocongestive Splenomegaly; Splenic Anemia; Hepatolienal Fibrosis) 115

General: Etiology portal hypertension due to thrombosis, compression, or aneurysm; insidious or sudden onset; most frequently occurs before age 35 years; slowly evolving.

Ocular: Subconjunctival hemorrhage.

Clinical: Pallor; mild jaundice or brown pigmentation of skin; enlarged liver; weakness; melena; flatulence; diarrhea; epistaxis; vomiting of blood.

Banti G. Dell'Anemia Splenica. *Arch Sc Anat Patol (Firenze)* 1883; 2:53-122.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1991.

Baraitser-Winter Syndrome 116

General: X-linked mental retardation, macrosomia, macrocephaly, and obesity syndrome.

Ocular: Ptosis; hypertelorism; down-slanting palpebral fissures.

Clinical: May be confused with Noonan syndrome; phenotypic features appear to be variable.

Megarbana A, Le Merrer M, el Kallab K. Ptosis, down-slanting palpebral fissures, hypertelorism, seizures and mental retardation: a possible new MCA/MR syndrome. *Clin Dysmorphol* 1997; 6:239-244.

Monteiro de Pina-Neto J, de Molfetta GA. The X-linked mental retardation, macrosomia, macrocephaly and obesity syndrome (Baraitser syndrome): a Brazilian case [Letter]. *Clin Dysmorphol* 1998; 7:233-234.

Verloes A. Iris coloboma, ptosis, hypertelorism, and mental retardation: Baraitser-Winter syndrome or Noonan syndrome? *J Med Genet* 1993; 30:425-426.

Bardet-Biedl Syndrome 117

General: Polydactyly; obesity; cognitive delay; retinal degeneration; nystagmus.

Ocular: 30% to 65% of patients have clinical nystagmus that may mimic spasmus nutans but either lacks head nodding or is not suppressed by head nodding; retinal degeneration with attenuated retinal vessels and pale optic disks.

Clinical: Nystagmus may be a presenting sign, but patients will have polydactyly, obesity, and motor/cognitive delay.

Gottlob I, et al. Head nodding is compensatory in spasmus nutans. *Ophthalmology* 1992; 99:1024-1031.

Gottlob I, et al. Signs distinguishing spasmus nutans (with and without central nervous system lesions) from infantile nystagmus. *Ophthalmology* 1990; 97: 1166-11 75.

Gottlob I, Helbling A. Nystagmus mimicking spasmus nutans as the presenting sign of Bardet-Biedl syndrome. *Am J Ophthalmol* 1999; 128:770-772.

Gottlob I, Reinecke RD. Eye and head movements in patients with achromatopsia. *Graefes Arch Clin Exp Ophthalmol* 1994; 232:392-401.

Gottlob I, Wizov SS, Reinecke RD. Quantitative eye and head movement recordings of retinal disease mimicking spasmus nutans. *Am J Ophthalmol* 1995; 119:374-376.

Leys MJ, et al. Visual acuities and dark-adapted thresholds of children with Bardet-Biedl syndrome. *Am J Ophthalmol* 1998; 106:561-569.

Bare Lymphocyte Syndrome 118

General: Rare, severe combined immunodeficiency characterized by the lack of expression of human leukocyte antigen (HLA) A, B, and C antigens with severe T and B deficiency.

Ocular: Horizontal nystagmus; candida retinitis.

Clinical: Recurrent pulmonary infections; bronchiectasis; gastroenteritis; hepatomegaly; developmental delay; respiratory failure; death.

Chess J, et al. Candida retinitis in bare lymphocyte syndrome. *Ophthalmology* 1986; 93:696-698.

Holland GN, et al. Ocular disorders associated with a severe acquired cellular immunodeficiency syndrome. *Am J Ophthalmol* 1982; 93:393-402.

Barlow Syndrome (Mitral Valve Prolapse) 119

General: Common; usually benign; asymptomatic; predominant in females; vague with psychoneurotic basis.

Ocular: Retinal branch arterial occlusion; retinal emboli; keratoconus; ophthalmic migraine; amaurosis fugax; bilateral retinal artery occlusion.

Clinical: Myxomatous degeneration of the mitral valve; palpitation; chest pain; dyspnea; hyperventilation.

Bergen RL, et al. Bilateral arterial occlusion secondary to Barlow's syndrome. *Ann Ophthalmol* 1982; 14:673-675.

Ebner R. Visual field examination during transient migrainous visual loss. *J Clin Neuro-Ophthalmol* 1991; 11: 114-117.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Van Rhee F, et al. Bilateral retinal artery occlusion due to mitral valve prolapse. *Br J Ophthalmol* 1991; 75:436-437.

Barre-Lieou Syndrome (Posterior Cervical Sympathetic Syndrome) 120

General: Irritation of the vertebral nerve causing circulatory disturbance in the area of the cranial nuclei; fifth and eighth nerves mainly involved; trauma and arthritic changes involving the third and fourth cervical vertebrae or cervical disk pathology may be etiologic factor; course chronic; occurrence in older patients.

Ocular: Reduced vision (transitory); corneal hypesthesia in association with persistent corneal ulcers confined to the lid fissure.

Clinical: Headache; vertigo; mild dizziness; vasomotor disturbances of face and facial pain; laryngeal and pharyngeal paresthesia; chronic cervical arthritis; ear noises are frequent; anxiety; depression; impaired memory; difficulty in thinking.

Barre JA. Chronic vertebral arthritis and medullar disturbances: chronic vertebral arthritis and tumor of spinal cord. *Paris Med* 1925; 2:226.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Barrier Deprivation Syndrome (Binkhorst Membrane Deprivation Syndrome; Worst Decompartmentalization of Eye Syndrome) 121

General: Intracapsular cataract extraction; trauma to posterior capsule with extracapsular cataract extraction; more frequent in blue-eyed patients; often bilateral; cause thought to be increased pigment loss, which releases prostaglandins, creating allergic reaction.

Ocular: Cystoid macular edema; corneal endothelial dystrophy; retinal detachment; leakage in peripheral retina and macula; iris pigment loss; uveitis; vitreous in anterior chamber; retinal holes; band keratopathy; glaucoma; iritis.

Clinical: None.

Alpar JJ. Contribution to Binkhorst's membrane deprivation and Worst's decompartmentalization of the eye syndromes. *Ann Ophthalmol* 1980; 12:1399-1402.

Waitzman MD. Topical indomethacin in treatment and prevention of intraocular inflammation with special reference to lens extraction and cystoid macular edema. *Ann Ophthalmol* 1979; 11:489-491.

Bartsocas-Papas Syndrome 122

General: A rare autosomal recessive variant of popliteal pterygium syndrome (PPS).

Ocular: Ptergium.

Clinical: Popliteal webbing; cleft lip; cleft palate; lower lip pits; syndactyly; genital and nail abnormalities; equinus feet.

Papadia F, Zimbalatti F, LaRosa CG. The Bartsocas-Papas syndrome: autosomal recessive form of popliteal pterygium syndrome in a male infant. *Am J Med Genet* 1984; 17:841-847.

Basal Cell Carcinoma 123

General: Most common malignant neoplasm of lids; it can occasionally occur as a primary basal cell cancer of the conjunctiva and in the lacrimal canaliculus.

Ocular: Neoplasm most common on lower lid and medial canthus; lacrimation.

Clinical: Tumors of skin and other regions, including sinuses.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Fraunfelder FT, et al. The role of cryosurgery in external ocular and periocular disease. *Trans Am Acad Ophthalmol Otolaryngol* 1977; 83:713-724.

Garrett AB, et al. Basal cell carcinoma originating in the lacrimal canaliculus. *Ophthalmic Surg* 1993; 24:197-199.

Husain SE, et al. Primary basal cell carcinoma of the limbal conjunctiva. *Ophthalmology* 1993; 100: 1720-1722.

Payne JW, et al. Basal cell carcinoma of the eyelids. A long-term follow-up study. *Arch Ophthalmol* 1969; 81: 553-558.

Basal Cell Nevus Syndrome (Nevoid Basal Cell Carcinoma Syndrome; Nevoid Basalioma Syndrome; Gorlin Syndrome; Gorlin-Goltz Syndrome; Multiple Basal Cell Nevi Syndrome) 124

General: Autosomal dominant; onset of skin lesions in childhood, usually at puberty.

Ocular: Basal cell carcinomas of eyelids; strabismus; hypertelorism; congenital cataracts; choroidal colobomas; glaucoma; medullated nerve fibers; prominence of supraorbital ridges; corneal leukoma; basalioma of the skin; coloboma of the choroid and optic nerve.

Clinical: Basal cell tumors with facial involvement; shallow pits of the skin of the hands and feet; jaw cysts; rib anomalies; kyphoscoliosis and fusion of vertebrae; medulloblastoma; frontal and temporoparietal bossing; broad nasal root.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:263-264.

Dejong PT, et al. Medullated nerve fibers: a sign of multiple basal cell nevi (Gorlin's) syndrome. *Arch Ophthalmol* 1985; 103: 1833-1836.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gorlin RJ. Goltz RW Multiple nevoid basal cell epithelioma, jaw cysts and bifid rib: a syndrome. *N Engl J Med* 1960; 262:908.

Manzi G, et al. The Gorlin-Goltz syndrome: case report. *Ophthalmologica* 1990; 200:104-106.

Basedow Syndrome (Graves Disease; Hyperthyroidism; Thyrotoxicosis; Exophthalmic Goiter; Parry Disease) 125

General: Diffuse toxic goiter; inherited as a simple autosomal recessive; penetrance greater in females; however, dominant mode of inheritance and variable penetrance are possible; uncommon in either sex before age 15 years.

Ocular: Exophthalmos; swelling of eyelids and discoloration of upper eyelids; lid lag (von Graefe); globe lag (Koeber); lid trembling on gentle closure (Rosenbach sign); reduced blinking (Stellwag); retraction of upper lid; difficulty in everting upper lid (Gifford sign); convergence weakness (Möobius); impaired fixation on extreme lateral gaze (Suker); possible external ophthalmoplegia (Ballet); Dalrymple sign (staring appearance); tearing; photophobia; epiphora; prolapse of lacrimal gland; neuroretinal edema; tortuous vessels; papilledema and papillitis; anisocoria; keratitis; increased intraocular pressure; increased intraocular pressure on upgaze; decreased visual acuity; enlargement of the extraocular muscles; increased volume of the extraorbital fat; superior rectus muscle enlargement; decreased venous outflow.

Clinical: Tachycardia; anxiety; insomnia; loss of weight; hyperhidrosis; restlessness; myocarditis (toxic); atrial fibrillation.

Cohen JS. Optic neuropathy of Graves' disease, hyperthyroidism, and ocular myasthenia gravis. *Arch Ophthalmol* 1973; 90:131.

Hirano T. Association of Graves' disease with Turner's syndrome. *JAMA* 1981; 246:2429.

Hudson HL, et al. Graves exophthalmos unrelated to extraocular muscle enlargement. Superior rectus muscle inflammation may induce venous obstruction. *Ophthalmology* 1991; 98:1495-1499.

Kendler DL, et al. The initial characteristics of Graves' orbitopathy vary with age and sex. *Arch Ophthalmol* 1993; 111:197-20l.

Kramar P. Management of eye changes of Graves' disease. *Surv Ophthalmol* 1974; 18:369-382.

Ravin JG, et al. Orbital radiation for the ocular changes of Graves' disease. *Am J Ophthalmol* 1975; 79:285-288.

Wartofsky L. Diseases of the thyroid, Graves' disease. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. McGraw-Hill, 1994:942-946.

Werner SC. Modification of the classification of the eye changes of Graves' disease. *Am J Ophthalmol* 1977; 83: 725-727.

Bassen-Kornzweig Syndrome (Abetalipoproteinemia; Acanthocytosis; Familial Hypolipoproteinemia) 126

General: Inability to absorb and transport lipids; predominant in males; autosomal recessive inheritance; acanthocytosis, a peculiar burr cell malformation of the red blood cells; the basic defect is thought to be an inability to synthesize the apolipoprotein B peptide of low-density and very-low-density lipoproteins.

Ocular: Ptosis (may be present); nystagmus; progressive external ophthalmoplegia; retinitis pigmentosa (usually atypical); retinopathy develops with age after 10 to 14 years; optic atrophy occasionally; epicanthal folds; cataract; optic nerve pallor; hypopigmentation of retina; macular degeneration; dyschromatopsia.

Clinical: Steatorrhea; hypocholesterolemia; neurologic disorder with ataxia (similar to Friedreich ataxia); areflexia; Babinski sign; muscle weakness (facial, lingual; proximal and distal); slurred speech; lordosis; kyphosis.

Adams RD, Victor M. Bassen-Kornzweig acanthocytosis. In: Adams RD, Victor M, eds. *Principles of Neurology,* 5th ed. New York: McGraw-Hill, 1993:830.

Bassen FE, Kornzweig AL. Malformation of the erythrocytes in a case of atypical retinitis pigmentosa. *Blood* 1950; 5:381.

Carr RE. Abetalipoproteinemia and the eye. *Birth Defects* 1976; 12:385-408.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Rowland LP, Pedley TA. Abetalipoproteinemia. In: Rowland LP, ed. *Merritt's Textbook of Neurology,* 9th ed. Philadelphia: Williams & Wilkins, 1995:594-595.

Yee RD, et al. Atypical retinitis pigmentosa in familial hypobetalipoproteinemia. *Am J Ophthalmol* 1976; 82:64-71.

Batten-Mayou Syndrome (Spielmeyer-Vogt Syndrome; Mayou-Batten Disease; Stock-Spielmeyer- Vogt Syndrome; Cerebroretinal Degeneration; Pigmentary Retinal Lipoid Neuronal Heredodegeneration; Vogt-Spielmeyer Syndrome; Juvenile Ganglioside Lipidosis; Neuronal Ceroid Lipofuscinosis; Myoclonic Variant of Cerebral Lipidosis; Batten Disease; Cerebromacular Dystrophy; Juvenile Amaurotic Family Idiocy; Spielmeyer-Sjögren Syndrome) 127

General: Autosomal recessive; some cases of autosomal dominant; possible disturbance in lipid metabolism; most common in Jewish families; onset between ages 5 and 8 years; mean age at death is 17 years; poor prognosis (see Tay-Sachs Disease; Dollinger-Bielschowsky Syndrome.) The lipopigment storage diseases are divided into four types based on clinical and electron microscopic features: infantile (Hagberg-Santavuori syndrome), late infantile (Jansky-Bielschowsky disease), juvenile (Spielmeyer- Vogt disease), and adult (Kufs disease).

Ocular: Vision initially reduced, progressing to total blindness; fat deposition in the retina with gradual development of pigment disturbances resembling retinitis pigmentosa; progressive primary optic atrophy; granular pigmentary change of macula; there is clinical evidence supporting the idea that the primary lesion of the retina is in the inner layers.

Clinical: Mental disturbances; convulsions (later); apathy; irritability; ataxia; upper and lower motor neuron palsies; rigidity; complete paralysis and dementia in terminal stage; hypertonus; death from intercurrent infection.

Bateman JB, Philippart M. Ocular features of the Hagberg-Santavuori syndrome. *Am J Ophthalmol* 1986; 102: 262-271.

Batten FE, Mayou MS. Family cerebral degeneration with macular changes. *Proc Roy Soc Med (Sect Ophthalmol)* 1914; 8:70.

Brod RD, et al. Diagnosis of neuronal ceroid lipofuscinosis by ultrastructural examination of peripheral blood lymphocytes. *Arch Ophthalmol* 1987; 105:1388-1393.

Hittner HM, Zeller RS. Ceroid-lipofuscinosis (Batten disease). *Arch Ophthalmol* 1975; 93: 1778.

Horiguchi M, Miyake Y. Batten disease: deteriorating course of ocular findings. *Jpn J Ophthalmol* 1992; 36:91-96.

Srensen JB, Parnas J. A *Clinical* study of 44 patients with juvenile amaurotic family idiocy. *Acta Psychiatr Scand* 1979; 59:449-461.

Zeman W. Batten disease: ocular features, differential diagnosis and diagnosis by enzyme analysis. *Birth Defects* 1976; 12:441-453.

Bazzana Syndrome (Angiospastic Ophthalmo-Auricular Syndrome) 128

General: Rare.

Ocular: Visual fields have concentric contraction; retinal vascular tortuosity and irregular contours.

Clinical: Progressive deafness (bilateral), caused by otosclerosis.

Bazzana EC, et al. Otosclerosi e campo visivo. *Arch Ital Otol Rinol Lar* 1950; 61:620.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology,* 4th ed. Baltimore: Williams & Wilkins, 1987.

BBB Syndrome (Hypertelorism-Hypospadias Syndrome; Opitz Syndrome) 129

General: X-linked inheritance possible; differentiated from G syndrome by facial features and onset in late childhood (see G Syndrome.) This disorder is compatible with normal intelligence and life span. The abnormal gene may be located in the duplicated region 5p13-p12.

Ocular: Epicanthal folds; strabismus; blepharophimosis; telecanthus; widely spaced eyebrows.

Clinical: High nasal bridge; hypospadias; cryptorchidism; cleft palate and lip; urinary malformations; mental retardation; osteochondritis dissecans; congenital heart defects; upper urinary tract anomalies.

Leichtman LG, et al. Apparent Opitz BBBG syndrome with a partial duplication of 5p. *Am J Med Genet* 1991; 40:173-176.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Stoll C, et al. Male-to-male transmission of the hypertelorism-hypospadias (BBB) syndrome. *Am J Med Genet* 1985; 20:221-225.

Beal Syndrome 130

General: Transient unilateral disease; becoming bilateral later, then resolving within 2 weeks.

Ocular: Acute follicular conjunctivitis (lymphoid follicles; cobblestoning of conjunctiva with rapid onset).

Clinical: No purulent discharge; associated with regional adenitis.

Ostler HB, et al. Acute follicular conjunctivitis of epizootic origin. *Arch Ophthalmol* 1969; 82:587.

Thygeson P. Follicular conjunctivitis: infectious diseases of the conjunctiva and cornea. In: *Symposium of the New Orleans Academy of Ophthalmology.* St. Louis: CV Mosby, 1965:103.

Beard Disease (Neurasthenia; Nervous Exhaustion) 131

General: Predominantly in women who are overworked or emotionally strained; occurs usually in the fourth or fifth decade; onset gradual; episodic recurrence.

Ocular: Hippus (visible, rhythmic but irregular pupillary oscillation, deliberate in time, and 2 mm or more excursion; it has no localizing significance).

Clinical: Muscle spasms; body aches; autonomic nervous system involvement; tiredness; insomnia; impotence; dyspepsia; phobic neurosis.

Duke-Elder S, Scott GI. *System of Ophthalmology. vol. XII.* St. Louis: CV Mosby, 1971:637-638.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Bee Sting of the Eye (Bee Sting of the Cornea) 132

General: Occurs when the toothed lancet of the stinging apparatus penetrates the cornea.

Ocular: Conjunctival hemorrhage, chemosis, and hyperemia; corneal abscess; keratitis; lid edema; iris depigmentation; iridoplegia; iritis; lacrimation; apoplectic visual loss; acute disk swelling secondary to acute demyelination.

Clinical: Laryngeal edema; anaphylaxis; death; localized tissue edema; fever.

Berrios RR, Serrano LA. Bilateral optic neuritis after a bee sting. *Am J Ophthalmol* 1994; 117:677-678.

Duke-Elder S, ed. *System of Ophthalmology. vol. XIV.* St. Louis: CV Mosby, 1972:1204-1207, 1346-1348.

Smolin G, Wong I. Bee sting of the cornea: case reports. *Ann Ophthalmol* 1982; 14:342-343.

Song HS, Wray SH. Bee sting optic neuritis. A case report with visual evoked potentials. *J Clin Neuro-Ophthalmol* 1991; 11:45-49.

Behçet Syndrome (Dermato-Stomato-Ophthalmic Syndrome; Oculobuccogenital Syndrome; Gilbert Syndrome) 133

General: Virus infection?; occurs in adults; chronic disease; complete remission is rare; etiology is unknown.

Ocular: Muscle palsies (occasional); nystagmus (occasional); conjunctivitis; hypopyon; iritis; recurrent uveitis; keratoconjunctivitis sicca; keratitis; vitreous hemorrhages; thrombophlebitis retinal veins (occasional); retinal hemorrhages; optic neuritis (occasional); macular edema; optic nerve atrophy; retinitis; secondary glaucoma; retinal vasculitis; disk edema; panophthalmitis; optic neuropathy; skin lesions, posterior uveitis and systemic complications have been associated with loss of vision with this disorder; corneal immune ring opacity.

Clinical: Aphthous lesions of mucous membranes of the mouth and genitalia; cerebellar signs; convulsions; paraplegia; skin erythema (multiforme, bullosum); arthritis; urethritis; glossitis; recurrent fever.

Behcet H. Uber Rezidivierende, Aphthose, Dutchein Virus Verursachte Geschwure am Mund, am Auge und an den Genitalien. *Derm Wschr* 1937; 105:1152.

Bietti GB, et al. Behcet's disease and immunosuppressive treatment. Our personal experience. *Mod Probl Ophthalmol* 1976; 16:314-323.

Cohen S, Kremer I. Bilateral corneal immune ring opacity in Behcet's syndrome. *Arch Ophthalmol* 1991; 109:324-325.

Demiroglu H, Barista I, Dundar S. Risk factor assessment and prognosis of eye involvement in Behcet's disease in Turkey. *Ophthalmology* 1997; 104:701-705.

Durieux P, et al. Multiple pulmonary arterial aneurysms in Behcet's disease and Hughes-Stovin syndrome. *Am J Med* 1981; 71:736-741.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

James DG, et al. Behcet's disease. *Ophthalmology* 1982; 89:1279-1284.

Kargi SH, Ozdal PC, Ortac S, et al. Autonomic nervous system involvement in Behcet Disease. *Ann Ophthalmol*. 2003; 35:108-110.

Kotake S, et al. Central nervous system symptoms in patients with Behcet diseases receiving cyclosporine therapy. *Ophthalmology* 1999; 106:586-589.

Sakamoto, M, et al. Prognostic factors of vision in patients with Behcet disease. *Ophthalmology* 1995; 102:317-321.

Shimizu K. Harada's, Behcet's, Vogt-Koyanaga syndromes: are they *Clinical* entities? *Trans Am Acad Ophthalmol Otolaryngol* 1973; 77:281.

Verity DH, et al. Factor V Leiden mutation is associated with ocular involvement in Behcet disease. *Am J Ophthalmol* 1999; 128:352-356.

Behr Syndrome (Optic Atrophy Ataxia Syndrome) 134

General: Infantile form of heredofamilial optic atrophy and hereditary ataxia; autosomal recessive; rare; temporary progression that after some years leads to a static condition; both sexes equally affected, although transmission of pure hereditary optic atrophy shows marked predominance in males; in most cases, the abnormalities do not progress after childhood.

Ocular: Nystagmus; central scotoma; severe progressive temporal atrophy of the optic nerve; bilateral retrobulbar neuritis; horizontal nystagmus.

Clinical: Pyramidal tract signs (increased tendon reflexes and positive Babinski sign); ataxia and disturbance of coordination; mental deficiency; vesical sphincter muscle weakness; muscular hypertonia; clubfoot; progressive spastic paraplegia; dysarthria; head nodding.

Behr C. Die Komplizierte, Hereditarfamiliare Optikusatrophie des Kindesalters; ein Fisher Nicht Beschriebener Symptomenkomplex. *Klin Monatsbl Augenheilkd* 1909; 47: 136.

Landrigan PJ, et al. Behr's syndrome: familial optic atrophy, spastic diplegia and ataxia. *Dev Med Child Neurol* 1973; 15:41-47.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Sheffer RN, et al. Behr's syndrome and 3-methylglutaconic aciduna. *Am J Ophthalmol* 1992; 114:494-497.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol. 1.*, 4th ed. Baltimore: Williams & Wilkins, 1982.

Bell Palsy (Idiopathic Facial Paralysis) 135

General: Unilateral facial nerve paralysis of sudden onset and gradual recovery involving the nerve as it runs through the fallopian canal; etiology unknown; more common in adults.

Ocular: Corneal ulcer; paralysis of seventh nerve; ectropion; lagophthalmos; ptosis; epiphora; decreased visual acuity; diplopia; ocular irritation; exposure keratitis.

Clinical: Aching in the ear or mastoid; tingling or numbness of cheek or mouth; alteration of taste; hyperacusis; epiphora; facial weakness; most commonly and frequently affected cranial nerve with herpes zoster is the facial nerve.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Wepman B, Baum JL. Ocular findings in Bell's palsy. *Ophthalmology* 1979; 86:1943-1946.

Sartoretti-Schefer S, et al. Idiopathic, herpetic and HIV-associated facial nerve palsies: abnormal MR enhancement patterns. *AJNR* 1994; 15:479-485.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol.* 5, *part* 2. 4th ed. Baltimore: Williams & Wilkins, 1995.

Fisch U. Surgery for Bell's palsy. *Arch Otolaryngol* 1981; 107:1-11.

Benedikt Syndrome (Tegmental Syndrome) 136

General: Lesion of the inferior nucleus tuber with obstruction of the third nerve; arteriosclerotic occlusion of branches of the basilar artery, trauma and hemorrhages in the midbrain, and neoplasm most common causes.

Ocular: Homolateral paralysis of cranial nerve III (oculomotor); involves associated movements of convergence, elevation, and depression of the eyes; loss of reflex to light and accommodation.

Clinical: Unilateral hyperkinesis; contralateral hemiparesis, coarse tremor of upper extremity (greatly increased during movement), hemihypoesthesia, and absent deep sensibility; ipsilateral ataxia. There is at least one reported case of an HIV-positive patient with Benedikt syndrome who had elevated immunoglobulin G (IgG) toxoplasma IgG titers.

Antworth MV, Beck RW Third nerve palsy as a presenting sign of acquired immune deficiency syndrome. *J Clin Neuro-Ophthalmol* 1987; 7: 125-128.

Benedikt M. Tremblement Avec Paralysie Croisee du Monteur Oculaire Commune. *Bull Med (Paris)* 1889; 3:547.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 2. 4th ed. Baltimore: Williams & Wilkins, 1995:3535.

Waltman SR. Ocular signs in mid-brain disease. In: Goy AJ, Burde RM, eds. *Clinical Concepts in Neuro-Ophthalmology*. *Int Ophthalmol Clin* 1967; 7:807.

Benign Mucosal Pemphigoid (Chronic Cicatricial Conjunctivitis; Cicatricial Pemphigoid; Essential Shrinkage of the Conjunctiva; Membrane Pemphigus; Ocularpemphigoid) 137

General: Etiology unknown; involving older age group, especially over 70 years; chronic autoimmune disorder characterized by fibrosis beneath the conjunctival epithelium; associated with the major histocompatibility complex class I alleles, which confer susceptibility to the disease; likely due to a multigene effect and associated with environmental factors; incidence in women is twice as frequent as men, no geographic or racial predilection.

Ocular: Conjunctivitis; absence of goblet cells of conjunctiva; conjunctival ulcer; pannus and keratitis; corneal opacity; entropion; trichiasis; cicatrization of lacrimal ducts; corneal perforation; symblepharon; dry eyes; bilateral involvement (may be asymmetrical); ocular shrinkage; xerosis; conjunctival and corneal bullae.

Clinical: Subepidermal and subepithelial blistering of mucous membranes; blisters may occur in pharyngeal, laryngeal, nasal, anal, and genital mucosa.

Bhol K, et al. Ocular cicatricial pemphigoid. A case report of monozygotic twins discordant for the disease. *Arch Ophthalmol* 1995; 113:202-207.

Kristensen EB, Norm MS. Benign mucous membrane pemphigoid, 1. Secretion of mucus and tears. *Acta Ophthalmol* 1974; 52:266-281.

La Maza MS, et al. Cataract surgery in ocular cicatricial pemphigoid. *Ophthalmology* 1988; 95:481-486.

Mondino BJ. Cicatricial pemphigoid and erythema multiforme. *Ophthalmology* 1990; 97:939.

Mondino BJ. Clinical immunologic disease. Dermatologic disease. In: Smolin G, Thoft RA, eds. *The Cornea,* 3rd ed. Boston: Little, Brown and Company, 1994:415-417.

Thomas CI, et al. Treatment of shrinkage diseases of the conjunctiva. *Ann Ophthalmol* 1974; 6:1289-1294.

Vandeveer MR. Ocular cicatricial pemphigoid. *J Am Optom Assoc* 1991; 62:166-169.

Benjamin-Allen Syndrome 138

General: Branchial arch syndrome; not hereditary.

Ocular: Bilateral dermoids of conjunctiva; marked follicular hyperplasia of conjunctiva.

Clinical: Lymphadenopathy; cutaneous nevoid lesions; incomplete alopecia; mental retardation; growth retardation.

Benjamin SN, Allen HF Classification of limbal dermoid choristomas and branchial arch anomalies: presentation of an unusual case. *Arch Ophthalmol* 1972; 87:305-3l4.

Mattes J, et al. Ring dermoid syndrome. *Arch Ophthalmol* 1980; 98: 1059.

Benson Disease (Asteroid Bodies of the Vitreous; Asteroid Hyalitis; Snowball Opacities of the Vitreous; Scintillatio Albescens) 139

General: Etiology unknown; occurs in people of advanced age who have been asymptomatic.

Ocular: Small, solid, stellate, spherical bodies in an otherwise normal vitreous; creamy, flat white, or shiny when viewed with an ophthalmoscope; may interfere with accurate measurement of axial length.

Clinical: Increased prevalence of diabetes mellitus, hypertension, atherosclerosis, and hyperopia.

Allison KL, et al. Asteroid hyalosis and axial length measurement using automated biometry. *J Cataract Refract Surg* 1991; 17:181-186.

Benson AH. A case of "monocular asteroid hyalitis." *Trans Ophthalmol Soc U K* 1894; 14:101-104.

Bergren RL, et al. Prevalence and association of asteroid hyalosis with systemic diseases. *Am J Ophthalmol* 1991; 111:289-293.

Gartner J. Whipple disease of the central nervous system, associated with ophthalmoplegia externa and severe asteroid hyalitis. A clinicopathologic study. *Doc Ophthalmol* 1980; 49:155-l87.

Berardinelli-Seip Syndrome (Congenital Generalized Lipodystrophy) 140

General: Autosomal recessive; disorder of the hypothalamus.

Ocular: Punctate corneal infiltrations (lipodystrophia corneae).

Clinical: Advanced bone age; dilation of the third ventricle and basal cistern; frequent elevation of growth hormone; severe lipid levels; enlarged liver; diabetes mellitus; hyperpigmentation of axillae and chest wall; phlebomegaly.

Berardinelli W. Undiagnosed endocrinometabolic syndrome: report of 2 cases. *J Clin Endocrinol* 1954; 14:195.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Klein S, et al. Generalized lipodystrophy: in vivo evidence of hypermetabolism and insulin-resistant lipid, glucose and amino acid kinetics. *Metabolism* 1992; 41:893-896.

Seip M, Trygstad O. Generalized lipodystrophy. *Arch Dis Child* 1962; 38:447.

Best Disease (Best Macular Degeneration; Vitelliruptive Macular Dystrophy; Polymorphic Macular Degeneration of Braley; Vitelliform Dystrophy) 141

General: Up to 7 years of age; a type of heredomacular dystrophy; autosomal dominant with variable expressivity.

Ocular: Egg yolk lesion at macula, later absorbed to leave atrophic scar; hemorrhagic or serous exudates beneath pigment epithelium; hyperopia; esotropia; strabismic amblyopia; unusual associations with full-thickness macular hole and extramacular multifocal vitelliform disease have been reported.

Best F. Uber eine Hereditase Maculaffektion; Beitrage zur Vererbungslehie. Z *Augenheilkd* 1905; 13: 199.

Galianos SO, et al. Multifocal Best's disease and sickle cell trait. *Ann Ophthalmol* 1981; 13:1181-1183.

Glacet-Bernard A, Coscas G. Full-thickness macular hole and retinal detachment complicating Best's disease. *Eur J Ophthalmol* 1993; 3:53-54.

Kingham JO, Lochen GP. Vitelliform macular degeneration. *Am J Ophthalmol* 1977; 84:526-531.

Merin S. Best's vitelliform macular dystrophy. In: Merin S, ed. *Inherited Eye Diseases: Diagnosis and Clinical Management.* New York: Marcel Dekker, 1991:151-159.

Miller SA, et al. Choroidal neovascular membrane in Best's vitelliform macular dystrophy. *Am J Ophthalmol* 1976; 82:252.

Miller SA. Multifocal Best's vitelliform dystrophy. *Arch Ophthalmol* 1977; 95:984-990.

Mohler CW, Fine SL. Long-term evaluation of patients with Best's vitelliform dystrophy. *Ophthalmology* 1981; 88: 692-699.

Pece A, et al. Best's multiple vitelliform degeneration. *Int Ophthalmol* 1992; 16:459-464.

Seddon JM, Sharma S, Chong S, et al. Phenotype and genotype correlations in two Best families. *Ophthalmology* 2003; 110: 1724-1731.

Beta-Glucuronidase Deficiency (Mucopolysaccharidosis VII; MPS VII) 142

General: Autosomal recessive disorder associated with enzyme deficiency of β-glucuronidase; disorder combines clinical and biochemical features of the Morquio and Sanfilippo syndromes.

Ocular: Clouding of the cornea.

Clinical: Dwarfism; hepatosplenomegaly; skeletal deformity; mental retardation; hernias; unusual facies; delayed psychomotor development; frequent symptomatic pulmonary infections.

Beaudet AL, et al. Beta-glucuronidase deficiency (mucopolysaccharidosis type VII). *Birth Defects* 1974; 10:246-250.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Sly WS, et al. P-glucuronidase deficiency: mucopolysaccharidosis. *Birth Defects* 1974; 10:239-245.

Bieber Syndrome 143

General: X-linked recessive inheritance.

Ocular: Microphthalmos; corneal pannus; cataracts; uveal hypoplasia; retinal dysplasia; optic nerve hypoplasia; congenital blepharoptosis.

Clinical: Microencephaly; mental retardation; agenesis of corpus callosum; hypospadias; cryptorchidism.

Bieber FR, et al. Prenatal detection of a new form of X-linked microencephaly. *Am J Hum Genet* 1983; 35:81A.

Duker JS, et al. Ocular findings in a new heritable syndrome of brain, eye, and urogenital abnormalities. *Am J Ophthalmol* 1985; 99:51-55.

Howard RO. Classification of chromosomal eye syndrome. *Int Ophthalmol Clin* 1981; 4:77.

Bielschowsky-Lutz-Cogan Syndrome (Internuclear Ophthalmoplegia) 144

General: Lesion in the medial longitudinal fasciculus; anterior internuclear ophthalmoplegia consists of paresis of convergence with paresis of homolateral medial rectus muscle during lateral gaze toward opposite side of the lesion; in posterior internuclear ophthalmoplegia, convergence is not affected, while the homolateral medial rectus muscle is paralytic on lateral gaze; the most common causes in young patients include a demyelinating process such as multiple sclerosis, whereas an ischemic process is more common in the elderly; other reported causes of brainstem infarction associated with internuclear ophthalmoplegia include sickle cell trait, periarteritis nodosa, Wernicke encephalopathy, "crack" cocaine smoking.

Ocular: Unilateral or bilateral palsy of the medial rectus muscle during conjugate lateral gaze but with or without normal function of this muscle during convergence, depending on the type of internuclear ophthalmoplegia; dissociated nystagmus in the maximal abducted contralateral eye.

Bielschowsky A. Die Innervation der Musculi Recti Internials Seitenwender. *Ber Detsch Ophth Gesell* 1902; 30:164.

Cogan DG, et al. Unilateral internuclear ophthalmoplegia: report of eight Clinical cases with one postmortem study. *Arch Ophthalmol* 1950; 44:783.

Crane TB, et al. Analysis of characteristic eye movement abnormalities in internuclear ophthalmoplegia. *Arch Ophthalmol* 1983; 101:206-210.

De La Paz MA, et al. Bilateral internuclear ophthalmoplegia in a patient with Wernicke's encephalopathy. *J Clin Neuro-Ophthalmol* 1992; 12:116-120.

Diaz-Calderon E, et al. Bilateral internuclear ophthalmoplegia after smoking "crack" cocaine. *J Clin Neuro-Ophthalmol* 1991; 11:297-299.

Leavitt JA, Butrus SI. Internuclear ophthalmoplegia in sickle cell trait. *J Neuro-Ophthalmol* 1994; 14:49-51.

Lutz A. Ueber die Balmen der Blickwendung und deren Dissoziierung. *Klin Monatsbl Augenheilkd* 1923; 70:213.

Metz HS. Saccade velocity measurements in internuclear ophthalmoplegia. *Am J Ophthalmol* 1976; 81:296-299.

Rich JR, et al. Bilateral internuclear ophthalmoplegia after trauma. *Arch Ophthalmol* 1974; 92:69.

Biemond Syndrome 145

General: Simple recessive; hypophyseal infantilism.

Ocular: Night blindness in the presence of retinal pigment degeneration; iris coloboma (occasionally); retinal pigmentary degeneration.

Clinical: Mental retardation; polydactyly; genital dystrophia (genital organs may have been arrested in their development; absence of secondary sex characteristics); obesity; hypogenitalism; postaxial polydactyly; hydrocephalus; hypospadias.

Biemond A. Infantilisme Hypophysaire Avec Colobome Irien, Polydactylie et Anomalies Physiques et Sequelettiques. *Ned Tijdschr Geneeskd* 1934; 78:1801.

Duke-Elder S, ed. *System of Ophthalmology, vol. III, part* 2. St. Louis: CV Mosby, 1964.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Bietti Disease (Bietti Marginal Crystalline Dystrophy) 146

General: Autosomal recessive; metabolic disturbance; histopathologic studies demonstrated advanced panchorioretinal atrophy with crystals and complex lipid inclusions seen in choroidal fibroblasts.

Ocular: Marginal corneal crystalline dystrophy with retinitis punctata albescens; panchorioretinal atrophy.

Clinical: Asymptomatic.

Bernauer W, Daicker B. Bietti's corneal-retinal dystrophy, a 16-year progression. *Retina* 1992; 12:18-20.

Kaiser-Kupfer MI, et al. Clinical biochemical and pathologic correlations in Bietti's crystalline dystrophy. *Am J Ophthalmol* 1994; 118:569-582.

Mauldin WM, O'Connor PS. Crystalline retinopathy (Bietti tapetoretinal degeneration without marginal corneal dystrophy). *Am J Ophthalmol* 1981; 92:640-646.

Welch RB. Bietti's tapetoretinal degeneration with marginal corneal dystrophy crystalline retinopathy. *Trans Am Ophthalmol Soc* 1977; 75:164-179.

Bing-Neel Syndrome 147

General: Association of macroglobulinemia and central nervous system symptoms; excessive production of gamma M globulin; over 50 years of age; anoxia secondary to blood sludging from increased viscosity is explanation for peripheral retinal vascular changes.

Ocular: Ptosis; paralysis of extraocular muscles; glaucoma; chorioretinitis; dilated and segmented retinal veins; vascular tortuosity; retinal hemorrhages; peripheral microaneurysms; mild papilledema.

Clinical: Chronic encephalopathy; peripheral neuropathy; strokes; subarachnoidal hemorrhages; weakness; fatigability; weight loss; splenomegaly; anemia.

Bing J, Neel AV. Two cases of hyperglobulinemia with affection of central nervous system on a toxi-infectious basis. *Acta Med Scand* 1936; 88:492.

Frase DM, et al. Mononeuritis multiplex in a patient with macroglobulinemia. *J Neurol Neurosurg Psychiatry* 1976; 39:711-715.

Imai F, et al. Intracerebral infiltration by monoclonal plasmacytoid cells in Waldenstrom's macroglobulinemia. Case report. *Neurol Med Chir (Tokyo)* 1995; 35:575-579.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol.* 2, 4th ed. Baltimore: Williams & Wilkins, 1987.

Bipolaris 148

General: Dematiaceous septate fungus found in dust, soil, and decaying matter; associated with acquired immunodeficiency syndrome (AIDS).

Ocular: Gradual progressive visual loss; afferent pupillary defect; proptosis; corneal ulcers; optic disk pallor; orbital cellulitis; ophthalmoplegia; endophthalmitis.

Clinical: Pansinusitis; sinusitis; headaches; nosebleeds; mucoid rhinorrhea; allergic rhinitis; nasal stuffiness; allergic fungal sinusitis.

Beasley FJ. Ocular involvement in mycotic sinusitis caused by Bipolaris. *Am J Ophthalmol* 1988; 106:240-241.

Jacobson M, et al. Bipolaris-induced orbital cellulitis. *J Clin Neuro-Ophthalmol* 1992; 12:250-256.

Jay WM, et al. Ocular involvement in mycotic sinusitis caused by Bipolaris. *Am J Ophthalmol* 1988; 105:366-370.

Pavan PR, Margo CE. Endogenous endophthalmitis caused by Bipolaris hawaiiensis in a patient with acquired immunodeficiency syndrome. *Am J Ophthalmol* 1993; 116:644-645.

B-K Mole Syndrome (Familial Atypical Multiple Mole Melanoma Syndrome; DNS; Dysplastic Nevus Syndrome) 149

General: Autosomal dominant.

Ocular: Ocular melanoma; metastasis to the anterior segment of the eye; occurs much less often than metastases to the choroid; iris nevi; choroidal nevi; conjunctival nevi.

Clinical: Many large, irregular, variable nevi predominantly occurring on the upper part of the trunk and extremities; atypical melanocytic hyperplasia; lymphocyte infiltration of the dermis and neovascularization; cutaneous melanoma (possible association).

Abramson DH, et al. B-K mole syndrome. *Arch Ophthalmol* 1980; 98:1397-1399.

Lynch HT, Krush AJ. Hereditary and malignant melanoma. *Cancer* 1968; 21: 119-125.

McCarthy JM, et al. Conjunctival and uveal melanoma in the dysplastic nevus syndrome. *Surv Ophthalmol* 1993; 37: 377-386.

Rodriguez-Sains RS. Ocular findings in patients with dysplastic nevus syndrome. *Ophthalmology* 1986; 93:661-665.

Rodriguez-Sains RS. Ocular findings in patients with dysplastic nevus syndrome. An update. *Dermatol Clin* 1991; 9:723-728.

Blackwater Fever 150

General: Usually occurs in association with malaria, *Plasmodium falciparum* infection; mortality 20% to 30%; recurrent hemolytic episodes with subsequent malarial infections.

Ocular: Scleral icterus; cotton-wool spots; retinal edema; optic disk edema; conjunctival calcium deposits; band keratopathy; cortical blindness; epibulbar hemorrhage of conjunctiva, and episclera; retinal hemorrhages.

Clinical: Fever; hemolysis; icterus; hemoglobinuria; malaria; uremia; nausea; vomiting; vertigo; convulsions; coma; acute renal failure; hypertension, azotemia; hypervolemia; metabolic disturbances; hyponatremia; hypercalcemia.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*, 11th ed. New York: McGraw-Hill, 1987.

Bruce-Chivatt LJ. Quinine and the mystery of blackwater fever. *Acta Leiden* 1987; 55:181-196.

Hidayat AA, et al. The diagnostic histopathologic features of ocular malaria. *Ophthalmology* 1993; 100:1183-1186.

Newton CR, et al. Retinal haemorrhages in falciparum malaria. *Arch Dis Child* 1991; 66:753.

Blastomycosis 151

General: Chronic fungal disease caused by *Blastomyces dermatitidis.*

Ocular: Hypopyon; mycotic keratitis; corneal ulcer; choroidal granuloma; nodules of iris; cicatrization of eyelid; ectropion; descemetocele; panophthalmitis; recurrent papillomatous lesion upper lid; granulomatous conjunctivitis.

Clinical: Granulomatous lesions of skin, lung, bone, or any part of the body.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Lopez R, et al. lntraocular blastomycosis: case report and review. *Clin Infect Dis* 1994; 18:805-807.

Slack JW, et al. Blastomycosis of the eyelid and conjunctiva. *Ophthal Plast Reconstr Surg* 1992; 8:143-149.

Vida L, Noel SA. Systemic North American blastomycosis with orbital involvement. *Am J Ophthalmol* 1974; 77: 240-242.

Witorsch P, Utz JP. North American blastomycosis: a study of 40 patients. *Medicine* 1968; 47: 169-200.

Blatt Syndrome (Cranio-Orbito-Ocular Dysraphia) 152

General: Autosomal dominant; characterized by distichiasis and anisometropia; both sexes affected; present from birth.

Ocular: Hypertelorism; microphthalmos; distichiasis with the meibomian glands usually absent; anisometropia.

Clinical: Meningocele or meningoencephalocele; cranial deformities; malformations of facial bones.

Blatt N. Cranio-orbito-ocular dysraphia and meningocele. *Rev Otoneuroophthalmol* 1961; 33:185.

Duke-Elder S, ed. *System of Ophthalmology, vol. III, part* 2. St. Louis: CV Mosby, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Blau Syndrome 153

General: Rare, autosomal dominant Iris bombe, uveitis, secondary angle closure glaucoma

Clinical: Skin rash, camptodactyly, early-onset granulomatous arthritis, flexion deformity

Kurokawa T, Kikuchi T, Ohta K, et al. Ocular manifestations in Blau syndrome associated with CARD 15/Nod 2 mutation. *Ophthalmology* 2003; 110: 2040-2044.

Latkany P. Blau syndrome. *Ophthalmology* 2004; 111: 853-854.

Blepharo-Naso-Facial Malformation Syndrome 154

General: Autosomal dominant.

Ocular: Telecanthus; lateral displacement of lacrimal puncta; lacrimal excretory obstruction.

Clinical: Masklike face; bulky nose; weak facial muscles; torsional dystonia; mental retardation.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Pashayan H, et al A family with blepharo-naso-facial malformations. *Am J Dis Child* 1973; 125:389-396.

Blepharophimosis Syndrome 155

General: Dominantly inherited tetrad of ptosis, epicanthus inversus, telecanthus, and blepharophimosis.

Ocular: Scarred or contracted in secondary blepharophimosis because of ocular pemphigus or trachoma; ectropion; epicanthus inversus; lacrimal puncta displacement; ptosis; telecanthus; optic nerve coloboma; angle dysgenesis; optic nerve hypoplasia; amblyopia; strabismus.

Clinical: Low-set ears; low nasal bridge.

Beaconsfield M, et al Visual development in the blepharophimosis syndrome. *Br J Ophthalmol* 1991; 75:746-748.

Chismire KJ, Witkop GS. Optic nerve hypoplasia and angle dysgenesis in a patient with blepharophimosis syndrome. *Am J Ophthalmol* 1994; 117:676-677.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kohn R, Romano PE. Blepharoptosis, blepharophimosis, epicanthus inversus, and telecanthus: a syndrome with no name. *Am J Ophthalmol* 1971; 72:625-632.

Owens V; et al Hereditary blepharophimosis, ptosis, and epicanthus inversus. *J Int Coll Surg* 1960; 33:558-574.

Blepharoptosis, Myopia, Ectopia Lentis 156

General: Autosomal dominant.

Ocular: Ptosis; high-grade myopia; ectopia lentis; displacement of crystalline lens of eye; connective tissue defect of sclera, zonules, and levator aponeurosis.

Clinical: None.

Gillum WN, et al. Dominantly inherited blepharoptosis, high myopia and ectopia lentis. *Arch Ophthalmol* 1982; 100: 282-284.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Bloch-Sulzberger Syndrome (Incontinentia Pigmenti; Siemens-Bloch-Sulzberger Syndrome) 157

General: Familial disorder affecting ectoderm; manifestations at birth; female predominance; X-linked dominant phenotype; disturbance of skin pigmentation.

Ocular: Orbital mass; retrolental fibroplasia; pseudoglioma; strabismus; blue sclera; cataract; optic nerve atrophy; papillitis; nystagmus; chorioretinitis; anomalies of chamber angle; neovascularization of retina; retinal hemorrhages and edema; microphthalmia; tractional retinal detachment.

Clinical: Dental and skeletal anomalies common; neurologic abnormalities; recurrent inflammatory lesions; skin melanin pigmentation on the trunk: (marble cake); occipital lobe infarct; neonatal infarction of the macula.

Berbich A, et al. Ocular findings in a case of incontinentia pigmenti (Bloch-Sulzberger syndrome). *Ophthalmologica* 1981; 182:119-129.

Goldberg MF, Custis PH. Retinal and other manifestations of incontinentia pigmenti (Bloch-Sulzberger syndrome). *Ophthalmology* 1993; 100:1645-1654.

Goldberg MF, The blinding mechanisms of incontinentia pigmenti. *Ophthalmic Genet* 1994; 15:69-76.

Jain RB, Willetts GS. Fundus changes in incontinentia pigmenti (Bloch-Sulzberger syndrome): a case report. *Br J Ophthalmol* 1978; 62:622-626.

Mensheha MO, et al. Retinal pigment epithelium in incontinentia pigmenti. *Am J Ophthalmol* 1975; 79:571-577.

Wald KJ, et al. Retinal detachments in incontinentia pigmenti. *Arch Ophthalmol* 1993; 111:614-617.

Watzke RC, et al. Retinal vascular changes of incontinentia pigmenti. *Arch Ophthalmol* 1976; 94:743-746.

Blocked Nystagmus Syndrome (Nystagmus Blockage Syndrome; Nystagmus Compensation Syndrome) 158

General: Von Noorden documented this syndrome with electrooculographic recordings.

Ocular: Bilateral or monocular convergence where the adducted eye(s) cannot be abducted to the midline; if monocular, it may alternate; esotropia increases with prolonged fixation; head turn; nystagmus.

Adelstein F, Coppers C. Augenmuskellahmungen. In: Hamburger FA, Hollouich F, eds. *Bucherei des Augenarztes.* Stuttgart: Enke, 1966:246-278.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1988.

Von Noorden GK, et al. Compensatory mechanisms in congenital nystagmus. *Am J Ophthalmol* 1987; 104:387-397.

Wright KW. Exotropia. In: Wright KW, ed. *Pediatric Ophthalmology and Strabismus*. St. Louis: Mosby, 1995:191.

Bloom Syndrome (Bloom-Torre-Mackacek Syndrome; Levi-Type Dwarfism; Telangiectasis; Facial Dwarfism) 159

General: Autosomal recessive inheritance; male preponderance; usually low birth weight following full-term gestation; full-term, abnormally small children.

Ocular: Erythema of the lower eyelids.

Clinical: Facial rash; erythema of any part of the body; hypersensitivity to light; failure to grow; microcephaly; dolichocephaly; abnormalities of ears, extremities, digits, and nose; facial rash from sensitivity to sunlight; predisposition to neoplasia (especially leukemia) and diabetes mellitus; multiple chromosomal breaks have been observed in deoxyribonucleic acid (DNA) from these patients.

Bloom, D. Congenital telangiectatic erythema resembling lupus erythematosus in dwarfs. *Am J Dis Child* 1954; 88: 754-758.

German J. Bloom's syndrome: incidence, age of onset, and types of leukemia in the Bloom's syndrome registry. In: Bartsocas CS, Loukopoulos D, eds. *Genetics of Hematological Disorders*. Washington, DC: Hemisphere Publishers, 1992:241-258.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Blue Rubber Bleb Nevus Syndrome (Bean Syndrome) 160

General: Onset after birth; autosomal dominant.

Ocular: Subconjunctival hemangioma with overlying fibrosis; raised hemorrhagic lesion near macula suggestive of a small arteriovenous malformation.

Clinical: Vascular lesions; cutaneous lesions found anywhere on the body; profuse sweating may occur over the skin lesions with pain or tenderness on palpation; visceral lesions are common; tender bluish papules on the trunk and extremities; colonic hemangiomas.

Bean WB. Blue rubber bleb naevi of the skin and gastrointestinal tract in vascular spiders and related lesions of the skin. Springfield, IL: Charles C. Thomas, 1958:178-185.

Chang EL, Rubin PA: Bilateral multifocal hemangiomas of the orbit in the blue rubber bleb nevus syndrome. *Ophthalmology* 2002; 109: 537-541.

Crompton JL, Taylor D. Ocular lesion in the blue rubber naevus syndrome. *Br J Ophthalmol* 1981; 65:133-137.

Moodley M, Ramdial P. Blue rubber bleb nevus syndrome: case report and review of the literature. *Pediatrics* 1993; 92:160-162.

Paules S, et al. Tender bluish papules on the trunk and extremities. Blue rubber-bleb nevus syndrome. *Arch Dermatol* 1993; 129:1505-1506.

Bobble-Head Doll Syndrome 161

General: Caused by massive dilation of the third ventricle; occurs in childhood.

Ocular: Pallor of the optic disk or optic atrophy; visual loss.

Clinical: Flexion extension movements of the head and neck on the trunk at a rate of two or three per second; pendular movements also may involve the trunk and upper limbs; hydrocephalus; obesity; head bobbing ceases during sleep; mental retardation.

Benton JW, et al. The bobble-head doll syndrome: report of a unique truncal tumor associated with third ventricular cyst and hydrocephalus in children. *Neurology* 1966; 16:725-729.

Kirkham TH. Optic atrophy in the bobble-head doll syndrome. *J Pediatr Ophthalmol* 1977; 14:299-301.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Bogorad Syndrome (Paroxysmal Lacrimation Syndrome; Crocodile Tear Syndrome) 162

General: Subsequent to facial palsy if the lesion is proximal to the geniculate ganglion; during regeneration, fibers supposed to reinnervate the sublingual and submandibular glands are partly interchanged with fibers innervating the lacrimal gland and, hence, gustatory stimulation causes lacrimation.

Ocular: Unilateral lacrimation while eating or drinking due to misdirected nerve fiber regeneration.

Clinical: Excessive salivation (occasionally); diffuse facial muscle response or facial contracture with lacrimation.

Bogorad FA. Symptoms of crocodile tears. *Vrach Delo* 1928; 11:1328.

McGovern FH. Paroxysmal lacrimation during eating following recovery from facial paralysis: syndrome of crocodile tears. *Am J Ophthalmol* 1954; 23:1388.

Bonnet-Dechaume-Blanc Syndrome (Cerebroretinal Arteriovenous Aneurysm Syndrome; Neuroretinoangiomatosis Syndrome; Wyburn-Mason Syndrome) 163

General: Dominant inheritance; unilateral or bilateral arteriovenous aneurysm of the midbrain with ipsilateral retinal angioma and skin nevi; severity and extent of symptoms depend on location of cerebral aneurysm and structures it may involve; not regarded as hereditary; incidence is equal in men and women; usually becomes symptomatic at age 30 years.

Ocular: Exophthalmos; ptosis; strabismus; nystagmus; hemianopsia due to lesion in optic tract or pulvinar; sluggish pupils; anisocoria; retinal arteriovenous aneurysm; varicosity of retinal veins; arteriovenous angiomas; papilledema; optic atrophy of fellow eye; vitreous hemorrhage; rubeosis iridis; optic neuropathy secondary to compression by vascular malformation; proptosis; partial ophthalmoplegia.

Clinical: Arteriovenous angiomas of the thalamus and mesencephalon; facial vascular and pigmented nevi, usually in the trigeminal distribution; psychic disturbances; slow and scanning speech; hydrocephalus; headache; dizziness; hemiplegia; congenital defects of bone, muscle, kidneys, and gastrointestinal tract.

Archer DB, et al. Arteriovenous communication of the retina. *Am J Ophthalmol* 1973; 75:224.

Bonnet P, et al. L'Anevrisme Cirsoide de la Retine (Anevrisme Racemeux): Ses Relations avec l'Anevrisme Cirsoide de la Face et l'Anevrisme-Cirsoide du Cerveau. *Bull Mem Soc Franc Opht* 1938; 51:521; *ibid J Med Lyon* 1937; 18:165.

Brodsky MC, et al. Bonnet-Dechaume-Blanc syndrome with large facial angioma. *Arch Ophthalmol* 1987; 105:854.

Ebert EM, Boger WP, Albert DM. Phakomatoses. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3317-3318.

Bornholm Disease (Epidemic Pleurodynia) 164

General: Associated with group B coxsackievirus; epidemic occurrence in summer and early fall; person-to-person contact; incubation 3 to 5 days; affects both sexes; prevalent in children and young adults; recurrent episodes of sudden excruciating pain in abdominal or thoracic regions, increased by movement and respiration.

Ocular: Optic neuritis.

Clinical: Malaise; sore throat; anorexia; muscle pain; abdominal pain; cutaneous hyperesthesia and paresthesia over affected area; meningitis; myocarditis; hepatitis; orchitis.

Geeraets WJ, *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Ikeda RM, et al. Pleurodynia among football players at a high school. An outbreak associated with coxsackievirus Bl. *JAMA* 1993; 270:2205-2206.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Botulism 165

General: Caused by a toxin-producing strain of *Clostridium botulinum;* occurs primarily after the ingestion of contaminated food; the organism can produce a neurotoxin, the effect of which can be life threatening.

Ocular: Absent optokinetic nystagmus, absent vertical gaze; marked limitation of horizontal gaze; ptosis; diplopia; decreased tear secretion; mydriasis; paralysis of accommodation; nystagmus; optic atrophy; optic neuritis; extraocular muscle paresis.

Clinical: Dizziness; severe respiratory impairment; gastrointestinal disturbances; dysphagia; dysarthria; postural hypotension.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Hambleton P. Clostridium botulinum toxins: a general review of involvement in disease, structure, mode of action and preparation of Clinical use. *J Neurol* 1992; 239:16.

Hedges TR, et al. Botulin ophthalmoplegia. *Arch Ophthalmol* 1983; 101:211-213.

Bourneville Syndrome (Bourneville-Pringle Syndrome; Tuberous Sclerosis; Epiloia) 166

General: Irregular dominant inheritance; more frequent in females; most patients die before age 24 years.

Ocular: Vitreous often cloudy; lens opacities; retinal mushroom-like tumor of grayish-white color; yellowish-white plaques with small hemorrhages and cystic changes in retina; papilledema; disk drusen; cerebral astrocytoma; 40% to 50% of patients have normal intelligence.

Clinical: Grand mal, petit mal, or jacksonian seizures (manifest first 2 years of life); mental changes from feeblemindedness to imbecility and idiocy; skin changes arranged usually about nose and cheeks (adenoma sebaceum); congenital tumors of kidney (hypernephroma or tubular adenoma) and heart (rhabdomyoma); cerebral astrocytoma.

Berberich MS, Hall BD. Penetrance and variability in tuberous sclerosis. *Birth Defects* 1979; 15:297-304.

Bourneville D. Contribution a l'Etude de l'Idiotie-Sclerose Tubereusedes Circonvulsions Cerebrales: Idiotie et Epilepsie Hemiplegique. *Arch Neurol (Paris)* 1880-1881; 1:69.

Ettl A, et al. Retinal phakomata associated with cerebral astrocytoma. An incomplete form of Bourneville-Pringle disease? *Ophthalmologica* 1993; 206:209-213.

Font RD, Ferry AP. The phacomatoses. *Int Ophthalmol Clin* 1972; 12: 1.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Margo CE, et al. Giant cell astrocytoma of the retina in tuberous sclerosis. *Retina* 1994; 13:155-159.

Williams R, Taylor D. Tuberous sclerosis. *Surv Ophthalmol* 1985; 30:143.

Boutonneuse Fever (Marseilles Fever) 167

General: Caused by *Rickettsia conorii* and transmitted by ticks.

Ocular: Conjunctivitis; central serous retinopathy; retinal detachment; perivasculitis; uveitis; papillitis; keratitis.

Clinical: Fever; lymph node enlargement; papular rash.

Alio J, et al. Rickettsial keratitis in a case of Mediterranean spotted fever. *Eur J Ophthalmol* 1992; 2:41-43.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Khairallah M, Ladjimi A, Chakroun M, et al. Posterior segment manifestations of *rickettsis conorii* infection. *Ophthalmology* 2004; 111: 529-534

Bowen Disease (Intraepithelial Epithelioma; Carcinoma In Situ; Dyskeratosis) 168

General: Squamous cell carcinomas *in situ* of the skin or conjunctiva.

Ocular: Dysplastic epithelium, intraepithelial epithelioma, or invasive squamous cell carcinoma of conjunctiva or cornea; infiltration of lacrimal system and sclera.

Awan KS. Intraepithelial epithelioma (Bowen's disease) of conjunctiva and chronic lymphocytic leukemia. *Ann Ophthalmol* 1978; 10:781-783.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Grossniklaus HE, et al. Conjunctival lesions in adults. A *Clinical* and histopathological review. *Cornea* 1987; 6:78.

Irvine AR Jr. Epibulbar squamous cell carcinoma and related lesions. *Int Ophthalmol Clin* 1972; 12:71-83.

Kamel S, Kamel KS. Bowen's disease: intraepithelial epithelioma. *Bull Ophthalmol Soc Egypt* 1975; 68:101-103.

Shields JA, Shields CL. Tumors of the conjunctiva and cornea. In: Smolin G, Thoft RA, eds. *The Cornea*, 3rd ed. Boston: Little, Brown and Company, 1994:587-588.

Brachymetapody-Anodontia-Hypotrichosis Albinoidism Syndrome (Anodontia-Hypotrichosis Syndrome) 169

General: Autosomal recessive.

Ocular: Strabismus; nystagmus; distichiasis; cataracts; high myopia.

Clinical: Congenital anodontia; small maxilla; short stature; shortening of metacarpals and metatarsals; little hair growth; albinoidism.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Taumaala P, Haapanen E. Three siblings with similar anomalies in the eyes, bones, and skin. *Acta Ophthalmol* 1968; 46:365-371.

Brain Dysfunction Syndrome (Dyscontrol Syndrome) 170

General: Suspected causes include tumors of the limbic system and/or localized atrophy of the brain.

Ocular: Visual field defects.

Clinical: Manic behavior; physical brutality without motive; sexual assaults; seizures; lack of memory; hallucinations; temporal lobe seizures; depression; speech difficulties.

Clements SD, Peters JE. Minimal brain dysfunctions in school-age child. *Arch Gen Psychiatry* 1962; 6:185.

Mark VH, et al. Brain disease and violent behavior. In: Smith JL, ed. *NeuroOphthalmology, vol. III.* St. Louis: CV Mosby, 1968:282.

Branched-Chain Ketoaciduria (Maple Syrup Urine Disease) 171

General: Deficiency in the oxidative decarboxylation of the corresponding α-ketoacids; possibly autosomal recessive inheritance; both sexes affected; onset in first week of life.

Ocular: Ptosis; epicanthal folds; hypertelorism; prominence of supraorbital ridges; cataract; strabismus; decreased or absent pupillary reaction to light; horizontal nystagmus; optic atrophy; ophthalmoplegia.

Clinical: Maple syrup odor of urine; neurologic symptoms; death may follow promptly or the patient may live for a decade during which severe mental retardation is apparent; vomiting; failure to thrive; absence of grasping reflex; generalized rigidity; hypoglycemic crisis; cortical blindness.

Burke JP, et al. Ophthalmic findings in maple syrup urine disease. *Metab Pediatr Syst Ophthalmol* 1991; 14:12-15.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:247.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Roy FH, Kelly ML. Maple syrup urine disease. *J Pediatr Ophthalmol* 1973; 10:70-73.

Branchial Clefts with Characteristic Facies, Growth Retardation, Imperforate Nasolacrimal Duct, and Premature Aging 172

General: Autosomal dominant.

Ocular: Strabismus; obstructed nasolacrimal ducts.

Clinical: Low birth weight; retarded growth; bilateral bronchial cleft sinuses; broad nasal bridge; protruding upper lip; carp mouth; premature aging; malformed ears; linear skin lesions behind the ears.

Fujimoto A, et al. New autosomal dominant branchiooculofacial syndrome. *Am J Med Genet* 1987; 27:943-951.

Lee WK., et al. Bilateral branchial cleft sinuses associated with intrauterine and postnatal growth retardation, premature aging, and unusual facial appearance: a new syndrome with dominant transmission. *Am J Med Genet* 1982; 11: 345-352.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Brill-Symmers Disease (Lymphosarcoma; Giant Follicular Lymphoma) 173

General: Occurs primarily in older patients.

Ocular: Unilateral or bilateral lid swelling; orbital masses with exophthalmos; lacrimal gland infiltration.

Clinical: Patients survive 6 to 7 years; special form of lymphoma.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1988.

Brittle Cornea Syndrome (Brittle Cornea, Blue Sclera and Red Hair Syndrome; Blue Sclera Syndrome) 174

General: Autosomal recessive; rare.

Ocular: Spontaneous perforation of cornea (brittle cornea); blue sclera; acute hydrops; microcornea; sclerocornea; cornea plana; keratoconus; keratoglobus.

Clinical: Red hair; associated with Ehlers-Danlos syndrome, osteogenesis imperfecta, and Marfan syndrome.

Cameron JA, et al. Epikeratoplasty for keratoglobus associated with blue sclera. *Ophthalmology* 1991; 98:446-452.

Cameron JA. Corneal abnormalities in Ehlers-Danlos syndrome type VI. *Cornea* 1993; 12:54-59.

Mehrotra AN, Bhatia RP. Syndrome of blue sclera, fragilitas ossium and otosclerosis. *Ind J Ophthalmol* 1974; 22: 42-44.

Osama O, et al. Brittle cornea syndrome. *Presented at the American Academy of Ophthalmology, Atlanta, Georgia*, 1995: 128(abst).

Paul AK, et al. Blue sclera with fragilitas ossium. *Ind J Ophthalmol* 1974; 22:38-39.

Stein E, et al. Brittle cornea. *Am J Ophthalmol* 1968; 66:67-69.

Ticho U, et al. Brittle cornea, blue sclera and red hair syndrome. *Br J Ophthalmol* 1980; 64:175-177.

Brown Syndrome (Superior Oblique Tendon Sheath Syndrome) 175

General: Etiology unknown; affects both sexes; present from birth; may be congenital or acquired (secondary to trauma, orbital surgery, or injections, or following delivery).

Ocular: Bilateral ptosis with associated backward head tilt; widening of palpebral fissure with attempted upward gaze; ocular movements show failure in direction of superior oblique action; may be associated with underaction of the inferior oblique; adduction or abduction restricted or completely abolished; choroidal coloboma.

Bert AT, et al. Congenital Brown's syndrome: *Clinical* and surgical approach. *Eur J Ophthalmol* 1994; 4: 138-143.

Brown HW. Congenital structural motor anomalies in strabismus. In: Allen JH, ed. *Ophthalmic symposium.* St. Louis: CV Mosby, 1950:205,229.

Brown HW. True and simulated superior oblique tendon sheath syndrome. *Doc Ophthalmol* 1973; 34:123-126.

Christiansen SP, Thomas AH. Postpartum Brown's syndrome. *Arch Ophthalmol* 1994; 112:23-25.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Nelson LB. Strabismus disorders (Brown syndrome). In: Nelson, Calhoun, Harley, eds. *Pediatric Ophthalmology*, 3rd ed. Philadelphia: WB Saunders, 1991:165.

Wang FM, et al. Acquired Brown's syndrome in children with juvenile rheumatoid arthritis. *Ophthalmology* 1984; 91:23-26.

Brown-Marie Syndrome (Brown-Marie Ataxic Syndrome; Sanger Brown Syndrome; Hereditary Ataxia Syndrome; Marie Hereditary Ataxia) 176

General: Cause unknown; simple recessive inheritance, although irregular dominant transmission has been observed.

Ocular: Nystagmus; strabismus; ophthalmoplegia; anisocoria; Argyll Robertson pupil; retinitis pigmentosa; optic nerve atrophy; retrobulbar optic neuritis.

Clinical: Hereditary ataxia; choreiform movements; athetosis; pyramidal tract paresis; speech difficulties; hyperreflexia.

Brown S. On hereditary ataxia, with a series of twenty-one cases. *Brain* 1892; 15:250.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Havener WH. Cerebellar-macular abiotrophy. *Arch Ophthalmol* 1951; 45:40.

Brown-McLean Syndrome 177

General: Following cataract surgery Photophobia, posterior staphyloma, peripheral corneal edema with underlying endothelial pigment, iridonesis

Clinical: Foreign body sensation

Hara T, Hara T: Brown-McLean syndrome associated with corneal endotheliitisin a pseudophakic eye. *J Cataract Refract Surg* 1993; 19: 780-786.

Martin EN, Alvarenza LS, SousLB, et al. Anterior stromal puncture in Brown-McLean syndrome. *J Cataract Refract Surg* 2004; 30: 1575-1577.

Reed JW, Cain LR, Weaver RG, et al. Clinical and pathologic findings of aphakic peripheral corneal edema: Brown-McLean syndrome. *Cornea* 1992; 11: 577-583

Brown-Sequard Syndrome 178

General: Caused by lesion (injury, tumor pressure) of spinal cord.

Ocular: Nystagmus (if lesion in upper cervical area); sluggish pupillary reaction to light (occasional finding); optic atrophy.

Clinical: Homolateral spastic paralysis with (i) loss of ipsilateral deep joint, tendon, and vibratory sensations below level of the lesion; (ii) loss of contralateral pain and temperature sensations; and (iii) sphincteral disturbances. Cases of this syndrome caused by meningomyelitis secondary to syphilis, herpes zoster, and multiple sclerosis have been reported.

Brown-Sequard CE. Sur la Transmission des Impressions Sensitives dans la Moelle Epiniere. *Compt Rend* 1850; 31: 700.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol.* 5, *part* 2. 4th ed. Baltimore: Williams & Wilkins, 1995.

Strom T, Schneck SA. Syphilitic meningomyelitis. *Neurology* 1991; 41:325-326.

Bruch Membrane Drusen 179

General: Autosomal dominant; round or oval lesions in grape like clusters in the posterior polar region; found on the vitreal side of Bruch membrane, secreted by the retinal pigment epithelial cells, apparently secondary to an inborn error of metabolism localized in the retinal pigment epithelium.

Ocular: Crystalline retinal degeneration; Doyne honeycomb choroiditis; fleck retina disease; macular edema; macular hemorrhage; pigmentary disturbances with secondary calcifications; central scotomata.

Clinical: None.

Deutman AF, Jansen LA. Dominantly inherited drusen of Bruch's membrane. *Br J Ophthalmol* 1970; 54:372-382.

Deutman AF. Macular dystrophies. In: Ryan SJ, ed. *Retina. vol. II* 2nd ed. St. Louis: Mosby, 1994.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Bruns Syndrome (postural Change Syndrome) 180

General: Caused by tumors of the third, fourth, or lateral ventricle or by lesions of the midline in the brain.

Ocular: Partial ophthalmoplegia (third nerve paralysis) and gaze paralysis; oculomotor paresis associated with postural change of head or body; amaurosis or transient blindness; flashes of light.

Clinical: Severe paroxysmal headache; nausea and vomiting; vertigo; irregular respiration; apnea; syncope; tachycardia; free-floating cysts within the fourth ventricle may produce intermittent foramen obstruction and Bruns syndrome; Kramer reported a patient with a free-floating cysticercus cyst with this condition.

Alpers BJ, Yaskin HE. The Bruns syndrome. *J Nerv Ment Dis* 1944; 100:115.

Bruns O. Neuropathologische Demonstrationen. *Neurol Centralbl* 1902; 21:561.

Geeraets WJ *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kramer J, et al. Transaqueductal migration of a neurocysticercus cyst. Case report. *J Neurosurg* 1992; 77:956-968.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5. *part* 1. 4th ed. Baltimore: Williams & Wilkins, 1995:3345-3346.

Buerger Disease (Thromboangiitis Obliterans) 181

General: Unknown etiology; males who smoke and are under age 35 years; affects small- and medium-sized arteries and veins of the extremities; segmented episodic inflammatory panarteritis with associated thrombosis.

Ocular: Exudative retinopathy; occlusion of retinal vessels; retinal hemorrhages; perivasculitis and endovasculitis; blindness; cataract.

Clinical: Intermittent claudication; coolness; paresthesia; hyperemia; cyanosis and gangrene may be present in the lower extremities.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Bullous Ichthyosiform Erythroderma (Collodion Baby; Congenital Ichthyosis; Epidermolytic Hyperkeratosis; Ichthyosis; Ichthyosis Vulgaris; Lamellar Ichthyosis; Nonbullous Ichthyosiform Erythroderma; Xeroderma; X-Linked Ichthyosis) 182

General: Autosomal inherited disorder; affects both sexes; normal at birth; onset within first 7 days; X-linked; pathogenesis may be secondary to physicochemical changes of corneal tissues including accumulation of cholesterol sulfate.

Ocular: Keratopathy; corneal scarring; keratitis; conjunctivitis; lagophthalmos; photophobia; ectropion; lid erythema; lacrimation; keratoconus; deep corneal punctate/filiform lesions

Clinical: At birth, the skin surface is moist, red, and tender; within several days, thick verrucous scales form.

Casaroli Marano RP, et al. Ocular findings associated with congenital X-linked ichthyosis. *Ann Ophthalmol* 1991; 23:167-172.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Frost P. Disorders of cornification. In: Moschella SL, et al., eds. *Dermatology.* Philadelphia: WB Saunders, 1975: 1056-1084.

Macsai MS, Doshi H. Clinical pathologic correlation of superficial corneal opacities in X-linked ichthyosis. *Am J Ophthalmol* 1994; 118:477-484.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Orth DH, et al. Collodion baby with transient bilateral lid ectropion. Review of ocular manifestations in ichthyosis. *Arch Ophthalmol* 1994; 91:206-207.

Burnett Syndrome (Milk Drinker Syndrome; Milk-Alkali Syndrome) 183

General: Characterized by alkalosis, hypercalcemia, and transient renal insufficiency with azotemia; develops during milk-alkali therapy for peptic ulcer; seen in excessive intake of milk or soluble alkali, as in therapy for peptic ulcer.

Ocular: Band-shaped keratopathy; conjunctivitis with calcification.

Clinical: Nausea, vomiting; headache; irritability; dizziness; depression; confusion.

Burnett CH, et al. Hypercalcemia without hypercalciuria or hypophosphatemia, calcinosis and renal insufficiency; syndrome following prolonged intake of milk and alkali. *N Engl J Med* 1949; 240:787.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Newmark K, Nugent P. Milk-alkali syndrome. A consequence of chronic antacid abuse. *Postgrad Med* 1993; 93: 149-150.

Schneider AB, Sherwood LM. Calcium homeostasis and the pathogenesis and management of hypercalcemic disorders. *Metabolism* 1974; 23:975

C

C Syndrome (Opitz Trigonocephaly Syndrome; Trigonocephaly Syndrome) 184

General: Autosomal recessive; consanguinity; early death.

Ocular: Hypertelorism; up-slanted palpebral fissures; strabismus.

Clinical: Polydactyly; unusual facies; cardiac abnormality; cryptorchidism; Omtra-oral anomalies; abnormally modeled ears; cardiac anomalies; neonatal hypotonia; severe mental retardation; short neck with loose skin.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Opitz JM, et al. The C syndrome of multiple congenital anomalies. *Birth Defects* 1969; 12:161-166.

Schaap C, et al. Opitz-C syndrome: on the nosology of mental retardation and trigonocephaly. *Genet Couns* 1992; 3: 209-215.

Zanini SA, et al. Trigonocephaly. *J Craniofacial Surg* 1992; 3:85-89.

Caffey Syndrome (Caffey-Silverman Syndrome; Infantile Cortical Hyperostosis) 185

General: Cause unknown, possibly collagen disease or viral infection; prevalent in females; onset in early infancy; sudden onset; benign; self-limited.

Ocular: Periorbital edema and tenderness; transient proptosis; mild conjunctivitis.

Clinical: Tender swelling over regions of cortical hyperostosis (may resemble periostitis); severe anemia; fever; dysphagia; pleurisy.

Borochowitz Z, et al. Familial Caffey's disease and late recurrence in a child. *Clin Genet* 1991; 40:329-335.

Caffey J, Silvermann WA. Infantile cortical hyperostoses. Preliminary report of a new syndrome. *Am J Roentgenol* 1945; 54:1.

Keeton BR. Vitamin E deficiency and thrombocytosis in Caffey's disease. *Arch Dis Child* 1976; 51:393-395.

Minton LR, Elliott JR. Ocular manifestations of infantile cortical hyperostosis. *Am J Ophthalmol* 1967; 64:902.

Caisson Syndrome (Bends Syndrome; Diver's Palsy; Compressed-Air Illness) 186

General: Under high atmospheric pressure (18 lb/in^2 at least) the blood becomes saturated with nitrogen; sudden decompression (e.g., return to normal atmospheric pressure when divers surface too rapidly) causes the nitrogen to bubble out in gas form, with resulting destruction of tissue spaces; symptoms appear usually within 3 hours after decompression.

Ocular: Nystagmus; diplopia; transient blindness; cataract formation with rapidly developing vacuoles and gray opacities; narrowing of the retinal vessels.

Clinical: Severe joint pain; dyspnea with sensation of chest constriction; giddiness; hemiplegia; vertigo; deafness; aphasia; paraplegia; convulsion; pruritus; abdominal pain.

Collier J. Caisson disease. In: Christian HA, ed. *Oxford Loose Leaf Medicine, vol. VI.* New York: Oxford University Press, 1920-1942:438-446.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology,* 4th ed. Baltimore: Williams & Wilkins, 1987.

California Syndrome 187

General: Functional and/or malingering visual complaints in people with psychosocial problems relating to parental divorce, poor school performance, and attention-getting behavior; seen in adults as well as children; usually secondary gain motive (e.g., disability benefits, Workmen's Compensation, litigation, or income tax).

Ocular: Blurred vision; abnormal vision fields; abnormal color vision; voluntary nystagmus; decreased visual acuity at near or far distance.

Clinical: Hysteria; malingering; social stresses.

Keltner JL, et al. The California syndrome. *Ophthalmology* 1985; 92:427-435.

Keltner JL. The California syndrome. *Arch Ophthalmol* 1988; 106:1053-1054.

CAMAK Syndrome (Cataract, Microcephaly, Arthrogryposis, Kyphosis Syndrome) 188

General: Low birth weight; autosomal recessive.

Ocular: Cataracts at birth or within 3 weeks.

Clinical: Mental retardation, stiffness of joints, microcephaly; birdlike facies; progressive curvature of the spine.

Lowry RB, et al. Cataracts, microcephaly, kyphosis, and limited joint movement in two siblings: a new syndrome. *J Med Genet* 1971; 15:227-229.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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CAMFAK Syndrome (Cataract, Microcephaly, Failure to Thrive, Kyphoscoliosis Syndrome) 189

General: Autosomal recessive; there is evidence supporting that it is a neurologic disease characterized by peripheral and central demyelination similar to that seen in Cockayne syndrome.

Ocular: Cataracts.

Clinical: Microcephaly; failure to thrive; mental retardation; spasticity; hip dislocation; kyphoscoliosis.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Scott-Emuakpor AB, et al. A syndrome of microcephaly and cataracts in four siblings: a new genetic syndrome. *Am J Dis Child* 1977; 131:167-169.

Talwar D, Smith SA. CAMFAK syndrome: a demyelinating inherited disease similar to Cockayne's syndrome. *Am J Med Genet* 1989; 34:194-98.

Canalis Opticus Syndrome 190

General: Loss of vision after blunt trauma to the head (mainly forehead) without direct eye injury; thought to occur because of sudden stretching of the fixed, as well as the movable, portions of the optic nerve during movement of the brain at the time of injury.

Ocular: Spontaneous unilateral or bilateral, reversible or irreversible amaurosis; absent pupil reaction in cases of complete blindness; spontaneous visual recovery has been reported anecdotally.

Clinical: Blunt head injury.

Berlin R. Uber Sehstorungen nach Verletzung durch Stumpfe Gewalt. *Klin Monatsbl Augenheilkd* 1878; 17.

Miller NR. The management of traumatic optic neuropathy. *Arch Ophthalmol* 1990; 108: 1086.

Seitz R. Canalis-opticus syndrome. *Ophthalmologica* 1969; 158:318.

Snebold NG. Neuro-ophthalmic manifestations of trauma. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994.

Wolin MJ, Lavin PJM. Spontaneous visual recovery from traumatic optic neuropathy after blunt head injury. *Am J Ophthalmol* 1990; 109:430.

Candidiasis 191

General: Yeastlike opportunistic fungal infection caused by *Candida albicans.*

Ocular: Uveitis; hypopyon; conjunctivitis; keratitis; corneal ulcer; blepharitis; endophthalmitis; dacryocystitis; papillitis; retinal atrophy; Roth spot; vitreous abscess; retrobulbar abscess; retinal detachment; panophthalmitis; chorioretinitis; infectious crystalline keratopathy.

Clinical: *C. albicans* normally is present as an intestinal saprophyte in 35% to 75% of the human population; in situations of internal environmental change, however, *Candida* can become pathogenic (e.g., obesity, diabetes mellitus, malignancy, and other debilitating conditions).

Donahue SS, et al. Intraocular candidiasis in patients with candidemia. Clinical implications derived from a prospective multicenter study. *Ophthalmology* 1994; 101:1302-1309.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Purgason PA, et al. Atypical presentation of fungal dacryocystitis. A report of two cases. *Ophthalmology* 1992; 99: 1430-1432.

Speer ME, et al. Candida endophthalmitis: a manifestation of candidiasis in the neonate. *South Med J* 1980; 73: 1407-1409.

Wagman RD, et al. Keratitis associated with the multiple endocrine deficiency, autoimmune disease, and candidiasis syndrome. *Am J Ophthalmol* 1987; 103:569-575.

Wilhelmus KR, Robinson NM. Infectious crystalline keratopathy caused by Candida albicans. *Am J Ophthalmol* 1991; 112:322-325.

Canine Tooth Syndrome (Class VII Superior Oblique Palsy) 192

General: Caused by trauma to the trochlear area, producing a "double Brown syndrome"; secondary to strengthening the superior oblique along with a residual superior oblique palsy, or a combination of local trauma to the trochlea causing restriction to upgaze along with closed head trauma producing a fourth nerve palsy.

Ocular: Underaction of the superior oblique and underaction of the inferior oblique on the same side.

Ellis FH, Helveston EM. Superior oblique palsy: diagnosis and classification. *Int Ophthalmol Clin* 1976; 16: 127-135.

Fraunfelder FT, Roy FR. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Capgras Syndrome (Illusion of Double Syndrome; L'illusion des Sosies; Phantom Double Syndrome; Nonrecognition-Misidentification Syndrome) 193

General: Characterized by misidentification or nonrecognition of a person by the patient who believes that this person appears in double in front of him or her; occurs in paranoid psychosis, and only people familiar or important to the patient appear in double; preponderance in women; agnosia of identification.

Ocular: Illusion of double perception with failure of recognition of a known person.

Clinical: General claims include statements that the person seen is an impostor, although the person is well known to the patient; hallucinations; delusions.

Anderson DN, Williams E. The delusion of inanimate doubles. *Psychopathology* 1994; 27:220-225.

Capgras J, Carrette P. Illusion des Sosies et Complexe d'Oedipe. *Ann Med Psychol* l924;82:48.

Luaute JP, Bidault E. Capgras syndrome: agnosia of Identification and delusion of reduplication. *Psychopathology* 1994; 27: 186-193.

Vogel BF. The Capgras syndrome and its psychopathology. *Am J Psychiatry* 1974; 131:922

Capsular Bag Distension Syndrome 194

General: Occurs after phacoemulsification as a result of occlusion of the anterior capsularhexis opening by the intraocular lens optic

Ocular: Shallow anterior chamber, pupil peaking, accumulation of turbid fluid in the capsular bag

Agrawal S, Agrawal J, Agrawal TP. Incomplet capsular bag distension syndrome after neodymium: YAG capsulotomy. *J Cataract Refract Surg* 2006; 32:351-352.

Davison JA. Capsular bag distention after endophacoemulsification and posterior chamber intraocular lens implantation. *J Cataract Refract Surg* 1990; 16: 99-108.

Capsular Block Syndrome 195

General: Following cataract surgery

Ocular: Complete sealing by the anterior capsule opening by the optic and displacement of the posterior capsule far behind the posterior optic surface

Liu TY, Chou PI. Capsular block syndrome associated with secondary angle-closure glaucoma. *J Cataract Refract Surg* 2001; 27: 1503-1505.

Mikae K, Ota I Iichihashi E, et. al. Capsular bag distention syndrome noted 5 years after intraocular lens implantation. Am J Ophthalmol 1998; 125: 545-547

Xiao Y, Wang U, Fu Z. Capsular block syndrome caused by a reversed-optic intraocular lens. *J Cataract Refract Surg* 2004; 30: 1130-1132

CAR Syndrome (Cancer-Associated Retinopathy Syndrome) 196

General: Rare; antiretinal antibodies in blood of cancer patients experiencing concomitant loss of vision; vision loss may be noted before cancer is diagnosed; mechanisms involved in the vision loss experienced by these patients is not understood, but serologic studies indicate they may include a series of autoimmune reactions directed at specific components of the retina.

Ocular: Vision loss usually progressive; retinal degeneration; retinal hole; abnormal visual fields; loss of color vision; retinal detachment; optic atrophy; ringlike scotoma; night blindness; retinal phlebitis.

Clinical: Carcinoma with or without metastasis to any part of the body.

Keltner JL, et al. Photoreceptor degeneration: possible autoimmune disorder. *Arch Ophthalmol* 1983; 101:564-569.

Ohnishi Y, et al. Cancer-associated retinopathy with retinal phlebitis. *Br J Ophthalmol* 1993; 77:795-798.

Thirkill CE, et al. Cancer-associated retinopathy. *Arch Ophthalmol* 1987; 105:372-375.

Thirkill CE, et al. The cancer-associated retinopathy antigen is a recovering-like protein. *Invest Ophthalmol Vis Sci* 1992; 33:2768-2772.

Carcinoid Syndrome 197

General: Slow-growing neoplasms of enterochromaffin cell; metastatic tumors usually arise from small primary tumors in the ileum.

Ocular: Lacrimation; periorbital edema; choroidal and orbital metastases.

Clinical: Cutaneous flushing; telangiectasia; intestinal hypermotility.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Fan JT, et al. Clinical features and treatment of seven patients with carcinoid tumor metastatic to the eye and orbit. *Am J Ophthalmol* 1995; 119:211-218.

Cardiac Myxomas 198

General: Myxomas of the heart account for approximately 50% of primary heart tumors; although benign, they can cause serious complications and death by obstruction or embolism; tumor arises from the mural endocardium.

Ocular: Central or branch retinal artery obstruction with ganglion cell edema and a "cherry-red macula"; choroidal and retinal infarct; ischemic optic neuropathy; conjunctival and caruncle pigmentation; eyelid pigmentation; transient loss of vision.

Clinical: Signs and symptoms are a result of emboli that travel to the extremities, brain, liver, spleen, kidney, cerebrum, and, rarely, the coronary arteries.

Bolo-Deoku J Jr, et al. Transient loss of peripheral vision as the presentation of left atrial myxoma. *Br J Ophthalmol* 1992; 76:113-114.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:221.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Kennedy RH, et al. The Carney complex with ocular signs suggestive of cardiac myxoma. *Am J Ophthalmol* 1991; 111:699-702.

Taylor RH, Deutsch J. Myxoma mix-up: a case report. *J Clin Neuro-Ophthalmol* 1992; 12:207-209.

Cardiorespiratory Obesity Syndrome (Pickwickian Syndrome) 199

General: Chronic pulmonary insufficiency caused by extensive obesity.

Ocular: Venous congestion; spontaneous hyphema; chorioretinal venous congestion with retinal hemorrhages and exudates; papilledema; rubeosis iridis (may be bilateral).

Clinical: Pronounced and greatly excessive obesity; headache; dyspnea; drowsiness; cyanosis; heart failure; muscular twitching; disturbed consciousness.

Burwell CS, et al. Extreme obesity associated with alveolar hypoventilation: a pickwickian syndrome. *Am J Med* 1956; 21:811.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Metzl K, et al. The pickwickian syndrome in a child: an extreme example of psychoneurotic obesity. *Clin Pediatr* 1969; 8:49.

Schneider TL, et al. Bilateral rubeosis iridis in a 36 year-old male as the presenting sign of obesity-hypoventilation syndrome: implications for angiogenesis[abstract]. *Invest Ophthalmol Vis Sci. Abs Issue* 1993; 34.

Carotid Artery Syndrome (Carotid Vascular Insufficiency Syndrome; Ocular Ischemic Syndrome) 200

General: Causes include microemboli, atherosclerotic plaques, arteritis, arterial compression by cicatricial tissue surrounding the vessel, and tumors; male preponderance; onset between ages 50 and 70 years.

Ocular: Lacrimation; homolateral transient, painless visual loss; photopsia; hemianopsia; retinal infarcts; cholesterol plaques may be seen in retinal arteries on funduscopic examination; optic atrophy; hypoxic retinopathy; low-tension glaucoma; anterior uveitis; cataract; visual acuity 20/400 or less; iris neovascularization; angle neovascularization; optic disk pale; retinal hemorrhages; Homer syndrome; amaurosis fugax; retinal artery occlusion; ophthalmoparesis; proptosis; chemosis; conjunctival hyperemia; acute orbital infarction.

Clinical: Transient cerebral ischemia with contralateral weakness of arm and leg; hemisensory disturbances; mental confusion and dysphasia; headache; dizziness; epileptiform seizures; carotid dissection.

Bogousslavsky J, et al. Isolated complete orbital infarction: a common carotid artery occlusion syndrome. *Eur Neurol* 1991; 31:72-76.

Brown GC. Macular edema in association with severe carotid artery obstruction. *Am J Ophthalmol* 1986; 102: 442-448.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fisher RG, Friedmann KR. Carotid artery thrombosis in persons fifteen years of age or younger. *J Am Med Assoc* 1959; 170: 1918.

Galetta SL, et al. Orbital ischemia, ophthalmoparesis and carotid dissection. *J Clin Neuro-Ophthalmol* 1991; 11: 284-287.

Marx JL, Hreib K, Choi IS, et al. Percutaneous carotid artery angioplasty and stenting for ocular ischemic syndrome. *Ophthalmology* 2004; 111: 2284-2291.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology*, 4th ed. Baltimore: Williams & Wilkins, 1987.

Mizener JB, et al. Ocular ischemic syndrome. *Ophthalmology* 1997; 104:859-864.

Muller M, et al. Carotid artery disease in vascular ocular syndromes. *J Clin Neuro-Ophthalmol* 1993; 13:175-180.

Carotid Artery Syndrome (Cavernous Sinus Fistula Syndrome; Red-Eyed Shunt Syndrome) 201

General: Seventy-five percent of cases caused by trauma; others occur spontaneously or are congenital; fistula from carotid artery to cavernous sinus.

Ocular: Progressive, pulsating exophthalmos; distended pulsating superior orbital vein; venous congestion of lids; variable ophthalmoplegia, depending on involvement of cranial nerves III to VI; secondary glaucoma; congestion of conjunctiva with chemosis; corneal ulcerations; eversion of the lower lid; loss of corneal sensation; retinal edema; engorgement of retinal veins; papilledema; optic atrophy; ocular bruit that may be subjective and/or objective; diplopia; visual decrease; choroidal folds; dilated superior ophthalmic vein.

Clinical: Severe unilateral headache; buzzing noise.

Dailey EJ, et al. Evaluation of ocular signs and symptoms in cerebral aneurysms. *Arch Ophthalmol* 1964; 71:463.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Flaharty PM, et al. Color Doppler imaging. A new noninvasive technique to diagnose and monitor carotid cavernous sinus fistulas. *Arch Ophthalmol* 1991; 109:522-526.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gonshor LG, Kline LB. Choroidal folds and dural cavernous sinus fistula. *Arch Ophthalmol* 1991; 109:1065-1066.

Phelps CD, et al. The diagnosis and prognosis of atypical carotid cavernous fistula. *Am J Ophthalmol* 1982; 93: 423-436.

Travers B. A case of aneurysm by anastomosis in the orbit, cured by ligation of common carotid artery. *Med Chir Trans* 1917; 2:1.

Carpenter Syndrome (Acrocephalopolysyndactyly Type II) 202

General: Hereditary; transmitted as an autosomal recessive trait; severe form of Apert syndrome; normal intelligence has been reported with this syndrome; polysyndactyly is not an absolute requirement for this diagnosis.

Ocular: Lateral displacement of inner canthus; epicanthal folds; microcornea; corneal opacities.

Clinical: Acrocephalopolysyndactyly; brachydactyly; peculiar facies; obesity; mental retardation; hypogonadism; generalized aminoaciduria; cryptorchidism; hypogenitalism.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Jamil MN, et al. Carpenter's syndrome (acrocephalopolysyndactyly type II) with normal intelligence. *Br J Neurosurg* 1992; 6:243-247.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Temtamy SA. Carpenter's syndrome: acrocephalopolysyndactyly. *J Pediatr* 1983; 69:111.

Cataract, Anterior Polar 203

General: Autosomal dominant; imperfect separation of lens from surface ectoderm during fifth week of embryologic development; abnormal mass in region of anterior pole and incomplete resorption of blood vessels and mesoderm at anterior pole of embryonic lens; they can be associated with chromosomal abnormalities including 3, 18 chromosomal translocation.

Ocular: Small opacities on anterior surface of lens; microphthalmia; cataracts usually do not interfere with vision; corneal astigmatism.

Clinical: None.

Bouzas AG. Anterior polar congenital cataract and corneal astigmatism. *J Pediatr Ophthalmol Strabismus* 1992; 29: 210-212.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Moross T, et al. Autosomal dominant anterior polar cataracts associated with a familial translocation. *J Med Genet* 1984; 21:52-53.

Rubin SE, et al. Anterior polar cataract in two sisters with an unbalanced 3, 18 chromosomal translocation. *Am J Ophthalmol* 1994; 117:512-515.

Cataract and Congenital Ichthyosis 204

General: Autosomal recessive; rare.

Ocular: Cortical cataract.

Clinical: Ichthyosis.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Pinkerton OD. Cataract associated with congenital ichthyosis. *Arch Ophthalmol* 1958; 60:393-396.

Cataract, Congenital or Juvenile (Cataract, Juvenile, Hutterite Type) 205

General: Autosomal recessive; seen most frequently in people of Japanese origin; autosomal dominant inheritance also has been reported.

Ocular: Retinitis pigmentosa; Usher syndrome (retinitis pigmentosa and congenital deafness); congenital cataract of the "i" phenotype; microphthalmos; keratoconus.

Clinical: Congenital deafness; galactokinase deficiency; epimerase deficiency.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Skokeir MHK, Lowry RB. Juvenile cataract in Hutterites. *Am J Med Genet* 1985; 22:495-500.

Cataract, Congenital Total with Posterior Sutural Opacities 206

General: Sex-linked; initial lens changes occur in both men and women with continuation of process in men; women show progression at much later age; it has been suggested that several X-linked cataract syndromes are due to deletions of different sizes in the X chromosome.

Ocular: Y-shaped sutural cataracts; congenital cataracts; nuclear cataract; cortical cataract; posterior subcapsular cataract; asymptomatic posterior y-sutural cataracts; severe visual impairment; bilateral pendular nystagmus; bilateral microcornea; exotropia; keratoconus.

Clinical: Mental retardation.

Crews SJ, Bundey SE. Is there an X-linked form of congenital cataracts? *Clin Genet* 1982; 21:351-353.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Waarburg M. X-linked cataract and X-linked microphthalmos: how many deletion families? [Letter] *Am J Med Genet* 1989; 34:451-453.

Cataract, Crystalline Aculeiform or Frosted 207

General: Autosomal dominant.

Ocular: Small crystal-like opacities of lens.

Clinical: None.

Gifford SR, Puntenney I. Coralliform cataract and a new form of congenital cataract with crystals in the lens. *Arch Ophthalmol* 1937; 17:885-892.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Cataract, Crystalline Coralliform 208

General: Autosomal dominant.

Ocular: Cataracts characterized by fine crystals in the axial region of the lens.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Riad M. Congenital familial cataract with cholesterin deposits. *Br J Ophthalmol* 1938; 22:745-749.

Cataract, Floriform 209

General: Autosomal dominant; rare.

Ocular: Lens opacity takes the form of annular elements, arranged either independently or grouped together like petals of a flower; lenticonus; aniridia.

Clinical: None.

Doggart JH. Congenital cataract. *Trans Ophthalmol Soc U K* 1957; 77:31-37.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Cataract, Membranous 210

General: Autosomal dominant.

Ocular: Total cataract that has undergone regression or resorption.

Clinical: None.

Gruber M. Ueber Primaere Familaere Linsendysplasie. *Ophthalmologica* 1945; 110:60-73.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Cataract, Microcornea Syndrome 211

General: Autosomal dominant; prominent in Sicilian families.

Ocular: Cataracts; microcornea; myopia.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Mellica F, et al. Autosomal dominant cataract and microcornea associated with myopia in a Sicilian family. *Clin Genet* 1985; 28:42-46.

Polomano RC, et al. Autosomal dominant cataracts and microcornea. *Can J Ophthalmol* 1979; 14:227-229.

Salmon JF, et al. Variable expressivity of autosomal dominant microcornea with cataract. *Arch Ophthalmol* 1988; 106:505-510.

Cataract, Microphthalmia and Nystagmus 212

General: Autosomal recessive.

Ocular: Miosis; cataract; nystagmus; microphthalmia.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Temtamy SA, Shalach BA. Genetic heterogeneity of the syndrome: microphthalmos with congenital cataract. *Birth Defects* 1974; 10:292-293.

Zeiter HJ. Congenital microphthalmos: a pedigree of 4 affected siblings and an additional report of 44 sporadic cases. *Am J Ophthalmol* 1963; 55:910-922.

Cataract, Nuclear (Coppock Cataract; Cataract, Discoid) 213

General: Autosomal dominant; epidemiologic evidence suggests that a single major gene can account for the correlation among siblings of nuclear sclerosis.

Ocular: Congenital zonular cataract; total nuclear cataract; fetal nucleus with scattered fine diffuse cortical opacities and incomplete cortical riders.

Clinical: None.

Harman NB. Congenital cataract: a pedigree of five generations. *Trans Ophthalmol Soc U K* 1909; 29:101-108.

Heiba JM, et al. Genetic etiology of nuclear cataract: evidence for a major gene. *Am J Med Genet* 1993; 47:1208- 1214.

Lee JB, Benedict WL. Hereditary nuclear cataract. *Arch Ophthalmol* 1950; 44:643-650.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Merin S. Inherited cataracts. In: Merin S, ed. *Inherited eye diseases*. New York: Marcel Dekker, 1991:86-120.

Cataract, Nuclear Diffuse Nonprogressive 214

General: Autosomal dominant; nonprogressive.

Ocular: Opacity of fetal nucleus resembles senile nuclear sclerosis.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Cataract, Posterior Polar 215

General: Autosomal dominant; onset in childhood; progressive.

Ocular: Congenital posterior polar opacity; scattered cortical opacities; choroideremia; myopia.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Valk LE, et al. A case of familial dwarfism, with choroideremia, myopia, posterior polar cataract and zonular cataract. *Ophthalmologica* 1956; 132:299.

Cat's-Eye Syndrome (Schachenmann Syndrome; Schmid-Fraccaro Syndrome; Partial Trisomy G Syndrome) 216

General: Causative factor is one extra chromosome, a G chromosome, which may be from a 13-15 or 21-22 chromosome; although the ocular findings of the syndrome are similar to the D 13-15 trisomy group, the systemic manifestations usually are less severe; this syndrome is associated with a supernumerary bisatellited marker chromosome derived from duplicated regions of 22pter→22q11.2; partial cat's-eye syndrome is characterized by the absence of coloboma.

Ocular: Hypertelorism; microphthalmos; antimongoloid slant of palpebral fissures; strabismus; inferior vertical iris coloboma (cat eye); cataract; choroidal coloboma; epicanthal folds.

Clinical: Anal atresia; preauricular fistulae (bilateral); umbilical hernia; heart anomalies.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Cory CC, Jamison DL. *The cat eye syndrome*. Arch Ophthalmol 1974; 92:259.

Liehr T, et al. Typical and partial cat eye syndrome: identification of the marker chromosome by FISH. *Clin Genet* 1992; 42:91-96.

Mears AJ, et al. Molecular characterization of the marker chromosome associated with cat eye syndrome. *Am J Hum Genet* 1994; 55:134-143.

Peterson RA. Schmid-Fraccaro syndrome ("cat's eye" syndrome): partial trisomy of G chromosome. *Arch Ophthalmol* 1973; 90:287.

Schachenmann G, et al. Chromosomes in coloboma and anal atresia. *Lancet* 1965; 2:290.

Weleber RG, et al. Cytogenetic investigation of cat-eye syndrome. *Am J Ophthalmol* 1977; 84:477.

Cebocephalia 217

General: Term derived from *Cebus* monkey-like head, defective nose, and eyes close together.

Ocular: Hypotelorism; mongoloid obliquity,

Clinical: Flat, incomplete nose; full cheeks; medial nostril; no palate or cleft lips.

Cohen MM. Holoprosencephaly revisited. *Am J Dis Child* 1974; 127:597.

Magalini SI, Scrascia E, *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Central Nervous System Deficiency Syndrome (Garland Syndrome; Spillan-Scott Syndrome) 218

General: Cause unknown; found in prisoners who had long been on a deficient diet; no improvement after normal diet resumed.

Ocular: Greatly reduced vision, particularly near vision, increasing over weeks or months but rarely progressing to complete blindness; relative or absolute central or paracentral scotomata; bitemporal pallor of the disks; optic neuropathy.

Clinical: Incomplete bilateral deafness, never proceeding to complete deafness; tinnitus; numbness and tingling in the legs, rarely in the hands; unsteadiness of gait; abnormal tendon reflexes (both hyperactive or absent); peripheral neuropathy.

Garland HG. A central nervous deficiency syndrome. *Proc Roy Soc Med* 1946; 39:178.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Tucker K, Hedges TR. Food shortages and an epidemic of optic and peripheral neuropathy in Cuba. *Nutr Rev* 1993; 51:349-357.

Wilkinson PB, King A. Amblyopia due to vitamin deficiency. *Lancet* 1944; 1:528.

Cerebellar Ataxia, Cataract, Deafness, and Dementia or Psychosis (Heredopathia Ophthalmo-Oto-Encephalica) 219

General: Autosomal dominant.

Ocular: Posterior polar cataracts.

Clinical: Tremor, paranoid psychosis; dementia; deafness.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Stromgren E. Heredopathia ophthalmo-oto-encephalica. Neurogenetic directory, part 1. *Handbook Clin Neurol* 1981; 42:150-152.

Cerebellar Ataxia, Infantile, with Progressive External Ophthalmoplegia 220

General: Autosomal recessive; neurologic lesion.

Ocular: Paralysis of all extraocular muscles; ptosis; retinal degeneration; blindness.

Clinical: Spinocerebellar degeneration; ataxia.

Jampel RS, et al. Ophthalmoplegia and retinal degeneration associated with spinocerebellar ataxia. *Arch Ophthalmol* 1961; 6:247-259.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Cerebellar Degeneration with Slow Movements 221

General: Autosomal dominant; described only in Indian families; associated with spinocerebellar degeneration and abnormal eye movements.

Ocular: Paramedian pontine reticular formation (horizontal gaze center); absent rapid movements of both eyes and abnormally slow movements.

Clinical: Brainstem lesion of paramedian pontine reticular formation; progressive mental deterioration.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Wadia NH, et al. A new form of heredofamilial spinocerebellar degeneration with slow eye movements. *Brain* 1971; 94:359-374.

Cerebral Autosomal Domint Arteriopathy 222

General: Automal dominant, generalized non-atherosclerotic nonamyloid arteriopathy

Ocular: Scotoma with migraine, cataract, iris atrophy, retinal microinfarction

Clinical: Recurrent stroke, cognitive decline, subcortical vascular dementia

Kreusel KM, Bechrakis N, Krause L, et al. Retinal angiomatosis in von Hippel-Lindau disease. *Ophthalmology* 2006; 113: 1418-1424.

Roine S, Harju M, Kivela TT, et al. Ophthalmologic findings in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. *Ophthalmology* 2006 113: 1411-1417.

Cerebral Cholesterinosis (Cerebrotendinous Xanthomatosis; CTX) 223

General: Autosomal recessive; lipid storage characterized by progressive neurologic dysfunction; large amounts of cholestanol and cholesterol in every tissue in body, particularly in brain and lungs.

Ocular: Cataracts; juvenile cataracts.

Clinical: Cerebellar ataxia; systemic spinal cord involvement; atherosclerosis; mental retardation; unsteady gait; liver damage; jaundice; chronic diarrhea.

Berginer VM, et al. Long-term treatment of cerebrotendinous xanthomatosis with chemodioxycholic acid. *N Engl J Med* 1984; 311:1649-1652.

Cruysberg JR, et al. Juvenile cataract associated with chronic diarrhea in pediatric cerebrotendinous xanthomatosis. *Am J Ophthalmol* 1991; 112:606-607.

Katy DA, et al. Peripheral neuropathy in cerebrotendinous xanthomatosis. *Arch Neurol* 1985; 42:1008-1010.

Cerebral Palsy 224

General: Group of diverse nonprogressive syndromes resulting from injury to the motor centers of the brain; lesions may occur prenatally, in infancy, or in childhood up to age 5 years or more; constitutes the most common cause of permanent physical handicap in children.

Ocular: Strabismus; ptosis; congenital cataract; optic nerve atrophy; papilledema; iris coloboma; nystagmus; uveitis; paresis of extraocular muscles; blepharospasm; leukoma.

Clinical: Systemic abnormalities, such as mental retardation, seizures, microcephalus, hydrocephalus, speech delays, and behavioral or emotional disturbances; motor defect; central visual impairment due to cerebral cortex and white matter malformation.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Harcourt B. Strabismus affecting children with multiple handicaps. *Br J Ophthalmol* 1974; 58:272-280.

Levy NS, et al. Strabismus in children with cerebral palsy. *J Pediatr Ophthalmol* 1976; 13:72-74.

Schenk-Rootlieb AJ, et al. Cerebral visual impairment in cerebral palsy: relation to structural abnormalities of the cerebrum. *Neuropediatrics* 1994; 25:68-72.

Cerebrofacial-Reno-Arthro-Syndactylia Syndrome 225

General: Cause familial?

Ocular: Trichiasis; slanted and asymmetrical size of lid fissure; blepharitis; contraction of visual fields; peripheral pigmentary anomalies of the retina.

Clinical: Mild oligophrenia; small head for body size; slight facial asymmetry; webbed digits; atrophic unilateral sternocleidomastoid muscle; spontaneous shoulder dislocation; small kidneys with renal dysplasia; chronic interstitial nephritis.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Greig DM. Hypertelorism: a hitherto undifferentiated congenital craniofacial deformity. *Edinburgh Med J* 1924; 31:560.

Hanley FJ. Congenital partial hemihypertrophy of the face. *J Oral Surg* 1968; 26:136.

Cerebro-Oculo-Facio-Skeletal Syndrome (COFS Syndrome) 226

General: Inherited as autosomal recessive disorder; death within the first 3 years of life; feeding difficulties secondary to incoordination of the swallowing mechanism.

Ocular: Microphthalmia; blepharophimosis; cataracts.

Clinical: Microcephaly; hypotonia; prominent nasal root; large ear pinnae; flexion contractures at elbows and knees; camptodactylia; osteoporosis; kyphosis; scoliosis; congenital muscular dystrophy.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gershoni-Baruch, et al. Cerebrooculo-facio-skeletal syndrome: further delineation. *Am J Med Genet* 1991; 41:74-77.

Lowry RB, et al. Cataracts, microcephaly, kyphosis and limited joint movement in two siblings. A new syndrome. *J Pediatr* 1971; 79:282.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*. 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pena SDJ, Shokeir MKH. Autosomal recessive cerebrooculo-facio-skeletal (COFS) syndrome. *Clin Genet* 1974; 5:285.

Cerebroretinal Vasculopathy 227

General: Autosomal dominant; frontoparietal lobe pseudotumor and retinal capillary abnormalities; neuropathologic findings are largely confined to the white matter.

Ocular: Capillary obliteration; retinal telangiectasia; glaucoma; cystoid degeneration; branch retinal vein occlusion; infectious retinitis; reduction in central visual acuity; progressive retinal ischemia.

Clinical: Jacksonian and grand mal seizures; headaches; loss of memory; slow speech; cytomegalic virus septicemia; perivascular inflammation; hepatic fibrosis; intracranial mass lesion; systemic collagen vascular disease; systemic lupus erythematosus; central nervous system disease.

Grand MG, et al. Cerebroretinal vasculopathy. *Ophthalmology* 1988; 95:649-659.

Grand MG, et al. Cerebroretinal vasculopathy: a new hereditary syndrome. *Ophthalmology* 1988; 95:649.

Jabs DA, et al. Severe retinal vasoocclusive disease in systemic lupus erythematosus. *Arch Ophthalmol* 1987; 104: 558-563.

Rizzo J. The retina. In: Lessel S, van Dalen JTW, eds. *Current neuro-Ophthalmology,* 3rd ed. St. Louis: Mosby-Year Book, 1991:237.

Ceroid Lipofuscinosis 228

General: Ceroid lipofuscinoses are disorders characterized by the accumulation of fluorescent lipopigments in a number of body tissues; included in this group are several diseases that were once considered variants of Tay-Sachs disease but are now classified separately; ceroid lipofuscinoses may be divided into infantile, late infantile (Bielschowsky-Jansky), juvenile (Spielmeyer- Vogt), adult (Kufs), and atypical forms (see Dollinger-Bielschowsky Syndrome; Kufs Disease; Infantile Neuronal Ceroid Lipofuscinosis; Batten-Mayou syndrome.)

Ocular: Tapetoretinal degeneration; pigmentary macular changes.

Clinical: Seizures; ataxia; dementia; cerebellar and extrapyramidal signs; "release" hallucinations.

Beckerman BH, Rapin I. Ceroid lipofuscinosis. *Am J Ophthalmol* 1971; 72:342.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Fenichel GM. Ceroid lipofuscinosis. In: Fenichel GM, ed. *Clinical pediatric neurology.* 2nd ed. Philadelphia: WB Saunders, 1993: 140-141.

Lake BD, Henderson DC, Oakhill A, et al. Bone marrow transplantation in Batten disease (neuronal ceroid lipofuscinosis). Will it work? Preliminary studies on coculture experiments and on bone marrow transplant in late infantile Batten disease. *Am J Med Genet* 1995; 57:369-373.

Lanska DJ, Lanska MJ. Visual "release" hallucinations in juvenile neuronal ceroid lipofuscinosis. *Pediatr Neurol* 1993; 9:316-317.

Cestan-Chenais Syndrome (Cestan [1] Syndrome) 229

General: Combination of Babinski-Nageotte and Avellis syndromes; lesion in the lateral portion of medulla oblongata.

Ocular: Enophthalmos; ptosis; nystagmus; miosis.

Clinical: Pharyngolaryngeal or glossopharyngeal paralysis; cerebellar hemiataxia; disturbance of sensibility; contralateral side of lesion.

Cestan R, Chenais L. Du Myosis dans Certaines Lesions Bulbaires en Foyer Hemiplegie du Type Avellis Associee au Syndrome Oculaire Sympathique. *Gaz D Hop* 1903; 76:1229.

Kistler JP, Ropper AH, Martin JB. Cerebrovascular diseases. In: Isselbacher KJ, ed. *Harrison's Principles of Internal Medicine,* 13th ed. New York: McGraw-Hill, 1994:2242-2243.

Sheehan S, et al. Vertebral artery compression in cervical spondylosis. Arteriographic demonstration during life of vertebral artery insufficiency due to rotation and extension of the neck. *Neurology* 1960; 10:968.

Charcot-Marie- Tooth Disease (Progressive Neuritic Muscular Atrophy; Progressive Peroneal Muscular Atrophy) 230

General: Dominant inheritance; onset between 5 and 15 years; rare disease; demonstrates autosomal dominant as well as recessive and X-linked recessive inheritance.

Ocular: Nystagmus; vision reduced if associated with optic nerve involvement; primary optic atrophy (rare).

Clinical: Positive familial history; atrophy of small muscles of hands and feet, slowly progressing to distal and then proximal arm and leg; fibrillary muscle twitchings (fasciculations) are common; cramps are common.

Chance PF, Lupski JR. Inherited neuropathies: Charcot-Marie- Tooth disease and related disorders. *Baillieres Clin Neurol* 1994; 3:373-385.

Charcot JM, Marie P. Sur one Forme Particuliere d' Atrophie Musculaire Progressive, Souvent Familiale, Debutant par les Pieds et les Jambes, et Atteignant Plus Tard les Mains. *Rev Med* 1886; 6:97.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

McClain LW. Charcot-Marie-Tooth disease. *JAMA* 1974; 229:767.
Siegel S. Charcot-Marie-Tooth disease. *JAMA* 1974; 228:873.
Tooth HH. The peroneal type of progressive muscular atrophy. London: HL Lewis, 1886.

Charcot-Wilbrand Syndrome 231

General: Lesion of artery of angular gyrus of dominant side; lesion can be bilateral; alexia.

Ocular: Visual agnosia; loss of ability to revisualize images; prosopagnosia.

Clinical: Occlusion of a portion of the posterior cerebral artery.

Charcot JM. Sur un Cas de Cecile Verbale. Oeuvres Completes de Charcot. Paris: Delahaye Lecrosnier, 1887.
Feinberg TE, et al. Associative Visual agnosia and alexia without prosopagnosia. Cortex 1994; 30:395-411.
Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.
Rentschler I, et al. Dissociation of local and global processing in visual agnosia. Vis Res 1994; 34:963-971.
Shelton PA, et al. Apperceptive visual agnosia: a case study. Brain Cogn 1994; 25:1-23.

Charge Association (Multiple Congenital Anomalies Syndrome; Coloboma, Heart Disease, Atresia, Retarded Growth, Genital Hypoplasia, Ear Malformation Association) 232

General: Syndrome consisting of four of six major manifestations of ocular coloboma, heart disease, atresia, retarded growth and development, genital hypoplasia, and ear malformations with or without hearing loss.

Ocular: Blepharoptosis; iris coloboma; optic nerve coloboma; macular hypoplasia; lacrimal canalicular atresia; nasolacrimal duct obstruction.

Clinical: Microcephaly; brachycephaly; malformed ear; bilateral finger contractures; heart disease; genital hypoplasia; heart disease; choanal atresia; retarded growth; hearing loss; facial nerve palsies; mental retardation.

Bowling BS, Chandna A. Superior lacrimal canalicular atresia and nasolacrimal duct obstruction in the CHARGE association, *J Pediatr Ophthalmol Strabismus* 1994; 31:336-337.
Lin CC, et al. Coloboma of optic nerve associated with serous maculopathy. A clinicopathologic correlative study. *Arch Ophthalmol* 1984; 102:1651.
Morgan D, et al. Ear-nose-throat abnormalities in the CHARGE association. *Arch Otolaryngol Head Neck Surg* 1993; 119:49-54.
Warburg M. Ocular coloboma and multiple congenital anomalies. The CHARGE association. *Ophthalmic Paediatr Genet* 1983; 2:189.

Charlin Syndrome (Nasal Nerve Syndrome; Nasociliaris Nerve Syndrome; Nasociliary Syndrome) 233

General: Neuritis of the nasal branch of the trigeminal nerve; three typical spots of pain according to the nerve distribution: (i) above and outside the nose; (ii) above the inner canthus; and (iii) inferior angle of the medial tarsal ligament (see Sluder Syndrome).

Ocular: Severe ocular and orbital pain, mainly upper nasal-orbital angle; slight inflammatory swelling of upper lid (occasional); photophobia; ciliary and conjunctival injection; pseudopurulent conjunctivitis; anterior uveitis; iritis; hypopyon; keratitis; corneal ulcers.

Clinical: Rhinorrhea; rhinitis always on same side of the ocular involvement; severe pain of ala nasi.

Charlin C. Le Syndrome du Nerf Nasal. *Ann Ocul* 1931; 168:86.
Ferrannini G. Charlin's syndrome. *Ann Ophthalmol Clin Ocul* 1969; 95:807.
Pau H. *Differential diagnosis of eye disease*. New York: Thieme, 1987.

Chediak-Higashi Syndrome (Anomalous Leukocytic Inclusions with Constitutional Stigmata) 234

General: Occurs in albinoid siblings born of consanguineous parents; tyrosinase-positive type of oculocutaneous albinism associated with a fetal reticuloendothelial incompetence.

Ocular: Decreased iris pigmentation; photophobia; narrowness and decreased number of vessels in retina; decreased pigmentation of choroid elevated disk; papilledema; infiltration of immature leukocytes in the uvea, retina, and optic nerve; nystagmus; ocular motor palsies; optic disk edema; oculocutaneous albinism.

Clinical: Anemia; neutropenia; thrombocytopenia; recurrent infections; hepatosplenomegaly; lymphadenopathy; oculocutaneous albinism; hyperpigmentation of sun-exposed areas.

Benezra D, et al. Chediak-Higashi syndrome: ocular findings. *J Pediatr Ophthalmol Strabismus* 1980; 17:68-74.
Chediak M. Nouvelle Anomalie Leucocytaire de Caractere Constitutionale et Familial. *Rev Hemat* 1952; 7:362.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fong DS, Pruett RC. Systemic associations with myopia. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3146.

Valenzuela R, Morningstar WA. The ocular pigmentary disturbance of human Chediak-Higashi syndrome. A comparative light and electron-microscopic study and review of the literature. *Am J Clin Pathol* 1981; 75:591-596.

Cherry-Red Spot Myoclonus Syndrome (Type I Sialidosis) 235

General: Morphologic changes in storage process in lysosomes of retinal ganglion cells, neurons of mesenteric plexus, hepatocytes, and Kupffer cells.

Ocular: Cherry-red spot of macula; horizontal nystagmus; white dot and flake lens changes; decreased visual acuity and gray perimacular halo.

Clinical: Development of a slowly progressive neurologic illness whose most crippling symptoms are myoclonus, seizures, decreased muscle tone, speech disorder, and hyperactive reflexes.

Engel J Jr, et al. Electrophysiological studies in two patients with cherry red spot-myoclonus syndrome. *Epilepsia* 1977; 18:73-87.

Neetens A, et al. Cherry red spot-ataxia-myoclonus. *Bull Soc Belg Ophthalmol* 1979; 186:67-76.

Rapin I. The cherry-red spot-myoclonus syndrome. *Ann Neurol* 1978; 3:234.

Chickenpox (Varicella) 236

General: Acute exanthematous disease; highly contagious; children between ages of 2 and 8 years.

Ocular: Conjunctival ulcer; corneal ulcer; descemetocele; corneal opacity; keratitis; paresis of third, fourth, and sixth nerves; optic neuritis; papilledema; retinitis; hemorrhagic retinopathy; uveitis; cataract; paralytic mydriasis; phthisis bulbi; unifocal choroiditis; dendritic keratitis; acute retinal necrosis (in a patient with acquired immunodeficiency syndrome [AIDS]); disciform keratitis.

Clinical: Fever; malaise; rash; pruritus.

Bierly JR, Ostler HB. Varicella dendritic keratitis. *J Pediatr Ophthalmol Strabismus* 1994; 31:53-56.

Deegan WF 3rd. Unifocal choroiditis in primary varicella zoster (chickenpox). *Arch Ophthalmol* 1994; 112:753-756.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Friedman SM, et al. Acute retinal necrosis after chickenpox in a patient with acquired immunodeficiency syndrome. *Arch Ophthalmol* 1993; 111:1607-1608.

Linnemann CC, Alvira MM. Pathogenesis of varicella-zoster angiitis in the CNS. *Arch Neurol* 1980; 37:239-240.

Nemet P, et al. Benign abducens palsy in varicella. *Am J Ophthalmol* 1974; 78:859.

Pavan-Langston D. Varicella-zoster ophthalmicus. *Int Ophthalmol Clin* 1975; 15: 171-185.

Chlamydia (Inclusion Conjunctivitis; Paratrachoma) 237

General: Organism that infects the epithelium of mucoid surfaces; sexually transmitted; major cause of nongonococcal urethritis in men and cervicitis in women; major cause of neonatal ophthalmia; *Chlamydia trachomatis* is an intracellular bacterium lacking respiratory enzymes that has an affinity for mucosal epithelium; serotypes A through C have been epidemiologically associated with trachoma; serotypes E through K have been associated with genital infection and keratoconjunctivitis in sexually active adults and neonates; other serotypes have been associated with lymphogranuloma venereum and Reiter syndrome.

Ocular: Follicular conjunctivitis; corneal opacities; keratitis; corneal ulcer; lid edema; uveitis.

Clinical: Pneumonia; gastrointestinal disturbances; genital discharge.

Dawson CR, et al. Inclusion conjunctivitis and Reiter's syndrome in a married couple. Chlamydia infections in a series of both diseases. *Arch Ophthalmol* 1970; 83:300-306.

Fraunfelder FT, Roy FR. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Grossman M, et al. Prospective studies in chlamydia in newborns. In: Mardh P, et al., eds. *Chlamydial infections.* Amsterdam: Elsevier Biomedical, 1982.

Malaty R. Chlamydia. In: Tabbara K, Hyundiuk RA, eds. *Infections of the eye.* Boston: Little, Brown and Company, 1986:107,114.

Stephens RS. Chlamydiology. In: Smolin G, Thoft RA, eds. *The Cornea*. Boston: Little, Brown and Company, 1994: 277-282.

Cholera 238

General: Acute illness that results from colonization of small bowel by *Vibrio cholerae;* rare cases of *V. cholerae* meningitis have been reported, some associated with bacteremia.

Ocular: Hyperemia of lids and conjunctivae; subconjunctival hemorrhages; madarosis of lids; xerosis of conjunctiva; lagophthalmos; keratomalacia; retinal ischemia; cataract.

Clinical: Diarrhea; shock; vomiting; muscle cramps; cyanosis; scaphoid abdomen; thready pulse; tachycardia; hypotension; tachypnea; acute tubular necrosis; metabolic acidosis; death.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*, 11th ed. New York: McGraw-Hill, 1987.

Naidu LS, et al. Vibrio cholerae non-0:1 meningitis in an infant. *Pediatr Infect Dis J* 1993; 12:879-88l.

Patil SN, et al. Sixth nerve paralysis following cholera inoculation. *Ind J Ophthalmol* 1977; 24:37.

Cholestasis with Gallstone, Ataxia, and Visual Disturbances 239

General: Autosomal recessive; not clear if distinct from Byler disease or another form of intrahepatic cholestasis; retinal neurologic features may be secondary to nutritional abnormalities.

Ocular: Retinal lesions; optic atrophy; ptosis.

Clinical: Congenital cholestasis; gallstone; cerebellar ataxia; jaundice; hepatitis; pruritus.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Tagawa Y, Konno T. Familial cholestasis with gallstone, ataxia and visual disturbance. *Tohoku J Exp Med* 1982; 137: 137-144.

Chorea (Acute Chorea; Sydenham Chorea; St. Vitus Dance; Huntington Hereditary Chorea) 240

General: Mendelian dominant trait.

Ocular: Lid retraction; spasmodic closures; apraxia of lid opening; disoriented ocular movements; anisocoria; mydriasis; hippus.

Clinical: Involuntary purposeless movements; emotional ability; muscle weakness.

Avanzini G, et al. Oculomotor disorders in Huntington's chorea. *J Neurol Neurosurg Psychiatry* 1979; 42:581-589.

DePen G, et al. Disturbance of eye movements in Huntington's chorea. *Arch Psychiatr Nervenkr* 1981; 229:205-213.

Duane TD. *Clinical Ophthalmology* Philadelphia: JB Lippincott, 1987.

Fahn S. Movements disorders. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1995:695-703.

Rowlan LP, Pedley TA. Abetalipoproteinemia. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1995:594-596.

Choroideremia (Tapetochoroidal Dystrophy, Progressive; Choroidal Sclerosis) 241

General: Sex-linked; onset at early age; progressive; primary degeneration may be of the retina, retinal pigment epithelium, or choriocapillaris; pigment stippling or granularity also evident in female carriers who possess normal and abnormal cells, through Barr body inactivation of one X chromosome.

Ocular: Reduction of central vision; constriction of visual fields; night blindness; choroidal and retinal atrophy.

Clinical: None.

Flannery JG, et al. Histopathologic study of a choroideremia carrier. *Invest Ophthalmol Vis Sci* 1990; 31:229.

Lesko JG, et al. Choroideremia is tightly linked to two proximal XQ chromosomal markers. *Am J Hum Genet* 1985; 37:165.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Merin S. Choroideremia. In: Merin S, ed. *Inherited Eye Diseases: Diagnosis and Clinical Management.* New York: Marcel Dekker, 1991:312-318.

Choroidoretinal Degeneration with Retinal Reflex in Heterozygous Women 242

General: Sex-linked; choroidoretinal degeneration differentiated by presence in heterozygous women of a tapetal-like retinal reflex; there is probably more than one X-linked locus leading to a retinitis pigmentosa type of picture.

Ocular: Retinitis pigmentosa; golden-hued, patchy appearance around macula.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Musarella MA, et al. Multipoint linkage analysis and heterogeneity testing in 20 X-linked RP families. *Genomics* 1990; 8:286-296.

Nussbaum RL, et al. Mapping X-linked ophthalmic diseases: linkage relationship of X-linked retinitis pigmentosa to X chromosomal short arm markers. *Hum Genet* 1985; 70:45-50.

Choroidoretinal Dystrophy 243

General: Sex-linked; similar to retinitis pigmentosa with absence of annular scotoma and little vascular change.

Ocular: Early poor central vision; retinitis pigmentosa; night blindness.

Clinical: None.

Hoare GW. Choroidoretinal dystrophy. *Br J Ophthalmol* 1965; 49:449-451.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press. 1998.

Chromosome 11 Long-Arm Deletion Syndrome 244

General: Patients with deletion of the long arm of chromosome 11 exhibit a distinctive countenance; female preponderance.

Ocular: Colobomas of the choroid, retina, and iris; retinal reduplication; retinal dysplasia; epicanthus; blepharoptosis; abnormal slanting of the interpalpebral fissures; bilateral uveal colobomas; hypertelorism; avascular retina (bilateral); abnormal pattern of retinal vessels.

Clinical: Keeled forehead; small carp-shaped mouth; low-set ears; highly arched palate; long upper lip with absent philtrum; short neck; widely spaced nipples; flexion contractures of the knees and elbows; hypoplastic nails; broad thumbs with low insertion; deeply pigmented skin on buttocks, lower back, and abdomen and in inguinal regions, axillas, and clavicular areas; congenital heart disease.

Ferry AD, et al. Ocular abnormalities in deletion of the long arm of chromosome 11. *Ann Ophthalmol* 1981; 13: 1373-1377.

Uto H, et al. A case of 11q- syndrome associated with abnormalities of the retinal vessels. *Ophthalmologica* 1994; 208:233-622.

Chromosome 13q Partial Deletion (Long-Arm Syndrome; 13q Syndrome) 245

General: No hereditary factor.

Ocular: Microphthalmos; antimongoloid slant of lid fissures; bilateral epicanthus; esotropia; cataract; choroidal coloboma; ptosis; retinoblastoma.

Clinical: Genital malformations; meningocele; short neck; small mouth; mental and physical retardation; small head; short stature; broad nasal bridge; simian crease; microcephaly; high nasal bridge; thumb hypoplasia.

Kennerknecht I, et al. Diagnosis of retinoblastoma in a presymptomatic stage after detection of interstitial chromosomal deletion 13q. *Ophthalmic Genet* 1994; 15:19-24.

Kivela T, Tuppurainen K, Riikonen P, et al. Retinoblastoma associated with chromosomal 13q14 deletion mosaicism. *Ophthalmology* 2003; 110: 1983-1988.

Larned DC, et al. Association of congenital ptosis and congenital heart disease. *Ophthalmology* 1986; 93:492-494.

Pratt CB, et al. Outcome for patients with constitutional 13q chromosomal abnormalities and retinoblastoma. *Pediatr Hematol Oncol* 1994; 11:541-547.

Weiss A, Margo C. Bilateral microphthalmos with cyst and 13q deletion syndrome. *Arch Ophthalmol* 1987; 105:25.

Wilson L, et al. Cytogenetic analysis of a case of "13q- syndrome" (46, XX, del 13) using band technique. *J Pediatr Ophthalmol Strabismus* 1980; 17:63-67.

Chromosome 18 Partial Deletion (Long-Arm) Syndrome (Monosomy 18 Partial [Long-Arm] Syndrome; De Grouchy Syndrome) 246

General: Deletion of approximately one half of the long arm of chromosome 18.

Ocular: Hypertelorism; epicanthal folds; narrow palpebral fissure; nystagmus (horizontal); strabismus; myopia; astigmatism; glaucoma; oval pupils; microcornea; posterior staphyloma; oblique disk; optic nerve staphyloma; optic nerve atrophy; microphthalmia; corneal opacities; iris hypoplasia; corectopia.

Clinical: Dwarfism; mental retardation; microcephaly; midface dysplasia; prominent antihelix and antitragus; congenital cardiac disease; abnormal, spindle-shaped fingers; genital defects.

de Grouchy J, et al. Deletion Partielle des Bras Longs du Chromosome 18. *Pathol Biol (France)* 1964; 12:579.

Ginsberg J, et al. Ocular manifestations in trisomy 18. *Am J Ophthalmol* 1968; 66:59.

Izquierdo NJ, et al. Anterior segment malformations in 18q- (de Grouchy) syndrome. *Ophthalmic Pediatr Genet* 1993; 14:91-94.

Levenson JE, et al. Partial deletion syndromes of chromosome 18. *Ann Ophthalmol* 1971; 3:756.

Chromosome 18 Partial Deletion (Short-Arm) Syndrome (Monosomy 18 Partial [Short-Arm] Syndrome) 247

General: Deletion of the short arm of chromosome 18 (note similarity of clinical features to those of the cri-du-chat syndrome or B1 deletion syndrome) (see Cri-du-Chat Syndrome).

Ocular: Hypertelorism; epicanthal folds; ptosis; mongolian or antimongolian slant; strabismus; eccentric pupil; cataract; corneal opacities; concentric visual field defects.

Clinical: Short stature; mental retardation; low-set ears; dysphagia; moon face; oliguria; arhinencephaly; microcephaly; congenital alopecia; flat bridge of nose; pyramidal tract signs; weakness and focal dystonia of the lower extremities.

Buhler EM, et al. Partial monosomy 18 and anomaly of thyroxine synthesis. *Lancet* 1964; 1:170.

Levenson JE, et al. Partial deletion syndromes of chromosome 18. *Ann Ophthalmol* 1971; 3:756.

Yanoff M, et al. Ocular and cerebral abnormalities in chromosome 18 deletion defect. *Am J Ophthalmol* 1970; 10:391.

Chronic Epstein-Barr Virus (Epstein-Barr Virus, Chronic; Chronic Infectious Mononucleosis) 248

General: Onset late adolescence or early adulthood; rare.

Ocular: Bilateral uveitis; cystoid macular edema; papilledema; cataract; keratitis; peripapillary and macular preretinal membranes; vitreitis; lacrimal gland swelling; conjunctivitis; corneal edema; keratoconjunctivitis; follicular conjunctivitis; subepithelial corneal opacities; retinitis; ophthalmoplegia; optic neuritis; endophthalmitis; dacryocystitis; nasolacrimal duct obstruction.

Clinical: Recurrent fever; pharyngitis; lymphadenopathy; fatigue; malaise; weight loss; splenomegaly.

Matoba AY, et al. Epstein-Barr virus stromal keratitis. *Ophthalmology* 1986; 93:746-75l.

Pflugfelder SC. et al. Ophthalmic manifestations of Epstein-Barr virus infection. *Int Ophthalmol Clin* 1993; 33: 95-101.

Steele RJ, Meyer DR. Nasolacrimal duct obstruction and acute dacryocystitis associated with infectious mononucleosis (Epstein-Barr virus). *Am J Ophthalmol* 1993; 115:265-266.

Tosato G, et al. Characteristic T cell dysfunction in patients with chronic active Epstein-Barr virus infection (chronic infectious mononucleosis). *J Immunol* 1985; 135:3082-3088.

Wong KW, et al. Ocular involvement associated with chronic Epstein-Barr virus disease. *Arch Ophthalmol* 1987; 105: 788-792.

Chronic Granulomatous Disease of Childhood 249

General: Genetically determined metabolic defect manifested by inability of the leukocytes to operate the hexose monophosphate shunt during phagocytosis.

Ocular: Conjunctivitis; keratitis; destructive chorioretinal lesions.

Clinical: Eczematous dermatitis; microabscesses of the skin, lymph nodes, and viscera.

Andersen SR. Vascular lesion (arteriovenous aneurysm or haemangioma) of the orbit in a case of chronic granulomatous disease. *Ophthalmologica* 1976; 173:145-15l.

Harley RD, ed. *Pediatric Ophthalmology*. 4th ed. Philadelphia: WB Saunders, 1998.

Lischner HW, Martyn LJ. Chorioretinal lesions, sea-blue histiocytes and other manifestations in familial chronic granulomatous disease. *Birth Defects* 1975; 11:73-76.

Chronic Progressive External Ophthalmoplegia (CPEO; Ophthalmoplegia Plus) 250

General: A general term covering many conditions; onset at any age; familial history; conditions associated with CPEO include myotonic dystrophy, Kearns-Sayre syndrome, Stephens syndrome, and oculopharyngeal dystrophy; disorders that rarely cause external ophthalmoplegia include congenital disorders (abetalipoproteinemia, Refsum disease, extraocular fibrosis syndrome, Möbius syndrome), progressive supranuclear palsy, endocrine exophthalmos, myasthenia gravis, and multiple sclerosis; now considered to be a mitochondrial cytopathy with varied clinical presentation; four distinct disorders of ophthalmic importance are (i) CPEO or Kearns-Sayre syndrome, (ii) myoclonus epilepsy with ragged red fibers (MERRF), (iii) mitochondrial encephalopathy, lactic acidosis, and strokelike episodes (MELAS), and (iv) Leber optic neuropathy.

Ocular: Exposure keratopathy; filamentary keratitis; keratoconjunctivitis sicca; corneal scarring; esotropia; exotropia; gaze paralysis; ptosis; levator paralysis; cataract; optic atrophy; diplopia; tapetoretinal degeneration; constriction of visual field; retinitis pigmentosa.

Clinical: Weakness; weight loss; myopathic or Hutchinson facies; cardiac abnormalities; central nervous system abnormalities.

Borchert MS. Diseases of the ocular muscles. In: Albert DM, Jakobiec FA. eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994:3102-3120.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Lane CM, Collin JRO. Treatment of ptosis in chronic progressive external ophthalmoplegia. *Br J Ophthalmol* 1987; 71:290-294.

Shapiro AHV, et al. Molecular basis of mitochondrial myopathies: polypeptide analysis in complex-I deficiency. *Lancet* 1988; 1:500.

Churg-Strauss Syndrome (Allergic Granulomatosis and Angiitis) 251

General: Severe multisystem vasculitis.

Ocular: Allergic granulomas in the extravascular tissues characterized by a central eosinophilic core consisting of necrotic eosinophilic leukocytes and fibrinoid swelling of collagen fibers; uveoscleritis; papilledema; anterior ischemic optic neuropathy; sensory motor neuropathy; reversible monocular blindness; myositis; episcleritis; amaurosis fugax, central branch retinal artery occlusion; retinal vasculitis; retinal hemorrhage; cranial nerve palsies; orbital inflammatory syndrome; corneal ulcer; conjunctival nodules.

Clinical: Bronchial asthma; fever; eosinophilia; necrotizing of small arteries and veins with an infiltration of vessels and perivascular tissues by eosinophils; systemic vasculitis.

Alberts AR, et al. Reversible monocular blindness complicating Churg-Strauss syndrome. *J Rheumatol* 1994; 21: 363-365.

Billing K, Malhotra R, Selva D, et al. Orbital myositis in Churg-Strauss syndrome. *Arch Ophthalmol* 2004; 122: 393-396.

Churg J, Strauss L. Anergic granulomatosis, allergic angiitis and periarteritis nodosa. *Am J Pathol* 1951; 27:277.

Kattah JC, et al. Anterior ischemic optic neuropathy in Churg-Strauss syndrome. *Neurology* 1994; 44:2200-2202.

Meisler DM, et al. Conjunctival inflammation and amyloidosis in allergic granulomatosis and angiitis (Churg-Strauss syndrome). *Am J Ophthalmol* 1981; 91:216-219.

Rosenblatt BJ, Foroozan R, Savino, PJ: Asymptomatic optic neuropathy associated with Churg-Strauss syndrome. *Ophthalmology* 2003; 110: 1650-1652.

Citrullinemia 252

General: Autosomal recessive; enzyme deficiency called argininosuccinic acid synthetase; mutations causing human citrullinemia are extremely heterogeneous; nonconsanguineous persons studied until 1991 had been found to be compound heterozygotes.

Ocular: Hypotonicity; low intraocular pressure; irregular choroidal and retinal pigment epithelium folding; engorged retinal vessels; pallor of optic disk.

Clinical: Mental retardation; nausea; vomiting; tremors; intermittent hyperammonemia.

Kobayashi K, et al. Additional mutations in argininosuccinate synthetase causing citrullinemia. *Mol Biol Med* 1991; 8:95-100.

Matsuda I, et al. Variant form of citrullinemia. *J Pediatr* 1985; 88:995-1006.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Claude Syndrome (Inferior Nucleus Ruber Syndrome; Rubro-Spinal-Cerebellar-Peduncle Syndrome) 253

General: Paramedian mesencephalic lesion starting in midbrain; often occlusion of terminal branches of the paramedian arteries supplying the inferior portion of the nucleus ruber.

Ocular: Paralysis of ipsilateral oculomotor and trochlear nerves (III, IV).

Clinical: May be associated with motor hemiplegia.

Claude H. Inferior nucleus ruber syndrome. *Rev Neurol* 1912; 1:311.

Cremieux G, Serratrice G. A case of retraction nystagmus associated with Claude's syndrome. *Mars Med (Fre)* 1972; 109:635.

Gaymard B, et al. Two mesencephalic lacunar infarcts presenting as Claude's syndrome and pure motor hemiparesis. *Eur Neurol* 1991; 31:152-155.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Clivus Edge Syndrome 254

General: Elevated intracranial pressure; root of oculomotor nerve pressed against bone as it enters cavernous sinus; includes subdural hematoma, temporal lobe tumor, and supraclinoid aneurysm.

Ocular: Brief miosis, then mydriasis and sluggish reaction of pupil; extraocular muscle paresis.

Fischer-Brugge E. Anatomische Ursachen Funktionaler Kreislaufstorungen des Gehirns und am N. Oculomotorius Bruns. *Beitr Klin Chir* 1951; 181:323-336.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Clostridium Perfringens 255

General: Gram-positive rod; most important cause of gas gangrene infection.

Ocular: Hypopyon; gas bubbles in anterior chamber; endophthalmitis; proptosis; glaucoma; coffee-colored discharge; eyelid edema; severe ocular pain; endophthalmitis after penetrating trauma or metastatic.

Clinical: Traumatized ischemic skeletal muscle, abdominal wall, or uterus; hemolytic anemia; shock; death.

Abu el-Asrar AM, Tabbara KF. Clostridium perfringens endophthalmitis. *Doc Ophthalmol* 1994; 87:177-182.

Frantz JF, et al. Acute endogenous panophthalmitis caused by Clostridium perfringens. *Am J Ophthalmol* 1974; 78*:* 295-303.

Nangia V, Hutchinson C. Metastatic endophthalmitis caused by Clostridium perfringens. *Br J Ophthalmol* 1992; 76: 252-253.

Obertynski H, Dyson C. Clostridium perfringens panophthalmitis. *Can J Ophthalmol* 1974; 9:258-259.

Coarctation of the Aorta 256

General: Congenital, local, and developmental; occurs in one of 2,000; not familial; not predictable by sex.

Ocular: Straightening of the arteriolar tree; arteriovenous crossing defects; focal caliber changes; cotton-wool spots; retinal edema; pronounced corkscrew tortuosity of the arterioles with little other retinal changes in 50% of patients; retinal vascular anomalies secondary to hemodynamic changes.

Clinical: Aorta narrowed to less than 1 cm in the region of the insertion of the ductus arteriosus; dyspnea; headache; epistaxis; palpitations of the heart; thoracic pain; intermittent claudication.

Asch AJ. Turner's syndrome occurring with Horner's syndrome. Coarctation of the aorta and aortic aneurysm. *Am J Dis Child* 1979; 133:827-830.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Granstrom KO. Retinal changes in coarctation of the aorta. *Br J Ophthalmol* 1951; 35:143.

Johns KJ, et al. Retinal vascular abnormalities in patients with coarctation of the aorta. *Arch Ophthalmol* 1991:109: 1266-1268.

Coats Disease (Leber Miliary Aneurysm; Retinal Telangiectasia) 257

General: Exudative retinitis; rare; more common in males than females; 95% unilateral.

Ocular: Leukocoria; telangiectatic retinal vessels; solid gray-yellow retinal detachment; optic atrophy; vitreous hemorrhage; anterior uveitis; glaucoma; intraocular calcification (rare); fibroosseous retinal nodules (atypical); hemorrhagic retinal macrocysts; cystoid macular edema.

Cameron JO. Coats' disease and Turner's syndrome. *Am J Ophthalmol* 1974; 78:852-854.

Campbell FP. Coats' disease and congenital vascular retinopathy. *Trans Am Ophthalmol Soc* 1976; 74:365-424.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Gartner J, Draf W. Aneurysms associated with telangiectasia of the nasal mucosa. *Am J Ophthalmol* 1975; 79:56-58.

Goel SD, Augsburger JJ. Hemorrhagic retinal macrocysts in advanced Coats disease. *Retina* 1991; 11:437-440.

Lee ST, et al. Cystoid macular edema secondary to juxtafoveolar telangiectasis in Coats' disease. *Ophthalmic Surg* 1991; 22:218-221.

Senft SH, et al. Atypical presentation of Coats disease. *Retina* 1994; 14:36-38.

Cocaine Intoxication Syndrome 258

General: Neonates born to cocaine-abusing mothers.

Ocular: Dilation and tortuosity of the iris vasculature; use of intranasal cocaine has been associated with optic neuropathy.

Clinical: Signs of withdrawal; jittery; irritable.

Goldberg RA, et al. Orbital inflammation and optic neuropathies associated with chronic sinusitis of intranasal cocaine abuse. *Arch Ophthalmol* 1989; 107:831.

Isenberg SJ, et al. Ocular signs of cocaine intoxication in neonates. *Am J Ophthalmol* 1987; 103:211-214.

Coccidioidomycosis 259

General: Caused by fungus *Coccidioides immitis.*

Ocular: Conjunctivitis; choroiditis; uveitis; retinal hemorrhages; vitreal opacity; vitreal floaters; episcleritis; hypopyon; granulomatous lesion of optic nerve head; paralysis of sixth cranial nerve; secondary glaucoma; papilledema; mutton fat keratitic precipitates; necrotizing granulomatous conjunctivitis; iridocyclitis.

Clinical: Mild respiratory illness; cavity lung lesion.

Bell R, Font RL. Granulomatous anterior uveitis caused by Coccidioides immitis. *Am J Ophthalmol* 1972; 74:93-98.

Blumenkranz MS, Stevens DA. Endogenous coccidioidal endophthalmitis. *Ophthalmology* 1980; 87:974-984.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Maguire LJ, et al. Coccidioidomycosis with necrotizing granulomatous conjunctivitis. *Cornea* 1994; 13:539-542.

Moorthy RS, et al. Coccidioidomycosis iridocyclitis. *Ophthalmology* 1994; 101:1923-1928.

Zakka KA, et al. Intraocular coccidioidomycosis. *Surv Ophthalmol* 1978; 22:313-321.

Cockayne Syndrome (Dwarfism with Retinal Atrophy and Deafness; Mickey Mouse Syndrome) 260

General: Autosomal recessive; onset in second year of life; wide spectrum of symptoms and severity of the disease suggest that biochemical and genetic heterogeneity exist.

Ocular: Enophthalmos; cataracts; pigmentary degeneration of retina; optic atrophy; band keratopathy; exotropia; nystagmus; absence of foveal reflex; corneal dystrophy; corneal perforation; anhidrosis; exposure keratitis; decreased blinking.

Clinical: Dwarfism (nanism) with disproportionately long limbs, large hands, and large feet; kyphosis; deformed limbs; thickened skull; intracranial calcifications; mental retardation; prognathism; deafness (often partial); precociously senile appearance; sensitivity to sunlight, with skin pigmentation and scarring; dental caries.

Broderick JD, Dark AJ. Corneal dystrophy in Cockayne's syndrome. *Br J Ophthalmol* 1973; 57:391.

Cockayne EA. Dwarfism with Retinal atrophy and deafness. *Arch Child* 1961; 11: 1.

Coles WH. Ocular manifestations of Cockayne's syndrome. *Am J Ophthalmol* 1969; 67:762-764.

Fraunfelder FT, Roy FH, eds. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Levin PS, et al. Histopathology of the eye in Cockayne's syndrome. *Arch Ophthalmol* 1983; 101:1093-1097.

Nance MA, Berry SA. Cockayne syndrome: review of 140 cases. *Am J Med Genet* 1992; 42:68-84.

Pearce WG. Ocular and genetic features of Cockayne's syndrome. *Can J Ophthalmol* 1972; 7:435-444.

Traboulsi EI, et al. Ocular findings in Cockayne syndrome. *Am J Ophthalmol* 1992; 114:579-583.

Coenurosis 261

General: Rare human infestation of the cystic larval stage of the dog tapeworm; usually infestation in the muscle, subcutaneous tissue, eye, nervous system, or brain; three species may be involved: *Multiceps taenia*, *Multiceps serialis*, and *Multiceps glomeratus.*

Ocular: Hypopyon; retinal detachment; retinal edema; anterior uveitis; conjunctivitis; proptosis; miosis; vitreal haze; increased intraocular pressure; coenurus cysts of the conjunctiva and iris.

Clinical: Ataxia; headache; loss of weight; somnolence; stiffness of neck and shoulders.

Boase AJ. Coenurus cyst of the eye. *Br J Ophthalmol* 1956; 40:183-185.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part I*, 4th ed. Baltimore: Williams & Wilkins, 1995:3318.

Scaravilli F. Parasitic and fungal infections. In: Adams JH, Duchen LW, eds. *Greenfield's Neuropathology*, 5th ed. New York: Oxford University Press, 1992:400-446.

Williams PH, Templeton AC. Infection of the eye by tapeworm Coenurus. *Br J Ophthalmol* 1971; 55:766-769.

Cogan (1) Syndrome (Nonsyphilitic Interstitial Keratitis) 262

General: Cause unknown; perhaps a generalized hypersensitivity reaction; most frequently affects young adults; unclear etiology; several studies suggest an autoimmunemediated process, possibly a vasculitis.

Ocular: Blepharospasm; lacrimation; congested conjunctival vessels; little or no reaction in anterior chamber but ciliary injection present; interstitial keratitis (unilateral or bilateral); granular-type infiltrates; patchy distribution in deeper stroma; later vascularization; conjunctivitis; corneal opacity; uveitis; nystagmus.

Clinical: Vestibuloauditory symptoms (similar to Ménière syndrome); nausea; vomiting; vertigo; tinnitus (abrupt onset); rapidly progressive deafness; loss of equilibration (see Ménière Syndrome); aortic insufficiency; sensorineural testing; lacunar infarcts.

Cheson BD, et al. Cogan's syndrome: a systemic vasculitis. *Am J Med* 1976; 70:549-555.

Cogan DG. Syndrome of non syphilitic interstitial keratitis and vestibuloauditory symptoms. *Arch Ophthalmol* 1945; 33:144.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gilbert WS, Talbot FJ. Cogan's syndrome. *Arch Ophthalmol* 1969; 82:633.

Karni A, et al. Cogan's syndrome complicated by lacunar brain infarcts. *J Neurol Neurosurg Psychiatry* 1991; 54: 169-171.

Oldenski R. Cogan syndrome: autoimmune-mediated audiovestibular symptoms and ocular inflammation. *J Am Board Fam Pract* 1993; 6:577-581.

Cogan (2) Syndrome (Oculomotor Apraxia Syndrome; Wieacker Syndrome) 263

General: X-linked; oculomotor apraxia and muscle atrophy; prevalent in males; corpus callosum can be hypoplastic.

Ocular: Rapid and frequent blinking; conjugate palsy; congenital oculomotor apraxia with patient unable to move eyes voluntarily to one side but with otherwise normal ocular movements; patient fixes objects by head tilt and turning, which causes further ocular deviation via the vestibular reflex; compensation for this overshoot is accomplished by some jerky eye movements with final fixation possible and gradual return of the head to the primary position; may be associated with abnormal electroretinographic responses.

Clinical: Slow progression, predominantly distal muscle atrophy; congenital contracture of feet; dyspraxia of face and tongue muscles; mild mental retardation.

Borchert MS, et al. Congenital oculomotor apraxia: findings with magnetic resonance imaging. *J Clin Neural Ophthalmol* 1987; 7:104.

Cogan DG. Type of congenital ocular motor apraxia presenting jerky head movement. Jackson Memorial Lecture. *Trans Am Acad Ophthalmol Otolaryngol* 1952; 56:853.

Magni R, et al. Electroretinographic findings in congenital oculomotor apraxia (Cogan's syndrome). *Doc Ophthalmol* 1994; 86:259-266.

Vassella F, et al. Cogan's congenital oculomotor apraxia in two successive generations. *Dev Med Child Neurol* 1972; 14:788.

Cogan-Guerry Syndrome (Microcystic Corneal Dystrophy; Map-Dot Fingerprint Dystrophy) 264

General: Etiology obscure; condition benign and asymptomatic; females predominantly affected; ultrastructural studies show discontinuous multilaminar thickened basement membrane under abnormal epithelium; the primary defect appears to be synthesis of abnormal basement membrane and adhesion complexes by the dystrophic epithelium.

Ocular: Reduced vision mainly with involvement of center of cornea; very fine wavy lines resembling fingerprints within or very close to corneal epithelium and best seen on biomicroscopy with retroillumination; fine grayish spheres (0.1- to O.5-mm diameter) in superficial corneal epithelium; maplike irregular border-lined slightly grayish area.

Clinical: None.

Cogan DG, et al. Microcystic dystrophy of the corneal epithelium. *Trans Am Ophthalmol Soc* 1964; 62:213.

Guerry D III. Fingerprint-like lines in the cornea. *Am J Ophthalmol* 1950; 33:724.

Luxenberg MN, et al. Superficial microcystic corneal dystrophy. *Arch Ophthalmol* 1975; 93:107.

Starck T, Hersh PS, Kenyon KR. Corneal dysgeneses, dystrophies and degenerations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. I.* Philadelphia: WB Saunders, 1994:26-30.

Coloboma of Macula (Agenesis of Macula) 265

General: Autosomal dominant; can be caused by intrauterine inflammation, birth hemorrhage; infantile inflammation.

Ocular: Defect in central area of fundus; coloboma can be pigmented, nonpigmented, or have abnormal vessels associated or completely absent; visual defect; absolute central scotoma; nystagmus; myopia; destruction of pigment epithelium; microphthalmos; coloboma of optic nerve (rare); keratoconus; paravenous retinochoroidal atrophy.

Clinical: Microencephaly.

Chen MS, et al. Bilateral macular coloboma and pigmented paravenous retinochoroidal atrophy. *Br J Ophthalmol* 1992; 76:250-251.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Coloboma of Macula with Type B Brachydactyly (Apical Dystrophy) 266

General: Autosomal dominant; bilateral pigmented macular coloboma and brachydactyly.

Ocular: Myopia; retinal detachment; coloboma of retina, choroid, sclera, and macula.

Clinical: Cleft palate; flexion deformity of distal interphalangeal joints of little fingers of hand; retarded growth; delayed sexual maturity; recurrent dislocation of left patella; short feet; coxa valga; genu valgum.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Smith RD, et al. Congenital macula colobomas and short limb skeletal dysplasia. *Am J Med Genet* 1980; 5:365-371.

Sorsby A. Congenital coloboma of macular, together with an account of familial occurrence of bilateral macular coloboma in association with apical dystrophy of hands and feet. *Br J Ophthalmol* 1935; 19:65-90.

Coloboma, Ocular 267

General: Autosomal recessive; congenital or secondary to faulty closure of embryonic fissure.

Ocular: Optic nerve coloboma; retinochoroidal coloboma; orbital cysts; retinal dysplasia; retinal detachment; iris coloboma.

Clinical: White sponge nevus.

Hayasaka S, et al. Typical ocular coloboma affects three generations in one family. *Ann Ophthalmol* 1992; 24: 209-212.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pagon RA, et al. Possible autosomal recessive ocular coloboma. *Am J Med Genet* 1981; 9:189-193.

Wright S, Levy IS. White sponge naevus and ocular coloboma. *Arch Dis Child* 1991; 66:514-516.

Colobomatous, Microphthalmia and Microcornea Syndrome 268

General: Autosomal dominant pattern of inheritance with complete penetrance

Ocular: Bilateral infernasal coloboma; axial enlargement; myopia; iridocorneal angle abnormalities; elevated IOP

Toker E, Elcioglu N, Ozean E, et al. Colomatous macrophthalmia with microcornea syndrome: report of a pedigree. *Am J Med Genet*. 2003; 121: 25-30.

Color Blindness, Blue-Mono-Cone Monochromatic Type 269

General: Sex-linked; progressive.

Ocular: Total to partial color blindness; macular scar; poor central vision; poor color discrimination; infantile nystagmus; nearly normal retinal appearance in most cases; there is evidence showing alterations in the red and green visual pigment gene cluster.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Nathans J, et al. Molecular genetics of human blue cone monochromacy. *Science* 1989; 245:831-838.

Spivey BE. The X-linked recessive inheritance of atypical monochromatis. *Arch Ophthalmol* 1965; 74:327-333.

Color Blindness, Partial, Deutan Series (Deuteranopia) 270

General: Sex-linked; affects males; deuteranopes can have one red pigment gene and one anomalous hybrid red-green gene with a spectral absorbance close to the red pigment gene.

Ocular: Red-green color blindness.

Clinical: None.

Adam A, Fraser GR. The linkage between protan and deutan loci. *Am J Hum Genet* 1970; 22:691-693.

Boger WP III, Petersen RA. Pediatric Ophthalmology. Protan and deutan color blindness. In: Pavan-Langston D, ed. *Manual of Ocular Diagnosis and Therapy*, 4th ed. Boston: Little, Brown and Company, 1995:285-286.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Nathans J, et al. Molecular genetics of inherited variation in human color vision. *Science* 1986; 232:203-210.

Comedo Cataract 271

General: Etiology unknown; possibly in the group of neurectodermal dysplasia syndromes (phakomatoses).

Ocular: Bilateral cataract or both unilateral comedo nevus and cataract; therapy is cataract operation; good prognosis.

Clinical: Bilateral comedo nevus (dermatosis characterized by groups of dilated, keratin-filled follicular spaces).

Mutton P, et al. Comedo naevus: an unusual association of infantile cataract. *Aust Paediatr J* 1975; 11:46-48.

Popov L, Boinaov L. Undescribed congenital cutaneo-ocular syndrome (congenital cataract-comedo syndrome). *Surv Med* 1962; 13:49-50.

Whyte HJ. Unilateral comedo nevus and cataract. *Arch Dermatol* 1968; 97:533-535.

Computer User Syndrome 272

General: Seen in people who use computers extensively.

Ocular: Ocular pain; asthenopia; excyclotorsion; depression of gaze; ocular synkinesis.

Clinical: Hand-wrist pronation; ulnar abduction; headaches; fatigue; various types of head and shoulder distress; carpal tunnel syndrome.

Bernbaum MH. Symposium on near point visual stress. *Am J Optom Physiol Opt* 1986; 62:361.

Grant AH. The computer users syndrome. *J Am Optom Assoc* 1987; 58:892-901.

Tsubota K, Nakamori K. Dry eyes and video display terminals [Letter]. *N Engl J Med* 1993; 328:584.

Cone Dysfunction Syndrome (Achromatopsia) 273

General: Male-linked recessive inheritance; condition is stagnant and nonprogressive; all modes of inheritance have been reported as well as many sporadic cases.

Ocular: Nystagmus; vision decreased 20/50 to 20/200 or less with no or reduced color vision; color vision might be affected with or without amblyopia; peripheral field loss if rods and cones are involved; photophobia; general fundus lesions, mainly macular involvement with depigmentation and degenerative changes; decreased central vision; difficulty adjusting from light to dark environment.

Clinical: Head movements.

Boger WP, Petersen RA. Achromatopsia. In: Pavan-Langston D, ed. *Manual of Ocular Diagnosis and Therapy*, 4th ed. Boston: Little, Brown and Company, 1995:283.

Goodman G, et al. Cone dysfunction syndromes. *Arch Ophthalmol* 1963; 70:214.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

O'Connor PS, Tredici TJ, Ivan DJ, et al. Achromatopsia. Clinical diagnosis and treatment. *J Clin Neuro-Ophthalmol* 1982; 2:219-226.

Reichel E. Hereditary cone dysfunction. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:1238-1248.

Schiefer U, Kurtenbach A, Braun E, et al. Centrally tinted contact lenses. A useful visual aid for patients with achromatopsia. *Ger J Ophthalmol* 1995; 4:52-56.

Zweifach PH, Wolf E. Acquired cone dysfunction and other photopic system diseases. *Arch Ophthalmol* 1968; 79: 18.

Cone-Rod Dystrophy (CRD) 274

General: Autosomal dominant; retinal dystrophy of photoreceptors, characterized by abiotrophic degeneration of rods and cones; onset before age 10 years; it has been suggested that a locus for cone-rod dystrophy may be located in the segment 18q21.1-q21.3 and 19q.

Ocular: Decreased central vision with progressive constriction of peripheral visual fields; degeneration of rods and cones.

Clinical: None.

Ferrell RE, et al. Autosomal dominant cone-rod dystrophy: a linkage study with 17 biochemical and serological markers. *Am J Med Genet* 1981; 8:363-369.

Hittner HM, et al. Dominant cone-rod dystrophy. *Doc Ophthalmol* 1975; 39:29-52.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Warburg M, et al. Deletion mapping of a retinal cone-rod dystrophy: assignment to 18q211. *Am J Med Genet* 1991; 39:288-293.

Congenital Cataract and Hypertrophic Cardiomyopathy Syndrome 275

General: Autosomal recessive; characterized by congenital cataract, hypertrophic cardiomyopathy, mitochondrial myopathy of voluntary muscles, and exercise-related lactic acidosis.

Ocular: Cataract; hyperplastic primary vitreous; aniridia; iris colobomas; microphthalmos; nystagmus; strabismus; myopia; keratoconus.

Clinical: Pulmonary stenosis; ventricular septal defects; structurally abnormal mito-chondria.

Cruysberg JRM, et al. Features of syndrome with congenital cataract and hypertrophic cardiomyopathy. *Am J Ophthalmol* 1986; 102:740-749.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Congenital Cataracts Facial Dysmorphism Neuropathy Syndrome 276

General: Autosomal recessive; motor and sensory neuropathy.

Ocular: Congenital cataracts, microcorneas, strabismus, pendular nystagmus, bilateral blepharoptosis.

Clinical: Patients are recognized in infancy by the presence of congenital cataracts and microcorneas; initially, a predominantly motor neuropathy begins in the lower limbs followed by upper limb involvement; severe disability occurs by the third decade; short stature, moderate nonprogressive cognitive deficits, pyramidal signs, and mild chorea are characteristic.

Mullner-Eidenbock A, Moser E, Klebermass N, et al. Ocular features of the congenital cataracts facial dysmorphism neuropathy syndrome. *Ophthalmology* 2004; 111: 1415-23.

Tournev I, et al. Congenital cataracts facial dysmorphism neuropathy syndrome, a novel complex genetic disease in Balkan Gypsies: *Clinical* and electrophysiological observations. *Am J Ophthalmol* 1999; 128:536-537.

Congenital Cataract, Microcornea, Abnormal Irides, Nystagmus, and Congenital Glaucoma Syndrome 277

General: Autosomal dominant.

Ocular: Microphakia; cataract with two concentric disks, with the anterior being swollen; microphthalmos; microcornea; nystagmus; congenital glaucoma; honey-colored iris with absence of pattern; peripheral anterior synechiae; pupillary abnormality; corneal edema; posterior synechiae; vitreous hemorrhage; shallow anterior chamber; vitreous loss; corneal staphyloma; keratoconus.

Clinical: High arched palate; increased webbing of fingers and toes; deafness.

Cebon L, West RH. A syndrome involving congenital cataracts of unusual morphology, microcornea, abnormal irides, nystagmus, and congenital glaucoma, inherited as an autosomal dominant trait. *Aust J Ophthalmol* 1982; 10: 237-242.

Henkind P, Friedman AH. Iridogoniodysgenesis with cataract. *Am J Ophthalmol* 1971; 72:949-954.

Congenital Cataract with Oxycephaly (Tower Skull Syndrome) 278

General: Autosomal dominant; craniostenosis.

Ocular: Congenital cataracts; keratoconus.

Clinical: Large fontanelles; deformed skull; dwarfing; osteopetrosis.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Roy FH. *Ocular Differential Diagnosis*, 7th ed. Philadelphia: Lippincott Williams & Wilkins, 2002.

Congenital Dyslexia Syndrome (Developmental Dyslexia of Critchley; Congenital Word Blindness of Hermann; Primary Dyslexia; Dyslexia Syndrome; Minimal Brain Dysfunction Syndrome; Attention Deficit Disorder; Congenital Word Blindness) 279

General: Primary reading disability in children with an average or above-average intelligence; male preponderance; dysfunction of the dominating parietotemporal lobe; Levinson postulates a primary cerebellar-vestibular (inner ear) dysfunction underlying this syndrome resulting in a secondary scrambled sensory input and motor output.

Ocular: No obvious connection seems to exist between coordination of ocular functions and dyslexia, although associated ocular findings may exist in reading problems (e.g., abnormal optokinetic nystagmus, metamorphopsia, defective color vision, convergence insufficiency, muscle imbalance, refractive errors); low accommodative converge/accommodation associated with decreased visual acuity and contrast sensitivity.

Clinical: General clumsiness; disorientation (time-space, right-left); behavioral changes; lack of integration of visual and auditory stimuli.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hermann K. Congenital word-blindness: poor readers in the light of Gerstmann's syndrome. *Acta Psychiatr Neurol Suppl* 1956; 103:117.

Hermann K. *Reading disability.* Springfield, IL: Charles C. Thomas, 1959.

Levinson HN. Abnormal optokinetic and perceptual span parameters in cerebellar-vestibular dysfunction and learning disabilities or dyslexia. *Percept Mot Skills* 1989; 68:35-54.

Preston MS. Visual evoked responses vs normal and disabled readers. *Psyche-physiology* 1974; 11:452.

Congenital Epiblepharon Inferior Oblique Insufficiency Syndrome 280

General: Prognosis is good with treatment; present in infancy; inversion of lash line occurs with epiblepharon and is exaggerated by the inferior oblique insufficiency.

Ocular: Narrow interpupillary distance; some ocular prominence; epicanthus; epiblepharon exaggerated in downward gaze; spastic entropion with retroflexion of the eyelashes; epiblepharon becomes less pronounced with growth and development; usually bilateral but in some cases asymmetrical; inferior oblique insufficiency usually unilateral; persistent unilateral keratoconjunctival irritation by the inverted cilia; lacrimation due to conjunctival and corneal irritation.

Clinical: Chubby cheeks (occasionally).

Duke-Elder S, ed. *System of Ophthalmology*, vol. III part 2. St. Louis: CV Mosby, 1976.

Swan KC. The syndrome of congenital epiblepharon and inferior oblique insufficiency. *Am J Ophthalmol* 1954; 39:130.

Congenital Heart Disease 281

General: Represents a wide variety of cardiac diseases or defects.

Ocular: Dilated, tortuous conjunctival vessels; tortuous retinal vessels; retinal edema; papilledema; retinal arterial macroaneurysm; Duane retraction syndrome; double elevator palsy.

Clinical: Hypoxia; increased cerebrospinal fluid; features vary because of a wide variety of cardiac diseases and defects.

Caltrider ND, et al. Retinal emboli in patients with mitral valve prolapse. *Am J Ophthalmol* 1980; 90:534-539.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

el-Asrar AM, et al. Retinal arterial macroaneurysm in a patient with congenital heart disease. *Br J Ophthalmol* 1993; 77:606-607.

Verma MJ, Faridi MM. Ocular motility disturbances (Duane retraction syndrome and double elevator palsy) with congenital heart disease, a rare association with Goldenhar syndrome-a case report. *Ind J Ophthalmol* 1992; 40: 61-62.

Congenital Hereditary Retinoschisis (CHRS; Juvenile X-Linked Retinoschisis) 282

General: X-linked recessive; bilateral; develops early in life but often stabilized toward the end of the second decade; severity varies widely.

Ocular: Bilateral vitreoretinal dystrophy; retinoschisis; vitreous veil; vitreous detachment; vitreous hemorrhage; decreased visual fields; maculopathy; cataract; neovascular glaucoma; vitreoretinopathy; proliferative retinal detachment; Mizuo phenomenon.

Brownstein S, et al. Nonglaucomatous cavernous degeneration of the optic nerve. *Arch Ophthalmol* 1980; 98: 354-358.

Condon GP, et al. Congenital hereditary (Juvenile X-linked) retinoschisis. *Arch Ophthalmol* 1986; 104:576-584.

de Jong PT, et al. Mizuo phenomenon in X-linked retinoschisis. Pathogenesis of the Mizuo phenomenon. *Arch Ophthalmol* 1991; 109:1104-1108.

Manschot WA. Pathology of hereditary juvenile retinoschisis. *Arch Ophthalmol* 1972; 88:131-138.

Regillo CD, et al. Surgical management of complications associated with X-linked retinoschisis. *Arch Ophthalmol* 1993; 111:1080-1086.

Congenital Lues (Congenital Syphilis) 283

General: Caused by intrauterine transplacental infection of fetus by *Treponema pallidum* (see Syphilis).

Ocular: Conjunctivitis; keratitis; dacryocystitis; optic nerve atrophy; periostitis; anisocoria; Argyll Robertson pupil; retinal degeneration; nystagmus; gumma of conjunctiva, eyelids, and orbit; paresis of extraocular muscles; secondary glaucoma; uveitis; iridoschisis.

Clinical: Cutaneous and mucous membrane lesions; periostitis; anemia; hepatosplenomegaly; ectodermal defects; central nervous system involvement; gummatous lesions.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Foss AJ, et al. Interstitial keratitis and iridoschisis in congenital syphilis. *J Clin Neuro-Ophthalmol* 1992; 12:167-170.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Salvador F, et al. Unilateral iridoschisis associated with syphilitic interstitial keratitis and glaucoma. *Ann Ophthalmol* 1993; 25:328-329.

Congenital Retinal Nonattachment 284

General: Autosomal dominant or recessive; acquired prenatally or perinatally; retinal dysplasia typical; x-ray irradiation has been reported to cause retinal nonattachment.

Ocular: Vascularized mass behind the lens; malformation of the chamber angle and elongation of the ciliary processes; retinal dysplasia.

Clinical: Severe fragility of the bones.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Warburg M. Heterogeneity of congenital retinal non-attachment falciform folds and retinal dysplasia: a guide to genetic counseling. *Hum Hered* 1976; 26:137-148.

Congenital Retinal Nonattachment with Mental Retardation, Osteoporosis, and Hypotonia 285

General: Autosomal recessive; well-demarcated entity; affects both males and females.

Ocular: Retinoblastoma; retina nonattached.

Clinical: Mentally retarded; osteoporosis; hypotonia; ligamentous laxity; dwarfism; microcephaly.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Warburg M. Heterogeneity of congenital retinal non-attachment falciform folds and retinal dysplasia: a guide to genetic counseling. *Hum Hered* 1976; 26:137-148.

Congenital Spherocytic Anemia (Congenital Hemolytic Jaundice; Hereditary Spherocytosis) 286

General: Hereditary deficiency of erythrocyte glucose-6-phosphate after exposure to certain drugs, chemicals, and foods such as fava beans.

Ocular: Congenital cataract; ring-shaped pigmentary deposits of cornea; tortuosity of retinal vessels; mongoloid palpebral aperture; microphthalmos.

Clinical: Leukemia; anemia.

Dalgleish R. Ring-like corneal deposit in a case of congenital spherocytosis. *Br J Ophthalmol* 1965; 49:40.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Congenital Tilted Disk Syndrome 287

General: Defective closure of embryonic cleft; disk appears tilted downward and nasal; affects both sexes; central vision is not affected; field defect improves with myopic correction.

Ocular: Myopia; astigmatism; coloboma; situs inversus; posterior inferonasal staphyloma; superotemporal, bitemporal, or superior altitudinal visual field defects; inferior fundus pallor; central retinal vein occlusion; hypoplastic retina and choroid; macular choroidal neovascularization.

Clinical: None.

Apple DJ, et al. Congenital anomalies of the optic disk. *Surv Ophthalmol* 1982; 27:3.

Giuffre G. Chorioretinal degenerative changes in the tilted disc syndrome. *Int Ophthalmol* 1991; 15:1-7.

Giuffre G. Tilted discs and central retinal vein occlusion. *Graefes Arch Clin Exp Ophthalmol* 1993; 231:41-42.

Stur M. Congenital tilted disk syndrome associated with parafoveal subretinal neovascularization. *Am J Ophthalmol* 1988; 105:98-99.

Congential Varicella Syndrome 288

General: Varicella passed in utero from mother to fetus

Ocular: Microphthalmia; microcornea; persistant hyperplastic primary vitreous

Clinical: Urinary tract infection; neurogenic bladder

Fujita H, Yoshii A, Maeda J, et al. Genitourinary anomaly in congenital varicella syndrome: case report and review. Pediatr Nephrol. 2004; 19: 554-7.

Congenital Vertical Retraction Syndrome 289

Aberrant regeneration of the oculomotor nerve; concurrent protective eyelid closure; congenital alterations in the extraocular muscle, its insertion, and its peripheral innervation; nystagmus retractorius; surgical or traumatic rearrangement of orbital structures may account for retraction.

Khodadoust AA, von Noorden GK. Bilateral vertical retraction syndrome: a family study. *Arch Ophthalmol* 1967; 78: 606-612.

Osher RH, et al. Acquired orbital retraction syndrome. *Arch Ophthalmol* 1980; 98: 1798-1802.

Pesando P. Vertical retraction syndrome. *Ophthalmologica* 1978; 177:254-259.

Conjunctivitis, Ligneous 290

General: Autosomal recessive; palpebral conjunctiva becomes the site of dense woody membrane that has global shape; associated with systemic use of tranexamic acid.

Ocular: Corneal scarring; dense membrane of the conjunctiva; may occur as a complication following strabismus surgery.

Clinical: None.

Bierly JR, et al. Ligneous conjunctivitis as a complication following strabismus surgery. *J Pediatr Ophthalmol Strabismus* 1994; 31:99-103.

Diamond JP, et al. Tranexamic acid-associated ligneous conjunctivitis with gingival and peritoneal lesions. *Br J Ophthalmol* 1991; 75:753-754.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pavan-Langston D. Cornea and external disease. Ligneous conjunctivitis. In: Pavan-Langston D, ed. *Manual of Ocular Diagnosis and Therapy*, 4th ed. Boston: Little, Brown and Company, 1995:103.

Conradi Syndrome (Multiple Epiphyseal Dysplasia Congenita; Dysplasia Epiphysealis Congenita; Chondrodystrophia Foetalis Hypoplastica; Calcinosis Universalis; Congenital Calcifying Chondrodystrophy; Stippled Epiphyses Syndrome; Conradi-Hünermann Syndrome; Chondrodysplasia Punctata) 291

General: Autosomal recessive; manifestations within the first 6 months of life; epiphyseal stippling present at birth; perinatal manifestations include disorganization of the spine, premature echogenicity of femoral epiphyses, and frontal bossing with depressed nasal bridge.

Ocular: Hypertelorism; heterochromia iridis (rare); bilateral total congenital cataract appearing at or shortly after birth; primary optic atrophy (rare); bilateral corneal punctate erosions.

Clinical: Short limbs (mainly proximal part) resulting in "short-limbed dwarfism"; deformities of hip, knee, and elbow joints by contraction and immobility, and possible transformation of muscles into fibrous tissue as a result; congenital heart defect with calcium deposits in the cardiac valves; skin anomalies (dyskeratosis); mental retardation.

Bellson FA. Optic nerve hypoplasia in chondrodysplasia punctata. *J Pediatr Ophthalmol* 1977; 14:144-147.

Conradi E. Vorzeitiges Auftreten von Knochenund Eigenartigen Verkalkungskernen bei Chondrodystrophia Fotalis Hypoplastica. Histologische un Rontgenuntersuchungen. *Jahrb Kinderhk* 1914; 80:86.

Happle R. Cataracts as a marker of genetic heterogeneity in chondrodysplasia punctata. *Clin Genet* 1981; 19:64-66.

Massey JY, Roy FH. Ocular manifestations of Conradi disease. *Arch Ophthalmol* 1974; 92:524.

Pryde PG, et al. Prenatal diagnosis of nonrhizomelic chondrodysplasia punctata (Conradi-Hunermann syndrome). *Am J Med Genet* 1993; 47:426-431.

Spierer A, Neumann D. Corneal changes in chondrodysplasia punctata syndrome. *Ann Ophthalmol* 1993; 25: 356-358.

Contact Dermatitis (Dermatitis Venenata) 292

General: Reaction of skin due to contact with foreign material; inflammatory disorder of the skin that may result from immunologic hypersensitivity (allergic contact dermatitis) or cutaneous injury not involving immunologic mechanisms (irritant contact dermatitis) from offending topical agents.

Ocular: Keratoconjunctivitis; chemosis; leukoma; corneal ulcer; pruritus of lids.

Clinical: Dermatitis; itching, erythema; vesiculation; edema with weeping and crusting.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Granstein RD. Skin and mucous membrane disorders. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3102-3120.

Korting GW. *The skin and the eye: a dermatologic correlation of diseases of the periorbital region*. Philadelphia: WB Saunders, 1973:7-15.

Lowney ED. Contact dermatitis. In: Moschella SL, Hurley HJ, eds. *Dermatology.* Philadelphia: WB Saunders, 1975: 239-258.

Convergence Insufficiency Syndrome (Asthenovergence of Stutterheim) 293

General: An exodeviation that is greater at near distances than at far ones; inadequate accommodative and fusional convergence impulses; prevalence is generally considered to be low in children under age 10 years and higher in females.

Ocular: Burning, itching, blurred vision; diplopia; difficulty in following moving objects; astigmatism; decreased visual acuity; exotropia; hypermetropia; orthotropia.

Clinical: Headache; associated with thyroid eye disease.

Burke JP, et al. Convergence insufficiency in thyroid eye disease. *J Pediatr Ophthalmol Strabismus* 1993; 30:127-129.

Fraunfelder FT, Roy FJ. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Harley RD, ed. *Pediatric Ophthalmology,* 4th ed. Philadelphia: WB Saunders, 1998.

Norm MS, Convergence insufficiency: incidence in ophthalmic practice. Results of orthoptic treatment. *Acta Ophthalmol* 1966; 44:132.

Cooley Anemia (Thalassemia; Thalassemia Major; Thalassemia Minor) 294

General: Autosomal dominant in synthesis of the □ or □ chain of hemoglobin; most prevalent in Mediterranean and Oriental populations.

Ocular: Retinal hemorrhages; angioid streaks; macular vascular abnormalities; pigmented chorioretinal scars (black sunbursts); occlusion of peripheral retinal arteries; vitreous hemorrhages.

Clinical: Hemolytic anemia; hypochromic anemia.

Condon PI, Serjeant GR. Ocular findings in sickle cell thalassemia in Jamaica. *Am J Ophthalmol* 1972; 74: 1105-1109,

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Goldberg MF. Ophthalmologic manifestations of sickle cell thalassemia. *Arch Intern Med* 1971; 128:33-39.

O'Donnell BF, et al. Angioid streaks in beta thalassemia minor. *Br J Ophthalmol* 1991; 75:639,

Rinaldi M, et al. Ocular involvement correlated with age in patients affected by major and intermedia beta-thalassemia treated or not with desferrioxamine. *Metab Pediatr Syst Ophthalmol* 1993; 16:23-25.

Cornea Plana 295

General: Autosomal dominant; may be inherited as autosomal dominant or recessive.

Ocular: Hyperopia; hazy corneal limbus; opacities in corneal parenchyma and marked arcus; posterior embryotoxon; iris and lens abnormalities.

Clinical: Associated with epidermolysis bullosa dystrophica.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Hemady RK, et al. Duplication of the lens, hourglass cornea, and cornea plana. *Arch Ophthalmol* 1993; 111:303,

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Sharkey JA, et al. Cornea plana and sclerocornea in association with recessive epidermolysis bullosa dystrophica. Case report. *Cornea* 1992; 11:83-85.

Corneal Crystals, Myopathy, and Nephropathy 296

General: Etiology unknown; may represent an atypical variant of myotonic dystrophy.

Ocular: Retinal pigment epithelial mottling; nystagmus; deep corneal crystals; conjunctival crystals.

Clinical: Weakness and atrophy of pharyngeal, facial, and intrinsic hand muscles; decreased hearing; hypertension; chronic renal disease with decreased glomerular filtration and proteinuria; asymmetrical smile; diminished gag reflex; chronic serous otitis media.

Arnold RW Corneal crystals, myopathy and nephropathy: a new syndrome? *J Pediatr Ophthalmol Strabismus* 1987; 24:151-155.

Reiss GR, et al. Infectious crystalline keratopathy. *Surv Ophthalmol* 1986; 31:69-72.

Corneal Dystrophy, Granular Type (Groenouw Type I Corneal Dystrophy) 297

General: Autosomal dominant; hyaline degeneration with absence of acid mucopolysaccharide deposition; autosomal dominant with complete penetrance; evidence links it with chromosome 5q; Avellino dystrophy is a variant of granular corneal dystrophy with lattice changes.

Ocular: Grayish-white granules in a disk-shaped area of central cornea; hyaline material separates epithelium from Bowman membrane; keratoconus.

Clinical: None.

Eiberg H, et al. Assignment of granular corneal dystrophy Groenouw type I to chromosome 5q. *Eur J Hum Genet* 1994; 2:132-138.

Holland EJ, et al. Avellino corneal dystrophy. Clinical manifestations and natural history. *Ophthalmology* 1992; 99: 1564-1568.

Jones SF, Zimmerman LE. Histopathologic differentiation of granular macular and lattice dystrophies of cornea. *Am J Ophthalmol* 1961; 51:394-410.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Moller HU Granular corneal dystrophy Groenouw type I. Clinical and genetic aspects. *Acta Ophthalmol Suppl* 1991; 198:1-40.

Corneal Dystrophy, Hereditary Polymorphous Posterior (PCD) 298

General: Autosomal dominant.

Ocular: Clouding of posterior cornea; reduced number of endothelial cells; thickening and opacities of Descemet membrane; associated with keratoconus; iridocorneal adhesions; glassy membranes; pupillary ectropion (rare).

Clinical: None.

Driver PJ, et al. Familial cases of keratoconus associated with posterior polymorphous dystrophy. *Am J Ophthalmol* 1994; 118:256-257.

Laganowski HC, et al. Distinguishing features of the iridocorneal endothelial syndrome and posterior polymorphous dystrophy: value of endothelial specular microscopy. *Br J Ophthalmol* 1991; 75:212-216.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Rodrigues MM, et al. Posterior polymorphous corneal dystrophy: recent developments. *Birth Defects* 1982; 18: 479-491.

Sekundo W, et al. An ultrastructural investigation of an early manifestation of the posterior polymorphous dystrophy of the cornea. *Ophthalmology* 1994; 101:1422-1431.

Corneal Dystrophy, Lattice Type (Lattice Corneal Dystrophy; LCD; Lattice Dystrophy Type I; Biber-Haab-Dimmer Dystrophy) 299

General: Autosomal dominant; progression to severe visual impairment by fifth or sixth decade.

Ocular: Grayish lines between the centers of cornea and periphery; rounded dots scattered over the cornea; elongated deposits that form reticular pattern in corneal stoma; keratoconus.

Clinical: Secondary form of inherited localized amyloidosis inherited as an autosomal dominant trait with low penetrance.

Kivlin JD, et al. Linkage analysis in lattice corneal dystrophy. *Am J Med Genet* 1984; 19:387-390.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pavan-Langston D. Lattice dystrophy (Biber-Haab-Dimmer). In: Pavan-Langston D, ed. *Manual of Ocular Diagnosis and Therapy*. 4th ed. Boston: Little, Brown and Company, 1995: 107.

Corneal Dystrophy, Macular Type (Groenouw Type II Corneal Dystrophy) 300

General: Autosomal recessive; onset in first decade, between 5 and 9 years; progressive; acid mucopolysaccharides found in corneal fibroblasts; it has been suggested that the defect may not be limited to the cornea.

Ocular: Minute gray, punctate opacities; reduced corneal sensitivity; photophobia; foreign body sensations; recurrent corneal erosions; keratoconus.

Clinical: Defect in metabolism of glycoprotein processing.

Klintworth GK, Smith CF. Macular corneal dystrophy: studies of sulfated glycosaminoglycans in corneal explant and confluent stromal cell cultures. *Am J Pathol* 1977; 89:167-182.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Merin S. Cornea: macular corneal dystrophy. In: Merin S, ed. *Inherited eye diseases.* New York: Marcel Dekker, 1991:29.

Corneal Dystrophy, Meesmann Epithelial (Meesmann Epithelial Dystrophy of Cornea) 301

General: Autosomal dominant; rare; onset first year of life; possible that a disturbance of the cytoplasmic ground substance results in cellular homogenization with cyst formation.

Ocular: Myriads of fine punctate opacities in epithelium and Bowman membrane of cornea; thickening of the epithelial basement membrane of cornea; keratoconus.

Clinical: None.

Fine BS, et al. Meesmann's corneal dystrophy: ultrastructural features. *Can J Ophthalmol* 1982; 17:24.

Fine BS, et al. Meesmann's epithelial dystrophy of cornea. *Am J Ophthalmol* 1977; 83:633-642.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Corneal Hypesthesia, Familial 302

General: Autosomal dominant; decreased corneal sensation, reflex tearing, blinking, and foreign body sensation.

Ocular: Punctate epithelial erosions; corneal edema; neurotrophic keratitis; corneal ulcers; poor blinking; decreased tearing; decreased corneal sensation.

Clinical: Trigeminal anesthesia; hypoplastic trigeminal nerves and gasserian ganglia.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Purcell JJ, et al. Familial corneal hypesthesia. *Arch Ophthalmol* 1979; 97:872~874.

Corneal Snowflake Dystrophy 303

General: Autosomal dominant; prevalence of green irides.

Ocular: Star-shaped chromatophore-like cells attached to anterior lens capsule; Bitot spots; white flecks on endothelium and Descemet membrane.

Clinical: Lactose intolerance; malabsorption of fat; vitamin A deficiency; dry skin; nevi; freckles.

Meretoja J. Inherited corneal snowflake dystrophy with oculocutaneous pigmentation disturbances and other symptoms. *Ophthalmologica (Basel)* 1985; 191:197-205.

Meretoja J. Inherited syndrome with corneal snowflake dystrophy, oculocutaneous pigmentary disturbances, pseudoexfoliation and malabsorption. *Ophthalmic Res* 1987; 19:245-254.

Corneo-Dermato-Osseous Syndrome (CDO Syndrome; Corneal Dystrophy, Epithelial with Skin and Skeletal Changes) 304

General: Autosomal dominant; similar lesions of palms, soles, and cornea occur III Richner-Hanhart syndrome.

Ocular: Epithelial and stromal corneal changes; photophobia; keratoconus.

Clinical: Palmoplantar hyperkeratosis; brachydactyly, short stature; premature birth; soft teeth.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Stern JK, et al. Corneal changes, hyperkeratosis, short stature, brachydactyly, and premature birth: a new autosomal dominant syndrome. *Am J Med Genet* 1984; 18:67-77.

Cortical Blindness, Retardation, and Postaxial Polydactyly 305

General: Autosomal recessive; combination of cortical blindness, retardation, and postaxial polydactyly.

Ocular: Cortical blindness.

Clinical: Growth and psychomotor development severely retarded; prominent forehead and short nose.

Hernandez A, et al. Cortical blindness, growth, and psychomotor retardation and postaxial polydactyly: a probably distinct autosomal recessive syndrome. *Clin Genet* 1985; 28:251-254.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Costen Syndrome (Temporomandibular Joint Syndrome) 306

General: Dental malocclusion; overaction of jaw joint followed by the development of a loose joint due to absorption of the meniscus, condyles, and bone.

Ocular: Headaches; facial pain.

Clinical: Severe neuralgia; "full" ear sensation with reduced hearing; tinnitus; dizziness; associated with vertigo.

Chole RA, Parker WS. Tinnitus and vertigo in patients with temporomandibular disorder. *Arch Otolaryngology Head Neck Surg* 1992; 118:817-821.

Costen JB. A syndrome of ear and sinus symptoms dependent upon disturbed function of the temporomandibular joint. *Ann Otol Rhinol Laryngol* 1934; 43:1.

Freese AS. Costen's syndrome: a reinterpretation. *Arch Otolaryngol* 1959; 70:309.

Cowden's Disease 307

General: Autosomal dominant; PTNE gene; tumor suppressor gene

Ocular: Cataract; angoid streaks; myopia; multiple trichilemmomas of eyelids

Clinical: Malignancies of breast, thyroid, ovary, uterus, colon or bladder; seizures; tremor; mental retardation; cerebellar gangliocytoma

Kiratli H, Sonmez B, Bilgic S, et al. Multiple astrocytic hamartoas in bilateral retina. *Can J Ophthalmo*. 2004; 39: 662-3

Sawada T, Okada T, Miwa K, et al. Two novel mutations of PTEN gene in Japanese patients with Cowden's disease. *Am J Genet A* 2004; 128: 12-4.

Cranial Nerves, Congenital Paresis 308

General: Autosomal recessive; damage to peripheral nerve and brainstem.

Ocular: Paralysis of pupillary sphincter; oculomotor synkinesis; ptosis; exotropia; hypotropia; amblyopia; diplopia.

Clinical: Vascular lesion from damage to peripheral nerve and brainstem; generalized developmental delay; seizures; facial paralysis; malformed external ears; abnormalities of motor system of arms and legs.

Balkan R, et al. Associated abnormalities in congenital third nerve palsies. *Am J Ophthalmol* 1984; 97:315-319.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Cranial Nerves, Recurrent Paresis 309

General: Autosomal recessive; rare, recurrent episodes of Bell palsy and external ophthalmoplegia; lack of iridoplegia distinguishes it from aneurysm.

Ocular: External ophthalmoplegia; third nerve palsy.

Clinical: Bell palsy; diabetes; polycythemia.

Currie S. Familial oculomotor palsy with Bell's palsy. *Brain* 1980; 93: 193-198.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Craniocervical Syndrome (Whiplash Injury) 310

General: Disturbed accommodation is due to a central lesion rather than a peripheral lesion of the ciliary muscle; Homer syndrome observed where a palsy of the cervical sympathetics occurs (see Homer Syndrome).

Ocular: General ocular pain; enophthalmos; mild ptosis; reduced ability to accommodate; disturbance in ocular movements; primarily those extraocular muscles innervated by the oculomotor nerve are involved; convergence insufficiency; nystagmus (gaze direction and vestibular, central, peripheral, and mixed type); vestibular impairment in more than 50% of cases; asthenopia; fogging; double vision; miosis; mydriasis; retinal arteriolar pressure may show changes in systolic and diastolic pressure and be more pronounced than changes in the brachial blood pressure; decreased stereoacuity; vitreous detachment.

Clinical: Headache; vertigo; dizziness; neck and back pain.

Bonica JJ. Hyperextension and hyperflexion cervical injuries. *The Management of Pain. vols. I and II.* 2nd ed. Philadelphia: Lea & Febiger, 1990:854-856.

Burke JP, et al. Whiplash and its effect on the visual system. *Graefes Arch Clin Exp Ophthalmol* 1992; 230:335-339.

Evans RW. Some observations on whiplash injuries. *Neural Clin* 1992; 10:975-997.

Narazaki S, et al. The ophthalmic disturbance as the craniocervical syndrome due to whiplash injury. *Jpn J Ophthalmol* 1969; *13:263.*

Wiesinger H, Guerry D III. Augenveranderungen bei "whiplash." *Verletzungen Klin Monatsbl Augenheilkd* 1961; 139:841.

Craniocleidodysostosis Syndrome (Cleidocranial Dysostosis Syndrome; Marie-Sainton Syndrome; Scheuthaurer Syndrome; Hulkcrantz Anosteoplasia; Mutational Dysostosis Syndrome) 311

General: Autosomal dominant; hypoplastic dysostosis of the skull; shows brachycephaly or platycephaly.

Ocular: Proptosis (unilateral); prominent orbital ridges; greater vertical diameter of the orbit compared with horizontal diameter; hypertelorism; antimongoloid palpebral fissure.

Clinical: Saddle nose; prominent forehead (frontal bossing); hypoplasia of facial bones and clavicles; high arched palate and protruding jaw; oligodontia; pathologic fractures; hyperlaxia of joints; kyphoscoliosis and spina bifida; scoliosis; hemiplegia; spastic paraplegia; mental deficiency; psychosis; incomplete closure of fontanelles; dwarfism; epilepsy; hypoplasia/aplasia of nasal bones; high and narrow orbital opening; absent or diminished paranasal sinuses; small sella; skeletal immaturity due to defect in bone remodeling.

Gorlin RJ, Cervenka J. Syndrome of facial clefting. *Scand J Plast Reconstr Surg* 1974; 8: 13-25.

Jensen BL. Cleidocranial dysplasia: craniofacial morphology in adult patients. *J Craniofac Genet Dev Biol* 1994; 14: 163-176.

Marie P, Sainton P. Sur la Dysostose Cleido-Cranienne Hereditaire. *Bull Soc Med Hop Paris 1898; 15:436.*

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Craniofacial, Deafness, Hand Syndrome 312

General: Autosomal dominant.

Ocular: Hypertelorism.

Clinical: Flat facial profile; hypoplastic nose with slitlike nares; hearing loss; ulnar deviation of hands.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Sommer A, et al. Previously undescribed syndrome of craniofacial, hand anomalies, and sensorineural deafness. *Am J Med Genet* 1983; 15:71-77.

Craniofrontonasal Dysplasia 313

General: Sex-linked; unexplained higher prevalence in females.

Ocular: Down-slanting palpebral fissures; hypertelorism.

Clinical: Coronal synostosis; brachycephaly; clefting of the nasal tip; joint anomalies; longitudinally grooved fingernails; other digital anomalies.

Keve J, et al. Craniofrontonasal dysostosis: variable expression in a three-generation family. *Clin Genet* 1992; 38: 441-446.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Reynolds JF, et al. Craniofrontonasal dysplasia: A new family. *Proc Greenwood Genet Center* 1983; 2:115.

Craniometaphyseal Dysplasia Syndrome (Pyle Syndrome; Familial Metaphyseal Dysplasia; Bakwin-Krida Syndrome; Leontiasis Ossea) 314

General: Autosomal recessive; absorption of secondary spongiosa is lacking, with resulting long-bone deformities.

Ocular: Hypertelorism.

Clinical: Splaying of metaphyseal ends of long bones; thick and dense base of bony skull; absent air filling of mastoid process and paranasal sinuses; late dentition; deafness; progressive headache; vomiting; low intelligence; prominent glabella and zygomatic arch; genu valgus deformity.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gupta D, et al. Craniometaphyseal dysplasia. *Australas Radial* 1993; 37:122-125.

Pyle E. A case of unusual bone development. *J Bone Joint Surg* 1931:13:874.

Cranio-Oro-Digital Syndrome (Otopalatodigital Syndrome; OPD II Syndrome; Facio-Palato-Osseous Syndrome; FPO) 315

General: Sex-linked; can occur as a sporadic condition.

Ocular: Downward-slanting palpebral fissures (antimongoloid obliquity).

Clinical: Microcephaly; small mouth; midface hypoplasia; cleft palate; flexed, overlapping fingers with syndactyly of digits 3 and 4; syndactyly of toes 2 and 5; bifid uvula; slight deviation of the terminal phalanges of the third fingers; radial deviation of the terminal phalanx of the right fourth finger; short first toe and long second toe; short first metacarpal; extra bone in the capitate-hamate complex; small thorax; bowed limbs with absent fibula; mild frontal bossing; conductive hearing impairment; flat facies; broad nasal base; wavy irregular clavicles and ribs; widely spaced eyes; prominent forehead.

Brewster TG, et al. Otopalato-digital syndrome, type II: an X-linked skeletal dysplasia. *Am J Med Genet* 1985; 20: 249-254.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Preis S, et al. Oto-palato-digital syndrome type II in two unrelated boys. *Clin Genet* 1994; 45:154-161.

Stoll C, Alembik Y. Oto-palato-digital syndrome type II. *Genet Couns* 1994; 5:61-66.

Craniopharyngioma 316

General: Benign congenital tumors arising from epithelial remnants of Rathke pouch; most common nonglial intracranial tumors in childhood; second most common sellar-parasellar tumor primarily in children or young adults; 35% of cases occur in patients over age 40 years.

Ocular: Paresis of third or sixth nerve; optic nerve atrophy; optic neuritis; papilledema; dilation of pupil; diplopia; hemianopsia; nystagmus; scotoma; visual field defects; visual loss.

Clinical: Hydrocephalus; infantilism; diabetes insipidus; abnormal sexual development; headaches; acute aseptic meningitis.

Amacher AL. Craniopharyngioma: the controversy regarding radiotherapy. *Child Brain* 1980; 6:57-64.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Sweet WH. Radial surgical treatment of craniopharyngioma. *Clin Neurosurg* 1975; 23:52-79.

Craniostenosis 317

General: Skull deformity caused by premature fusion of cranial sutures.

Ocular: Optic atrophy; exophthalmos; strabismus; papilledema; nystagmus; ocular colobomas; swollen optic nerves; dissociated eye movements; ptosis; anisometropia; corneal exposure; amblyopia.

Clinical: Elevated cerebrospinal fluid; abnormal development of the skull.

Cheng H, et al. Dissociated eye movements in craniosynostosis: a hypothesis revived. *Br J Ophthalmol* 1993; 77: 563-568.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hertle RS. Visual loss in patients with craniofacial synostosis. *J Pediatr Ophthalmol Strabismus* 1991; 28: 344-349.

Iannaccone G, Gerlini G. The so-called "cloverleaf skull syndrome." A report of three cases with a discussion of its relationships with thanatophoric dwarfism and the craniostenoses. *Pediatr Radiol* 1974; 2:175-184.

Newman SA. Ophthalmic features of craniosynostosis. *Neurosurg Clin N Am* 1991; 2:587-610.

Craniosynostosis-Mental Retardation-Clefting Syndrome 318

General: Autosomal recessive.

Ocular: Choroidal coloboma.

Clinical: Craniosynostosis; mental retardation; seizures; dysplastic kidneys; bat ears; cleft lip and palate; beaked nose; small posterior fontanelle.

Baraitser M, et al. A new craniosynostosis-mental retardation syndrome. *Clin Genet* 1982; 22:12-15.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Craniotelencephalic Dysplasia 319

General: Autosomal recessive.

Ocular: Optic nerve hypoplasia.

Clinical: Frontal bone protrusion; encephalocele; craniosynostosis; developmental retardation; agenesis of the corpus callosum; lissencephaly; arhinencephaly.

Hughes HE, et al. Craniotelencephalic dysplasia in sisters: further delineation of a possible syndrome. *Am J Med Genet* 1983; 14:557-565.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Cretinism (Hypothyroid Goiter; Hypothyroidism; Juvenile Hypothyroidism; Myxedema) 320

General: Deficient thyroid function.

Ocular: Blepharitis; ptosis; enophthalmos; temporal madarosis; decreased tear secretion; glaucoma; proptosis; optic atrophy; optic neuritis; blue dot cataract; conjunctivitis; scleritis; optic disk hemorrhage and arcuate scotoma associated with glaucoma.

Clinical: Myxedema; larynx and tongue swollen; hoarse speech; dry, yellowish skin; slow pulse; mental retardation; infertility; pericardial effusion; cardiac enlargement; physical development retarded.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Girkin CA, McGwin G, McNeal SF, et al. Hypothyroidism and the development of open-angle glaucoma in a male population. *Ophthalmology* 2004; 111:1649-1652

Mahto RS. Ocular features of hypothyroidism. *Br J Ophthalmol* 1972; 56:546-550.

McLenachan J, Davies DM. Glaucoma and the thyroid. *Br J Ophthalmol* 1965; 49:444.

Smith KD, et al. An ocular dynamic study supporting the hypothesis that hypothyroidism is a treatable cause of secondary open-angle glaucoma. *Can J Ophthalmol* 1992; 27:341-344.

Smith KD, et al. Reversal of poorly controlled glaucoma on diagnosis and treatment of hypothyroidism. *Can J Ophthalmol* 1992; 27:345-347.

Creutzfeldt-Jakob Syndrome (Spastic Pseudosclerosis; Corticostriatospinal Degeneration; Disseminated Encephalopathy; Heidenhaim Syndrome; Presenile Dementia with Spastic Paralysis; Presenile Dementia-Cortical Degeneration Syndrome) 321

General: Heredofamilial occurrence; caused by degenerative changes in cerebral cortex, basal ganglion, and spinal cord; disease is progressive; begins in middle or later age; occurs in both sexes.

Ocular: Cortical blindness; myoclonic conjugate eye movements; paralysis of seventh nerve; ptosis; dyschromatopsia; homonymous hemianopsia; nystagmus; slow vertical saccades; mild demyelination of the optic nerve.

Clinical: Mental deterioration; psychosis; stupor; weakness and stiffness of extremities; slow development of pyramidal signs; loss of reflexes; tremor, rigidity; dysarthria; aphasia; ataxia; myoclonus; convulsive seizures; cerebellar ataxia decerebrate posture.

Creutzfeldt HG. Uber Fine Eigenartige Herdformige Erkrankung des Zentralnervensystems. In: Nissl F, Alzheimer A, eds. *Histologie und Histopathologie.* Jena: Fischer, Arbeit Erganzungs-band, 1921; and Z *Ges Neurol Psychiat* 1920; 57:1.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Grant MP, et al. Abnormal eye movements in Creutzfeldt-Jakob disease. *Ann Neurol* 1993; 34:192-197.

Jacob A. Uber Eine Eigenartige Erkrankung des Zentralnervensystems mit Bemerkenswertem Anatomischen Befund. Z *Ges Neural Psychiatr* 1921; 64:147.

Roos R, et al. The *Clinical* characteristics of transmissible Creutzfeldt-Jakob disease. *Brain* 1973; 96:1-20.

Cri-du-Chat Syndrome (Cat-Cry (5p-) Syndrome; Crying Cat Syndrome; BI Deletion Syndrome; Lejeune Syndrome) 322

General: Short arm deletion of a no. 5 chromosome (5p-); increased inheritance risk; 13% have one parent with balanced translocation; female preponderance 2:1 (see Wolf Syndrome).

Ocular: Hypertelorism; epicanthal folds; antimongoloid slanting of palpebral fissures; strabismus; increased tortuosity of retinal vessels.

Clinical: High-pitched, plaintive cry by an infant (reminiscent of a crying cat); mental retardation; broad nasal root; micrognathia or retrognathia; low-set ears; simian crease; congenital heart defect; small larynx and epiglottis.

Bateman JB. Chromosomal anomalies and the eye. In: Wright KW; ed. *Pediatric Ophthalmology and strabismus.* St. Louis: Mosby, 1995:595.

Howard RD. Ocular abnormalities in the cri-du-chat syndrome. *Am J Ophthalmol* 1972; 73:939.

Lejeune J, et al. Trois cas de Deletion Partielle du Bras Court d'un Chromosome 5. *C R Acad* Sci *(Paris)* 1963; 257: 3098.

Wilkins LE, et al. Clinical heterogeneity in 80 home-reared children with cri-du-chat syndrome. *J Pediatr* 1983; 102: 528-533.

Criswick-Schepens Syndrome (Familial Exudative Vitreoretinopathy) 323

General: Familial exudative vitreoretinopathy, similar to retrolental fibroplasia; bilateral; slowly progressive; full-term babies; no oxygen therapy; autosomal dominant; may be inherited as X-linked or autosomal dominant condition.

Ocular: Posterior vitreous detachment of organized membranes of vitreous; snowflake-like opacities of vitreous; heterotropia of macula; subretinal exudates; retinal detachment; degenerative retinal changes; retinal hemorrhage; retinal folds; enophthalmos; phthisis; intraretinal exudate; vitreous hemorrhage; amblyopia; falciform retinal fold.

Clinical: Normal general development; normal birth weight.

Brockhurst R, et al. Uveitis. I. Gonioscopy. *Am J Ophthalmol* 1956; 42:545.

Criswick VG, Schepens CL. Familial exudative vitreoretinopathy. *Am J Ophthalmol* 1969; 68:578-594.

Ebert EM, Mukai S. Familial exudative vitreoretinopathy. *Int Ophthalmol Clin* 1993; 33:237-247.

Fullwood P, et al. X-linked exudative vitreoretinopathy: *Clinical* features and genetic linkage analysis. *Br J Ophthalmol* 1993; 77:168-170.

Nicholson DH, Galvis V. Criswick-Schepens syndrome. *Arch Ophthalmol* 1984; 102:1519-1522.

Plager DA, et al. X-linked recessive familial exudative vitreoretinopathy. *Am J Ophthalmol* 1992; 114:145-148.

Van Nouhuys CE. Signs, complications, and platelet aggregation in familial exudative vitreoretinopathy. *Am J Ophthalmol* 199l; 111:34-41.

Crohn Disease (Granulomatous Ileocolitis) 324

General: Autoimmune or hypersensitivity inflammatory change; slight prevalence in males; Jewish people most frequently affected; onset at any age; more severe in young people; remission; relapses.

Ocular: Recurrent conjunctivitis; marginal corneal ulcers; keratitis; blepharitis; dry eye; scleritis; episcleritis; iris atrophy; uveitis; pupil immobility and dilatation; macular edema; macular hemorrhages; extraocular muscles palsy; vitreal haze; retinal vasculitis; subconjunctival nodules; conjunctival ulcer; pannus; acute dacryoadenitis; orbital pseudotumor.

Clinical: Inflammatory bowel disease; abdominal distention; tenderness of abdomen; mass in right lower quadrant of abdomen; diarrhea; abdominal cramps; bloating; flatulence; weight loss; nervousness; tension; depression; pyoderma gangrenosum.

Duker JS, et al. Retinal vasculitis in Crohn's disease. *Am J Ophthalmol* 1987; 103:664-668.

Dutt S, et al. Acute dacryoadenitis and Crohn's disease: findings and management. *Ophthal Plast Reconstr Surg* 1992; 8:295-299.

Ellis PP, Gentry JH. Ocular complications of ulcerative colitis. *Am J Ophthalmol* 1964; 58:779-784.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Grewal RK, et al. Subconjunctival nodules: an unusual ocular complication of Crohn's disease. *Can J Ophthalmol* 1994; 29:238-239.

Hegab SM, al-Mutawa SA. Conjunctival ulcer in a patient with Crohn's disease. *Ophthalmic Surg* 1994; 25:638-639.

Knox DL, et al. Primary, secondary and coincidental ocular complications of Crohn's disease. *Ophthalmology* 1984; 91:163-173.

Knox DL, et al. The keratopathy of Crohn's Disease. *Am J Ophthalmol* 1980; 90:862-865.

Macoul KL. Ocular changes in granulomatous ileocolitis. *Arch Ophthalmol* 1970; 84:95-97.

Tierney LM. Crohn's disease. In: Tierney LM, ed. *Current Medical Diagnosis and Treatment*, 33rd annual revision. Stamford: Appleton and Lange, 1994:518-521.

Crome Syndrome 325

General: Fatal; death usually at 4 to 8 months; autosomal recessive.

Ocular: Congenital cataracts.

Clinical: Epileptic seizures; mental retardation; small stature; renal tubular necrosis; encephalopathy.

Crome L, et al. Congenital cataracts, renal tubular necrosis and encephalopathy in two sisters. *Arch Dis Child* 1963; 38:505-515.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Cronkhite-Canada Syndrome 326

General: Hypoproteinemia; hypocalcemia

Ocular: Nuclear and posterior subcapsular cataract

Clinical: Gastrointestinal polyposis; hyperpigmentation of skin; hair loss; nail atrophy

Goto A. Cronkhite-Canada syndrome: observation of the 180 cases reported in Japan. *Nippon Rinsho* 1991; 49: 221-226

Simcock PR, Zamarakji HJ, Muller BR: Cataract formation in the Cronkhite-Canada syndrome. *J Cataract Refract Surg* 1996; 22: 1125-1126.

Crouzon Syndrome (Dysostosis Craniofacialis; Oxycephaly; Craniofacial Dysostosis; Parrot-Head Syndrome; Möbius-Crouzon Syndrome; Hereditary Craniofacial Dysostosis) 327

General: Autosomal dominant; manifestations present at birth.

Ocular: Bilateral exophthalmos; hypertelorism (wide interpupillary distance); obliquity of palpebral fissures with outer canthus slanting downward; nystagmus; exotropia; upper field defects due to pressure upon the optic nerve on its lower part; bluish sclera; exposure keratitis in extreme exophthalmos; cataract; papilledema; secondary optic atrophy; corneal dystrophy; ptosis; strabismus; keratoconus.

Clinical: Prognathism; maxillary hypoplasia with short upper lip; synostosis of coronal and lambda sutures; parrot-beaked nose (psittachosrhina); widening temporal fossae; headaches.

David DJ, Sheen R. Surgical correction of Crouzon syndrome. *Plast Reconstr Surg* 1990; 85:344-354.

Gray TL, Casey T, Selva D, et al. Ophthalmic sequelae of Crouzon syndrome. *Ophthalmology* 2005; 112: 1129-1134.

Krause AC, Buchanan DN. Dysostosis craniofacialis (Crouzon). *Am J Ophthalmol* 1959; 22:140.

Kreiborg S, Aduss H. Pre- and postsurgical facial growth in patients with Crouzon's and Apert's syndromes. *Cleft Palate J* 1986; 23[Suppl l]:78-90.

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Muhlbauer W, et al. Early operation in craniofacial dysostosis. *World J Surg* 1989; 13:366-372.

Posnick JC. Craniofacial dysostosis. Staging of reconstruction and management of the midface deformity. *Neurosurg Clin N Am* 1991; 2:683-702.

Renier D, Marchac D. Craniofacial surgery for craniosynostosis: functional and morphological results. *Ann Acad Med Singapore* 1988; 17:415-426.

Strohecker B. Surgical reconstruction of patients with Crouzon's syndrome: a lifetime of needs. *Plast Surg Nurs* 1995; 15:23-29.

Turvey TA, Long RE Jr, Hall DJ. Multidisciplinary management of Crouzon syndrome. *J Am Dent Assoc* 1979; 99: 205-209.

Crowded Disc Syndrome (Bilateral choroidal folds and optic neuropathy) 328

Ocular: Bilateral choroidal folds; optic disk congestion; optic atrophy; hyperopia; shortened axial length

Clinical: Elevated intracranial ruled out

Sarraf D, Schwartz SD: Bilateral choroidal folds and optic neuropathy: a variant of the crowded disk syndrome. *Ophthalmology* 2003; 110: 1047-1052.

CRST Syndrome (Thibierge-Weissenbach Syndrome; Calcinosis, Raynaud Phenomenon, Sclerodactyly, and Telangiectasia) 329

General: Scleroderma variant; possible autosomal dominant transmission; resembles Rendu-Osler- Weber disease; well-established association with primary biliary cirrhosis; prevalent in females; average age of onset 45 years; reported only in whites.

Ocular: Conjunctival hyperemia; keratitis; profuse tear lysozyme; gritty, burning sensations; bilateral optic neuropathy.

Clinical: Dermal and subcutaneous calcinosis; Raynaud phenomenon; sclerodactyly; telangiectasia; calcinosis cutis; multiple intracranial aneurysms.

Boschi A, et al. Bilateral optic neuropathy associated with the crest variant of scleroderma. *Eur J Ophthalmol* 1993; 3:219-222.

Frayha RA, et al. Familial CRST syndrome with sicca complex. *J Rheumatol* 1977; 4:53-58.

Ortiz JR, et al. CREST-associated multiple intracranial aneurysms and bilateral optic neuropathies. *J Clin Neuro- Ophthalmol* 1991; 11:233-240.

Thibierge G, Weissenbach RJ. Concretions Calcaires Souscutanees et Scleroderme. *Ann Derm Syph (Paris)* 1911; 2: 129-155.

Cryoglobulinemia 330

General: Hematologic disorder in which a group of proteins precipitate from serum on exposure to cold but redissolve when warmed. Frequently occurs in diseases of reticuloendothelial systems, multiple myeloma, and Hodgkin disease.

Ocular: Congestive retinopathy; vitreous hemorrhage; retinal detachment; rubeosis iridis; neovascular glaucoma; optic disk edema; conjunctival vascular congestion.

Clinical: Widespread intravascular coagulation in small vessels throughout body.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. Clinical *Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Cryptococcosis (Torulosis) 331

General: A pulmonary infection caused by *Cryptococcus neoformans*, a saprophyte found in weathered pigeon droppings, soil, and unpasteurized cow's milk; infection acquired through respiratory system and usually manifests as meningoencephalitis; higher incidence in patients with AIDS.

Ocular: Blurred or poor vision; diplopia; uveitis; papilledema; retinal detachment; retinal hemorrhage and exudates; secondary glaucoma; vitreous reaction; retinitis; proptosis; a mass over the optic nerve head; disease process can be bilateral or unilateral; cranial nerve VI palsy; visual loss; conjunctivitis.

Clinical: Severe headache; dizziness; ataxia; vomiting; tinnitus; memory disturbances; jacksonian convulsions; fever usually is absent; occurs frequently in patients with leukemia or lymphoma.

Avendano J, et al. Ocular cryptococcosis. *Am J Ophthalmol* 1978; 86: 110-113.

Balmes R, et al. Conjunctival cryptococcosis preceding human immunodeficiency virus seroconversion. *Am J Ophthalmol* 1992; 113:719-721.

Cohen DB, Glasgow BI Bilateral optic nerve cryptococcosis in sudden blindness m patients with acquired immune deficiency syndrome. *Ophthalmology* 1993; 100:1689-1694.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Hester DE, et al. Isolated ocular cryptococcosis in an immunocompetent patient. *Ophthalmic Surg* 1992; 23:129-131.

Kestelyn P, et al. Ophthalmic manifestations of infections with Cryptococcus neoformans in patients with the acquired immunodeficiency syndrome. *Am J Ophthalmol* 1993; 116:721-72 7.

Kupfer C, et al. A possible cause of decreased vision in cryptococcal meningitis. *Invest Ophthalmol* 1974; 13: 801-804.

Lesser RL, et al. Cryptococcal meningitis and internal ophthalmoplegia. *Am J Ophthalmol* 1979; 87:682-687.

Cryptophthalmia Syndrome (Cryptophthalmos Syndactyly Syndrome; Fraser Syndrome) 332

General: Autosomal recessive.

Ocular: Microphthalmia; epibulbar dermoid; cryptophthalmos; enophthalmia; eyebrows partially or completely missing; skin from forehead completely covers one or both eyes, but the globes can be palpated beneath the skin; in unilateral cases, the fellow eye may present lid coloboma; buphthalmos; conjunctival sac partially or totally obliterated; absence of trabeculae, Schlemm canal, and ciliary muscles; cornea is differentiated from the sclera; lens anomalies from complete absence to hypoplasia, dislocation, and calcification.

Clinical: Syndactyly (finger, toes) (about 40%); coloboma of alae nasi and nostrils; urogenital abnormalities, including pseudohermaphroditism and renal hypoplasia; abnormal, bizarre hairline; narrow external auditory meatus and malformation of ossicles; cleft lip and palate may occur; atresia or hypoplasia of larynx in some cases; hoarse voice; dysplastic pinna; meatal stenosis; glottic web and subglottic stenosis.

Ford GR, et al. ENT manifestations of Fraser syndrome. *J Laryngol Otol* 1992; 106:1-4.

Goldhammer Y, Smith JL. Cryptophthalmos syndrome with basal encephaloceles. *Am J Ophthalmol* 1975; 80: 146.

Ide CH, Wollschlaeger PB. Multiple congenital abnormalities associated with cryptophthalmia. *Arch Ophthalmol* 1969; 81:638.

Morax S, et al. Orbito-palpebral reconstruction in two cases of incomplete cryptophthalmos. *Adv Ophthalmic Plast Reconstr Surg* 1992; 9:81-91.

Pankau R, et al. Fraser (cryptophthalmos syndactyly) syndrome: a case with bilateral anophthalmia but presence of normal eyelids. *Genet Couns* 1994; 5:191-194.

Curly Hair-Ankyloblepharon-Nail Dysplasia Syndrome (CHANDS) 333

General: Autosomal recessive with pseudodominance.

Ocular: Congenital ankyloblepharon (fused eyelids).

Clinical: Curly hair; hypoplastic nails.

Baughman FA. CHANDS: the curly hair-ankyloblepharon-nail dysplasia syndrome. Clinical delineation of birth defects. *Birth Dejects* 1971; 7:100-102.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Curtius Syndrome (Ectodermal Dysplasia with Ocular Malformations) 334

General: Ectodermal dysplasia of the skin with ocular involvement; occasional combinations such as hypoplasia of nails and hair, and malformation of cheeks, temples, and breasts.

Ocular: Hypertelorism; sparse eyelashes (hypotrichosis); nystagmus; decreased tear secretion; congenital cataract; tapetoretinal degeneration; coloboma.

Clinical: Hidrotic ectodermal dysplasia; ichthyosis vulgaris; acrofacial syndactylic dysostosis; hypodontia.

Bowen P, Armstrong HB. Ectodermal dysplasia, mental retardation, cleft lip/palate and other anomalies in three sibs. *Clin Genet* 1976; 9:35-42.

Bystrom EB, et al. The syndrome of electrodactyly, ectodermal dysplasia, and clefting (EEC). *J Oral Surg* 1975; 33: 192-198.

Curtius F. Kongenitaler Partieller Riesenwuchs mit Endokrinen Storungen. *Deutsch Arch Klin Med* 1925; 147:310.

Cushing (1) Syndrome (Adrenocortical Syndrome; Hyperadrenalism Syndrome; Pituitary Basophilism; Suprarenal Syndrome) 335

General: Excessive secretion of adrenal cortical hormones due to primary or secondary adrenal hyperplasia or induced by adrenal or extraadrenal neoplastic tissue; common in females of childbearing age.

Ocular: Proptosis (rare); ocular muscle palsies; uncharacteristic visual field changes (not necessarily resembling bitemporal hemianopsia); optic nerve and/or chiasmal compression either unilateral or bilateral; posterior subcapsular cataract; central serous retinopathy; Lisch nodules.

Clinical: Hirsutism; obesity; "buffalo hump"; hypertension; diabetes; skin pigmentation; osteoporosis; abdominal striae; polycythemia and lymphopenia; weakness; nervousness; irritability; dysmenorrhea.

Bouzas EA, et al. Central serous chorioretinopathy in endogenous hypercortisolism. *Arch Ophthalmol* 1993; 111: 1229-1233.

Bouzas EA, et al. Lisch nodules in Cushing's disease. *Arch Ophthalmol* 1993; 111:439-440.

Bouzas EA. et al. Posterior subcapsular cataract in endogenous Cushing syndrome: an uncommon manifestation. *Invest Ophthalmol Vis Sci* 1993; 34:3497-3500.

Cushing H. The basophil adenomas of the pituitary body and their *Clinical* manifestations (pituitary basophilism). *Bull Johns Hopkins Hosp* 1932; 50: 137.

Rovit RL, Duane TD. Eye signs in patients with Cushing's syndrome and pituitary tumors: some observations related to chromophobe tumors and hyperadrenalism. *Trans Am Ophthalmol Soc* 1967; 65:52.

Cushing (2) Syndrome (Angle Tumor Syndrome; Cerebellopontine Angle Syndrome; Pontocerebellar Angle Tumor Syndrome; Acoustic Neuroma Syndrome) 336

General: Tumor involving cranial nerves V, VI, VII, and VIII and brainstem; occurs between ages 30 and 45 years.

Ocular: Paresis orbicularis muscle (VII); paresis external rectus muscle (VI); mixed nystagmus with head tilt; palsies of extraocular muscles are accounted for by increased intracranial pressure if the aqueduct of Sylvius is closed by the growing tumor; decreased corneal reflex V (homolateral and early sign); bilateral papilledema (increased intracranial pressure).

Clinical: Deafness (homolateral); labyrinth function disturbed or lost; tinnitus; hyperesthesia of the face; homolateral facial nerve paresis (total paralysis rare); hoarseness; difficulties in swallowing; unilateral limb ataxia; gait ataxia; nuchal headache; emesis; facial pain, numbness and paresis; progressive unilateral hearing loss.

Berg EF. The ocular signs and symptoms of posterior fossa disorders. In: Gay AJ, Burde RM, eds. *Clinical Concepts in Neuro-Ophthalmology*. *Int Ophthalmol Clin* 1967; 7:801.

Cushing H. *Tumors of the nevus acusticus.* Philadelphia: WB Saunders, 1917.

Cushing (3) Syndrome (Chiasmal Syndrome) 337

General: Suprasellar meningioma, aneurysm in the anterior part of the circle of Willis, and craniopharyngioma are the three most common lesions; usually occurs in adult patients.

Ocular: Bitemporal hemianopsia (progressive); in early stages, optic disk may appear normal or only slightly pale; later, sharp, border-lined white optic disk.

Clinical: Craniopharyngiomas; pituitary adenoma; aneurysm; chiasmal glioma; nasopharyngeal carcinoma; germinoma; glioma; choristoma; chordoma; hemangioma; leukemia; lymphoma; metastatic tumor; arteriovenous malformation; sarcoidosis; pituitary abscess; sphenoid sinus mucocele; arachnoid cyst.

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Gittinger JW Jr. Chiasmal disorders. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology. vol. IV.* Philadelphia: WB Saunders, 1994:2615.

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Waller RR, et al. A rare cause of the chiasmal syndrome. *Arch Ophthalmol* 1972; 88:269.

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Cutis Marmorata Syndrome (Marble Skin Syndrome) 338

General: Prominent in children; etiology unknown; possibly a mild form of livedo reticularis; in severe cases, vessel dilation and skin changes may be permanent; occurs with exposure to cold and subsides in a warm environment; extremities and pectoral region most frequently affected.

Ocular: Congenital glaucoma; corneal and scleral thinning; staphyloma; sclerotic appearance to trabecular meshwork; corneal edema; cataracts; optic nerve atrophy; heterochromia iridium; iris anterior layer dysplasia; intraoperative suprachoroidal hemorrhage (rare).

Clinical: Bluish-red mottling of skin; spasmodic narrowing of arterioles with dilation of vessels; ulceration and scaling of skin; congenital hypothyroidism.

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Mayatepek E, et al. Congenital glaucoma in cutis marmorata telangiectatica congenita. *Ophthalmologica* 1991; 202: 191-193.

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Sato SE, et al. Congenital glaucoma associated with cutis marmorata telangiectatica congenita: two case reports. *J Pediatr Ophthalmol Strabismus* 1988; 25: 13-17.

Cystic Fibrosis Syndrome (Fibrocystic Disease of Pancreas) 339

General: Autosomal recessive; Caucasians; lungs, pancreas, and salivary glands are mainly involved.

Ocular: Ischemic retinopathy caused by carbon dioxide retention and chronic respiratory insufficiency; vein congestion and capillary dilation around the optic nerve; retinal hemorrhages; macular degeneration; papilledema; optic atrophy; xerosis of conjunctiva; optic neuritis; abnormal pupillary responses; decreased contrast sensitivity.

Clinical: Failure to gain weight properly; recurrent pulmonary infections; salty skin; pancreatic insufficiency with malabsorption; abdominal cramps; diarrhea; increased appetite; dyspnea; chronic cough; production of viscous tenacious sputum; fever; retarded growth; delayed puberty; distended abdomen; hyperresonant chest; depressed diaphragm; clubbing of fingers.

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Spaide RF, et al. Ocular findings in cystic fibrosis. *Am J Ophthalmol* 1987; 103:204-210.

Cysticercosis 340

General: Caused by *Taenia solium.*

Ocular: Tenonitis; endophthalmitis; optic atrophy; papilledema; retinal detachment; retinal hemorrhages and exudates; vitreal hemorrhages; hypopyon; uveitis; paresis of extraocular muscles; periretinal proliferation; cysts may be present almost anywhere in or around the eye; orbital lesion; extraocular myositis; subretinal cysticercosis; acute preseptal cellulitis.

Clinical: Dead larvae may cause muscle pain; weakness; fever; eosinophilia; calcification of tissues.

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Luger MH, et al. In toto removal of a subretinal Cysticercus cellulosae by pars plana vitrectomy. *Br J Ophthalmol* 1991; 75:561-563.

Perry HD, Font RL. Cysticercosis of the eyelid. *Arch Ophthalmol* 1978; 96:1255-1257.

Pollard ZF. Cysticercosis: an unusual cause of papilledema. *Ann Ophthalmol* 1975; 7:110-112.

Stewart CR, et al. Cysticercosis as a cause of severe medial rectus muscle myositis. *Am J Ophthalmol* 1993; 116: 510-511.

Cystinuria 341

General: Caused by abnormal protein metabolism; recessive or incomplete recessive.

Ocular: Pigmentary retinopathy; gyrate atrophy.

Clinical: Impaired intestinal absorption and renal tubular reabsorption of cystine, lysine, arginine, and ornithine leads to excretion of these dibasic amino acids into urine; associated with renal dwarfism, pyramidal and extrapyramidal symptoms, and deaf mutism.

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Khan MY, et al. Gyrate atrophy of the choroid and retina with hyperornithinaemia, cystinuria and lysinuria. *Eye* 1994; 8[Pt 3]:284-287.

Cytomegalic Inclusion Disease (Cytomegalovirus; Congenital Cytomegalic Inclusion Disease) 342

General: Cytomegalovirus passes transplacentally from an asymptomatic mother to fetus.

Ocular: Uveitis; cataract; optic atrophy; inclusion bodies in the aqueous humor; severe conjunctivitis; corneal opacities; microphthalmos; strabismus; dacryoadenitis; chorioretinitis; cytomegalovirus retinitis (most common cause of acquired viral retinitis, primarily because of the AIDS virus); glaucoma.

Clinical: Cerebral calcifications; microcephaly; mental retardation; inclusion bodies in the urine; spastic diplegia; seizures; cerebellar hypoplasia; intraventricular hemorrhage; hydrocephalus.

Becroft DM. Prenatal cytomegalovirus infection: epidemiology, pathology and pathogenesis. *Perspect Pediatr Pathol* 1981; 6:204-241.

Chumbley LC, et al. Adult cytomegalovirus inclusion retinouveitis. *Am J Ophthalmol* 1975; 80:807-816.

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Sekhsaria S, et al. An unusual case of congenital cytomegalovirus infection with glaucoma and communicating hydrocephalus. *Clin Pediatr* 1992; 31:505-507.

Yoser SL, et al. Systemic viral infections and their retinal and choroidal manifestations. *Surv Ophthalmol* 1993; 37: 313-352.

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Danbolt-Closs Syndrome (Acrodermatitis Enteropathica; Brandt Syndrome) 343

General: Etiology unknown; autosomal recessive; occurs in both sexes with onset in early infancy; characterized by intermittent simultaneous occurrence of diarrhea and dermatitis with failure to thrive.

Ocular: Loss of eyebrows; blepharitis; ectropion; loss of eyelashes; photophobia; conjunctivitis; scattered superficial corneal opacities; keratitis; lacrimal punctal stenosis; corneal superficial punctate lesions, nebulous subepithelial opacities and linear epithelial erosions.

Clinical: Symmetrical skin eruptions on hands, feet, elbows, knees, and buttocks usually dry up to an erythematosquamous type; glossitis and stomatitis; alopecia; paronychia with nail dystrophy; gastrointestinal disturbances; diarrhea (intermittent).

Cherry PM, Falcon MG. Punctual stenosis-caused by idoxuridine or acrodermatitis enteropathica? *Arch Ophthalmol* 1976; 94:1632.

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Gellis SS, Feingold M. Acrodermatitis enteropathica. *Am J Dis Child* 1969; 117:201.

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Prabriputaloong A, Prakitrittranon W Corneal involvement in acrodermatitis enteropathica: a case report. *J Med Assoc Thailand* 1992; 75:423-427.

Racz P, et al. Bilateral cataract in acrodermatitis enteropathica. *J Pediatr Ophthalmol Strabismus* 1979; 16:180-182.

Sandstrom B, et al. Acrodermatitis enteropathica, zinc metabolism, copper status and immune function. *Arch Pediatr Adolesc Med* 1994; 148:980-985.

Dandy-Walker Syndrome (Atresia of the Foramen of Magendie) 344

General: Manifested in infants; malformation and stenosis of the foramina of Luschka and Magendie; dilation of fourth ventricle.

Ocular: Ptosis; sixth nerve paralysis; papilledema.

Clinical: Hydrocephalus (varies in severity) with enlargement of the skull and thinning of the bone predominantly in occipital region; loss of tendon reflexes; basilar impression; scoliosis; hydromelia.

Carrier H, et al. Dandy-Walker malformation. Study of an anatomical case and current concepts. *Ann Anat Pathol (Paris)* 1973; 18:405.

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Darier-White Syndrome (Keratosis Follicularis; Dyskeratosis Follicularis Syndrome; Psorospermosis) 345

General: Unknown etiology; defect in the synthesis, organization, and maturation of tonofilament-desmosome complex; irregular dominant inheritance; both sexes equally affected, with onset in childhood; chronic but relatively benign and more aggravated in the summer.

Ocular: Conjunctival keratosis; bilateral corneal subepithelial infiltrations and sometimes corneal ulceration; cataract formation (rare).

Clinical: Confluent flesh-colored keratotic papules on head, neck, back, abdomen, and groin; small stature; mild mental retardation; hair loss; genital hypoplasia; oral-mucosal lesions; hypertrophic flexural involvement; acral signs.

Blackman HJ, et al. Corneal epithelial lesions in keratosis follicularis (Darier's disease). *Ophthalmology* 1980; 87: 931-943.

Burge SM, Wilkinson JD. Darier- White disease: a review of the *Clinical* features in 163 patients. *J Am Acad Dermatol* 1992; 27:40-50.

Darier J. Psorospermatose folliculaire vegetante. *Ann Dermatol Syphil* 1889; 10:597.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Mohamed KN. Darier's disease and corneal opacity. *Dermatologica* 1991; 182:205.

Dawson Disease (Dawson Encephalitis; Subacute Sclerosing Panencephalitis; Inclusion-Body Encephalitis) 346

General: Sclerosing panencephalitis classified as a degenerative, progressive neurologic disorder caused by a measles virus infection of the central nervous system.

Ocular: Nystagmus; ptosis; papilledema; optic neuritis; macular pigmentation and degeneration; focal retinitis; ocular motor palsies; optic atrophy; preretinal vitreous membrane; exophthalmos; visual agnosia; chorioretinitis; retinal vasculitis; macular chorioretinitis.

Clinical: Chronic inflammation of brain with neuronal degeneration; gliosis; eosinophilic inclusion bodies in brain tissue; decline in intellect; behavioral changes; slurred speech; drooling; motor abnormalities; disorientation; seizures; death.

Fenichel GM. Subacute sclerosing panencephalitis. In: Fenichel GM, ed. *Clinical Pediatric Neurology,* 2nd ed. Philadelphia: WB Saunders, 1993:137.

Gravina RF, et al. Subacute sclerosing panencephalitis. *Am J Ophthalmol* 1978; 86:106-109.

Johnston HM, et al. Visual deterioration as presentation of subacute sclerosing panencephalitis. *Arch Dis Child* 1980; 55:899-90l.

Kovacs B, Vastag O. Fluoroangiographic picture of the acute stage of the retinal lesion in subacute sclerosing panencephalitis. *Ophthalmologica* 1978; 177:264-269.

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Zagami AS, Lethlean AK. Chorioretinitis as a possible very early manifestation of subacute sclerosing panencephalitis. *Aust N Z J Med* 1991; 21:350-352.

DeBarsy Syndrome 347

General: Rare progeroid syndrome associated with characteristic ocular, facial, skeletal, dermatologic, and neurologic abnormalities.

Ocular: Congenital corneal opacification (loss of Bowman layer); cataracts.

Clinical: Short stature, pectus excavatum, skeletal dysplasia with short legs, multiple joint dislocations, especially involving the hands; skin redundancy (as seen in cutis laxa); midface hypoplasia; thin transparent skin with prominent superficial veins; frontal bossing and aged, progeroid facies; early death; hypotonia; mental retardation; brisk deep tendon reflexes.

Aldave AJ, et al. Congenital corneal opacification m DeBarsy syndrome. *Arch Ophthalmol* 2001; 119:285-288.

Bartsocas CS, et al. De Barsy syndrome. *Prog Clin Biol Res* 1982; 104:157-160.

De Barsy AM, Moens E, Dierckx L. Dwarfism, oligophrenia and degeneration of the elastic tissue in skin and cornea: a new syndrome. *Helv Paediatr Acta* 1968; 3:305-313.

Deerfly Fever (Francis Disease; Rabbit Fever; Tularemia; Deerfly Tularemia) 348

General: Acute infectious disease caused by *Francisella (Pasteurella) tularensis.*

Ocular: Chemosis; conjunctivitis; corneal ulcer; endophthalmitis; dacryocystitis; optic atrophy; iris prolapse; chalazion; corneal opacity; pannus.

Clinical: Local ulcerative lesion; suppuration of regional lymph nodes; fever; prostration; myalgia; severe headache; pneumonia.

Bloom ME, et al. Oculoglandular tularemia in an inner city child. *Pediatrics* 1973; 51:564-566.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WE Saunders, 2000.

Hanna C, Lyford JH. Tularemia infection of the eye. *Ann Ophthalmol* 1971; 3:1321-1325.

Degos Syndrome (Malignant Atrophic Papulosis; Degos-Delort-Tricot Syndrome; Cutaneointestinal Syndrome; Kolmeier-Degos Syndrome) 349

General: Rare cutaneovisceral disease; male preponderance; death occurs within a few months after diffuse eruption of skin lesions; multiple cerebral infarcts and/or thrombosis of small arteries; lymphocytic-mediated necrotizing vasculitis with mucin deposits in the dermis.

Ocular: Atrophic skin of eyelids; intermittent diplopia; conjunctiva may be atrophic; telangiectasias of conjunctiva with microaneurysms; peripheral choroiditis; papilledema has occurred with progressive central nervous system involvement; necrotic papules of lids, bulbar conjunctivae, and episcleral tissues.

Clinical: Porcelain-white skin lesions (asymptomatic and diffuse); anorexia and/or weight loss; gastrointestinal involvement; peritonitis; intermittent paresthesias with early central nervous system involvement; signs of progressive cerebral and cerebellar atrophy; peripheral telangiectatic rim.

Degos R, et al. Dermatite papulo-squameuse atrophiante. *Bull Soc Fr Dermatol Syphil* 1942; 49:148,281.

Degos R. Malignant atrophic papuloses. *Br J Dermatol* 1979; 100:21-35.

Demitsu T, et al. Malignant atrophic papulosis (Degos' syndrome). *Int J Dermatol* 1992; 31:99-102.

Howard RO, et al. Malignant atrophic papulosis. *Arch Ophthalmol* 1968; 79:262.

Kohlmeier W Multiple Hautenekrosen bie Thromboangiitis Obliterans. *Arch Dermatol Syphil* l941; 181:783-792.

Leslie TA, et al. Degos disease and spastic paraplegia. *Clin Exp Dermatol* 1993; 18:344-346.

Premalatha S, et al. Malignant atrophic papulosis (Degos syndrome). First case report from India. *Clin Exp Dermatol* 1980; 5:370-371.

Dejean Syndrome (Orbital Floor Syndrome) 350

General: Usually secondary to a traumatic lesion involving the floor of the orbit.

Ocular: Enophthalmos; exophthalmos; lid hematoma; diplopia due to displacement of the globe or restricted function of the inferior rectus and/or inferior oblique muscles; orbital emphysema.

Clinical: Severe pain in superior maxillary region; numbness in area of first and second branches of trigeminal nerve; nausea and vomiting.

Bireer RB, et al. Orbital emphysema: how common, how significant? *Ann Emerg Med* 1994; 24:1115-1118.

Dejean C. Le Syndrome du Plancher de l'Orbite. *Bull Mem Soc Fr Ophthalmol* 1935; 48:473.

McKeown CA. Ocular motility disorders and orbital trauma. *Int Ophthalmol Clin* 1992; 32:123-149.

Milauskas AT, Fueger GF. Serious ocular complications associated with blowout fractures of the orbit. *Am J Ophthalmol* 1966; 62:670.

Dejerine-Klumpke Syndrome (Lower Radicular Syndrome; Klumpke Syndrome; Klumpke Paralysis) 351

General: Lesion involving the inferior roots of the brachial plexus with nerves derived from the eighth cervical and first thoracic root.

Ocular: Enophthalmos; ptosis; narrowed palpebral fissure; miosis.

Clinical: Paralysis and atrophy of the small muscles of forearm and hand (flexor carpi ulnaris, flexor digitorum, interossei, thenar, hypothenar); decreased sensation or increased sensibility on the inner side of the forearm.

Dejerine-Klumpke A. Contribution a l'Etude des Paralysies Radicularies du Plexus Brachial; Paralysies Radicularies Totales; Paralysies Radicularies Inferieures; de la Participation des Filets Sympathiques Oculopupillaries dans ces Paralysies. *Rev Med (Paris)* 1885;5:591,739.

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Menkes JH. Perinatal asphyxia and trauma: perinatal injuries of the peripheral nerves, brachial plexus. In: *Textbook of Child Neurology*. 5th ed. Baltimore: Williams & Wilkins. 1995:66-369.

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Dejerine-Roussy Syndrome (Retrolenticular Syndrome; Dejerine-Roussy Syndrome; Posterior Thalamic Syndrome; Thalamic Hyperesthetic Anesthesia Syndrome; Thalamic Syndrome) 352

General: Posterior thalamic lesion.

Ocular: Hemianopsia when the thalamogeniculate artery is thrombosed near its origin from the posterior cerebral artery, because there is involvement of the medial aspect of the lateral geniculate body; if the posterior cerebral artery is thrombosed, complete hemianopsia with macular sparing results; clinical transient acute esotropia; possible association between thalamic lesions and mononuclear supranuclear palsy; unilateral blepharospasm.

Clinical: Sensory disturbances: contralateral; hemiataxia: contralateral; hemiplegia (transient): contralateral; choreoathetoid movements: contralateral; spontaneous pain: contralateral.

Barroso L, Hoyt WF. Episodic exotropia from lateral rectus neuromyotonia-appearance and remission after radiation therapy for a thalamic glioma. *J Pediatr Ophthalmol Strabismus* 1993; 30:56-57.

Dejerine J, Roussy G. Le Syndrome Thalamique. *Rev Neurol* 1906; 14:521.

Kulisevsky J, et al. Unilateral blepharospasm stemming from a thalamomesencephalic lesion. *Mov Disord* 1993; 8: 239-240.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology,* 4th ed. Baltimore: Williams & Wilkins, 1987.

Scoditti U, et al. Acute esotropia from small thalamic hemorrhage. *Acta Neurol Belg* 1993; 93:290-294.

Deletion of Chromosome 12q 15-q23 353

General: Interstitial deletion of chromosome 12; autosomal dominant or autosomal recessive

Ocular: Corneal plana

Clinical: Deletion of the KERA gene

Tocyap ML, Azar N, Chen T, et al. Clinical and molecular characterization of a patient with an interstitial deletion of chromosome 12q15-q23 and peripheral corneal abnormalities Am J Ophthalmol 2006; 141(3):566-567.

De Lange Syndrome (I) (Congenital Muscular Hypertrophy Cerebral Syndrome; Brachmann-De Lange Syndrome) 354

General: Etiology not known; autosomal recessive inheritance.

Ocular: Antimongoloid slant of palpebral fissures; mild exophthalmos; hypertrichosis of eyebrows; long eyelashes; telecanthus; ptosis; blepharophimosis; nystagmus on lateral gaze; constant coarse nystagmus; strabismus; alternating exotropia; high myopia; anisocoria; chronic conjunctivitis; blue sclera; pallor of optic disk.

Clinical: Mental retardation; growth retardation; extrapyramidal motor disturbances; multiple skeletal abnormalities with congenital muscular hypertrophy; long philtrum; thin lips; crescent-shaped mouth.

Beck B. Psycho-social assessment of 36 de Lange patients. *J Ment Defic Res* 1987; 31[Pt 3]:251-257.

Bull MJ, et al. Gastrointestinal abnormalities: a significant cause of feeding difficulties and failure to thrive in Brachmann- de Lange syndrome. *Am J Med Genet* 1993; 47:1029-1034.

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Jackson L, et al. deLange syndrome: a *Clinical* review of 310 individuals. *Am J Med Genet* 1993; 47:940-946.

Jelsema RD, et al. Prenatal diagnosis of congenital diaphragmatic hernia not amenable to prenatal or neonatal repair. Brachmann-de Lange syndrome. *Am J Med Genet* 1993; 47:1022-1023.

Milot J, Demay F. Ocular anomalies in de Lange syndrome. *Am J Ophthalmol* 1972; 74:394.

Rosenbach Y, Zahavi I, Dinari G. Gastroesophageal dysfunction in Brachmann-de Lange syndrome. *Am J Med Genet* 1992; 42:379-380.

Singh NN, Pulman RM. Self-injury in the de Lange syndrome. *J Ment Defic Res* 1979; 23:79-84.

Demodicosis 355

General: *Demodex folliculorum* and *Demodex brevis* infestation; exact role in causing blepharitis is unclear; most patients are asymptomatic.

Ocular: Blepharitis; follicular distention and hyperplasia; lid hyperemia; lid hyperkeratinization; madarosis; meibomian gland destruction; mite colonies of eyelashes and eyebrows.

Clinical: Pruritus.

English FP, Nutting WB. Demodicosis of ophthalmic concern. *Am J Ophthalmol* 1981; 91:362-372.

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Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Ruff T, et al. The hair follicle mites Demodex folliculorum and Demodex brevis: biology and medical importance: a review. *Dermatologica* 1987; 162:1-11.

De Morsier Syndrome (Septooptic Dysplasia) 356

General: Absence of the septum pellucidum; agenesis of corpus callosum; enlargement of ventricles; infundibulum primary site of structural derangement in patients with optic nerve hypoplasia.

Ocular: Optic disk hypoplasia; bitemporal hemianopia; poor vision; nystagmus.

Clinical: Growth retardation; pituitary insufficiency; diabetes insipidus; normal cognitive development; intact neurologic status; normal language development; late appropriate behavior; abnormal early poor motor coordination; subtle visual alterational problems; association between optic nerve hypoplasia and cerebral hemispheric abnormalities, especially schizencephaly, due to migration anomalies.

Brodsky MC, Glasier CM. Optic nerve hypoplasia. Clinical significance of associated central nervous system abnormalities on magnetic resonance imaging. *Arch Ophthalmol* 1993; 11:66-74.

de Morsier G. Median craniocephalic dysraphia and olfactogenital dysplasia. *World Neurol* 1962; 3:485.

Rush JA, Bajandas FJ. Septo-optic dysplasia (de Morsier syndrome). *Am J Ophthalmol* 1978; 86:202-205.

Williams J, et al. Septo-optic dysplasia: the *Clinical* insignificance of an absent septum pellucidum. *Dev Med Child Neurol* 1993; 35:490-501.

Dengue Fever 357

General: Endemic over the tropics and subtropics; caused by four distinct serogroups of dengue viruses, types 1,2,3, and 4, group B arboviruses; transmitted solely by mosquitoes of the genus *Aedes.*

Ocular: Lid edema; conjunctivitis; ocular and retrobulbar pain accentuated by ocular movement; dacryoadenitis; keratitis; corneal ulcer; iritis; retinal or vitreous hemorrhages; ocular motor paresis; optic atrophy.

Clinical: Hemorrhagic fever, severe headache; backache; joint pain; rigors; insomnia; anorexia; loss of taste; epistaxis; rashes; maculopapular rash; myalgia; human infection with of four serotypes of Dengue virus causing two diseases; classic Dengue fever; Dengue hemorrhagic fever (50% mortality).

Brown SI, Grayson M. Marginal furrows. *Arch Ophthalmol* 1968; 79:563-567.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hayes EB, Gubler DJ. Dengue and Dengue hemorrhagic fever. *Pediatr Inject Dis J* 1992; 11:311-317.

Lim WK, Mathur R, Koh A, et al. Ocular Manifestations of Dengue Fever. *Ophthalmology* 2004; 111: 2057-2064.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part II*, 4th ed. Baltimore: Williams & Wilkins, 1995:4002.

Dental-Ocular-Cutaneous Syndrome 358

General: Abnormal tooth roots; distinctive features separate this syndrome from oculodentodigital or faciodentodigital syndromes.

Ocular: Entropion lower eyelids; glaucoma (juvenile type).

Clinical: Unusual upper lip with lack of "cupid's bow" and thickening and widening of the philtrum; syndactyly; cutaneous hyperpigmentation overlying the interphalangeal joints; clinodactyly; single conical roots in all primary teeth and permanent first molars; scant body hair; horizontal ridging of fingernails.

Ackerman JL, et al. A new dental, ocular and cutaneous syndrome. *Int J Dermatol* 1973; 12:285.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger. 1976.

Dermatitis Herpetiformis (Duhring-Brocq Disease) 359

General: Malignant; atypical; does not respond well to sulfone or sulfapyridine therapy; uncommon; autoimmune blistering dermatosis; pruritic eruption involving the scalp, buttocks, lower back, and extensor surface of arms; autoantibody is generally of immunoglobulin A class causing deposition at the dermal-epidermal junction.

Ocular: Bullae of conjunctiva, skin, and mucous membranes; blisters are intraepithelial (acantholysis) and usually do not leave scars; epithelium desquamates in patches; corneal and conjunctival vascularization; symblepharon; cataract.

Clinical: Vesicles; erythema; pruritus; burning; eruption classically involves extensor surface of the knees, elbows, buttocks, sacrum, scapula, and scalp.

Duane TD. *Clinical Ophthalmology, vol. IV.* Philadelphia: JB Lippincott, 1987.

Foster SC. Immunologic disorders of conjunctiva, cornea and sclera. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:200.

Fraser NG, et al. Dermatitis herpetiformis and Sjogren's syndrome. *Br J Dermatol* 1979; 100:213-215.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Dermatophytosis (Epidermophytosis; Epidermomycosis; Rubrophytia; Tinea; Trichophytosis) 360

General: Superficial infection of the skin; ringworm fungi; most frequently seen in children during hot, humid weather.

Ocular: Conjunctivitis; corneal ulcer; madarosis; scaly rash; folliculitis; blepharitis; lid edema.

Clinical: Scalp, facial, and lid ringworm lesions.

Fedukowicz HB. *External Infections of the Eye: Bacterial, Viral, and Mycotic.* 3rd ed. Norwalk, CT: Appleton-Century-Crofts, 1984.

Francois J, Rysselaere M. *Oculomycoses.* Springfield, IL: Charles C. Thomas, 1972:229-245.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Dermoid (Dermoid Choristoma; Dermoid Cyst; Dermolipoma; Lipodermoid) 361

General: Benign tumors composed of epidermal tissue, dermal adnexal structures, skin appendages, hair follicles, sebaceous gland, and sweat glands; slowly growing.

Ocular: Dermoid of conjunctiva, cornea, and lids; keratitis; extraocular muscle paralysis; exophthalmos; astigmatism; visual loss; orbital lesions causing diplopia and proptosis; may be connected with the lacrimal canaliculum.

Clinical: Subcutaneous dermoids of the skin; aplasia cutis congenita possibly associated with strabismus has been reported.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gardner J, Viljoen D. Aplasia cutis congenita with epibulbar dermoids: further evidence for syndromic identity of the ocular ectodermal syndrome. *Am J Med Genet* 1994; 53:317-320.

Grove AS Jr. Giant dermoid cysts of the orbit. *Ophthalmology* 1979; 86:1513-1520.

Leonardo D, et al. Recurrent giant orbital dermoid of infancy. *J Pediatr Ophthalmol Strabismus* 1994; 31:50-52.

Paris GL, Beard C. Blepharoptosis following dermolipoma surgery. *Ann Ophthalmol* 1973; 5:697-699.

Sharman P, et al. Dermoids: *Clinical* presentation and management. *Br J Ophthalmol* 1984; 68:642-652.

Desert Lung and Cataract Syndrome 362

General: Nonoccupational pneumoconiosis; common in desert areas; excessive exposure to atmospheric dust; inhalant of fine, sandy dust.

Ocular: Cataracts (posterior subcapsular most prevalent); corneal opacities.

Clinic: Miliary infiltrates; thickening of bronchial walls; mild restrictive changes with pulmonary function.

Hawass ND. An association between desert lung and cataract: a new syndrome. *Br J Ophthalmol* 1987; 71:694-697.

Newell FW *Ophthalmology: Principles and Concepts*, 6th ed. St. Louis: CV Mosby, 1986.

Devic Syndrome (Ophthalmoencephalomyelopathy; Optic Myelitis; Neuromyelitis Optica) 363

General: Etiology unknown; frequent between the ages of 20 and 50 years; mortality rate up to 50%; associated with chickenpox.

Ocular: Ptosis is rare; ocular muscle palsy (rare); abducens and oculomotor palsy; paralysis of conjugate gaze; blindness; onset usually very sudden in one eye, followed soon by blindness in the other eye; miosis; bilateral optic neuritis (unilateral involvement is rare); optic atrophy; pupillary dysfunction.

Clinical: Prodromal signs: headache; sore throat; fever and malaise; ascending myelitis with resulting pain, which may be severe; numbness; weakness; paralysis.

Ahasan HA, et al. Neuromyelitis optics (Devic's disease) following chicken pox. *Trop Doct* 1994; 24:75-76.

al-Deeb SM, et al. Devic's neuromyelitis optica and varicella. *J Neurol* 1993; 240:450-451.

Chusid MJ, et al. Neuromyelitis optica (Devic disease) following varicella infection. *J Pediatr* 1979; 95:737-738.

Devic E. Myelite Aigue Dorso-lumbaire, avec Nevrite Optique; Autopsie. *Cong Fr Med* 1894-1895; 1:434.

Francois J. Ocular manifestations in demyelinating diseases. *Adv Ophthalmol* 1979; 39:1-36.

Hauser SL. Multiple sclerosis and other demyelinating diseases. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:2287-2294.

Kinney EL, et al. Devic's syndrome and systemic lupus erythematosus: a case report with necropsy. *Arch Neurol* 1979; 36:643-644.

Sadig SA, Miller JR. Devic syndrome. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1995:825.

Williamson PM. Neuromyelitis optica following infectious mononucleosis. *Proc Aust Assoc Neurol* 1975; 12: 153-155.

Diabetes Mellitus (Willis Disease) 364

General: Metabolic disorder that affects carbohydrate and lipid metabolism; familial, usually occurs over age 40 years; frequently occurs in people who are overweight.

Ocular: Ectropion uveae; rubeosis iridis; cataract; optic nerve atrophy; microaneurysms of retina; lipemia retinalis; dilation of the retinal veins; cotton-wool spots; hard yellow exudates; irregular sheathing of retinal veins; macular edema; retinal neovascularization; vitreal hemorrhages; hypotony; glaucoma; asteroid hyalosis; paresis of third or sixth nerve; anterior ischemic optic neuropathy; decreased corneal sensation.

Clinical: Hyperglycemia with resulting glycosuria and polyuria; imperfect combustion of fats leading to acidosis, dyspnea, lipemia, ketonuria, and coma; atherosclerosis; pruritus; lowered resistance to pyogenic infections; proteinuria predicts vision loss, indicates more advanced diabetic retinopathy; glycosylated hemoglobin also is a risk factor for retinopathy and macular edema.

Barner SS, Fledelius HC. Anterior ischemic optic neuropathy as an initial sign of diabetes mellitus. *Ann Ophthalmol* 1982; 14:492-496.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Feman SS. The natural history of the first *Clinical*ly visible features of diabetic retinopathy. *Trans Am Ophthalmol Soc* 1994; 92:745-773.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Michels RG. Vitrectomy for complications of diabetic retinopathy. *Arch Ophthalmol* 1978; 96:237-246.

Moss SE, et al. Ten-year incidence of visual loss in a diabetic population. *Ophthalmology* 1994; 101:1061-1070.

Pavan PR, et al. Optic disc edema in juvenile-onset diabetes. *Arch Ophthalmol* 1980; 98:2193-2195.

Ruben ST. Corneal sensation in insulin dependent and non-insulin dependent diabetics with proliferative retinopathy. *Acta Ophthalmol* 1994; 72:576-580.

Dialinas-Amalric Syndrome (Amalric-Dialinas Syndrome; Deaf Mutism-Retinal Degeneration Syndrome) 365

General: Retinal pigmentary disturbances and deafness as outstanding findings but without severe general systemic disorders as seen in the syndromes of Hallgren, Cockayne, Alport, Laurence-Moon-Bardet-Biedl (see Hallgren Syndrome; Cockayne Syndrome; Alport Syndrome; Laurence-Moon-Bardet-Biedl Syndrome).

Ocular: No night blindness but heterochromia iridis; atypical retinitis pigmentosa with small, scattered, fine-pigmented deposits in the macular region with some accumulations and accompanied by small white and yellow spots.

Clinical: Deaf mutism.

Amalric P. Nouveau Type de Degenerescence Tapeto-Retinienne au Cours de la Surdumutite. *Bull Soc Ophthalmol Fr* 1960; 196:211.

Charamis J, et al. Deaf-mutism and ophthalmic lesions. *J Pediatr Ophthalmol* 1968; 5:230.

Dialinas NP. Les Alterations Oculaires chez les Sourds-Muets. *Genet Hum* 1959; 8:225.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Diamond Blackfan Syndrome 366

General: Rare, congenital hematologic disorder characterized by isolated erythroid hypoplasia (hypoplastic anemia).

Ocular: Strabismus, hypertelorism, microphthalmos, and infantile glaucoma.

Clinical: May have musculoskeletal abnormalities.

Balaban EP, et al. Diamond-Blackfan syndrome in adult patients. *Am J Med* 1985; 78:533-538.

Schofield KP, Evans DI. Diamond-Blackfan syndrome and neutropenia. *J Clin Pathol* 1991; 44:742-744.

Young TL, Schaffer DB, Cohen AR. Infantile glaucoma associated with the Diamond-Blackfan syndrome. *J Pediatr Ophthalmol Strabismus* 1992; 29:55-58.

Diencephalic Syndrome (Diencephalic Epilepsy Syndrome; Autonomic Epilepsy Syndrome; Penfield Syndrome; Anterior Diencephalic Autonomic Epilepsy Syndrome) 367

General: Occurs in males at ages 6 to 7 years; caused by hypothalamic dysfunction and a localized epileptic stimulus originating in the dorsal nucleus of the thalamus.

Ocular: Proptosis (occasionally); excessive lacrimation; pupillary abnormalities.

Clinical: Abdominal pain; headache; irritability; rapid pulse; elevated blood pressure; salivation; hiccup; chills; dyspnea (Cheyne-Stokes); seizures (possible); sudden onset of vasodilation of the skin (cervical sympathetic).

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Pele S, et al. Brain scanning in the diencephalic syndrome. *Neuroradiology* 1974; 7:109.

Penfield W. Diencephalic autonomic epilepsy. *Arch Neurol Psychiatr* 1929; 22:358.

Diffuse Keratoses Syndrome 368

General: Ophthalmic complications and manifestations following a group of dermatologic conditions for which the etiology is unknown, including scleroderma, CRST syndrome, Rendu-Osler- Weber syndrome, progressive systemic sclerosis with calcinosis, Spanlang-Tappeiner syndrome, and Savin syndrome (see Rendu-Osler-Weber Syndrome; Progressive Systemic Sclerosis; Spanlang-Tappeiner Syndrome; Savin Syndrome; CRST Syndrome; Scleroderma).

Ocular: Dermatologic and cicatricial skin changes; hydrophthalmos; corneal changes with nodular thickening in the stroma, which worsen in fall and improve during spring (particularly Savin syndrome); retinal phlebitis.

Clinical: Ichthyosis (usually congenital); microcephalus (CRST); facial dermatoses; deafness; urticarial (allergic) manifestations; subcutaneous calcinous plaques, mainly on hands; sclerodactyly.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976

Kleine-Natrop HE, Haustein UF. "Benign mucus membrane pemphigoid" with rapid loss of sight and generalized scarring skin change. *Hautarzt* 1968; 19:6.

Savin LH. Corneal dystrophy associated with congenital ichthyosis and allergic manifestations in male members of a family. *Br J Ophthalmol* 1956; 40:82.

Thibierge G, Weissenbach RJ. Concretions Calcaires Souscutanees et Sclerodermie. *Ann Dermatol Syph* 1911; 2:129.

Diffuse Unilateral Subacute Neuroretinitis Syndrome (DUSN; Unilateral Wipeout Syndrome; Wipeout Syndrome) 369

General: Caused by a nematode that is not *Toxocara canis,* that is, at least two nematodes of different sizes; usually occurs in children or young adults; nematode may remain viable in eye for 3 years or longer.

Ocular: Vitreitis; papillitis; gray-white lesions of retina; optic atrophy; retinal vessel narrowing; diffuse pigment epithelial degeneration; endophthalmitis; nematode in fundus; the pathognomonic finding in DUSN is the presence of a motile intraocular nematode.

Clinical: Weight loss; lack of appetite; cough; fever; pulmonary infiltration; hepatomegaly; leukocytosis; persistent eosinophilia.

Gass JD, Braunstein RA. Further observation concerning the diffuse unilateral subacute neuroretinitis syndrome. *Arch Ophthalmol* 1983; 101:1689-1697.

Gass JD, et al. Diffuse subacute neuroretinitis. *Ophthalmology* 1978; 85:521-545.

Gass JDM, Olsen KR. Diffuse unilateral subacute neuroretinitis. In: Ryan SJ, ed. *Retina, vol. II,* 2nd ed. St. Louis: Mosby, 1994.

Sivalingam A, Godberg RE. Diffuse unilateral subacute neuroretinitis. *Arch Ophthalmol* 1991; 109:1028.

Dimmer Syndrome (Keratitis Nummularis) 370

General: Onset after minor ocular trauma.

Ocular: Photophobia; ocular pain; excessive lacrimation; discoid infiltration of superficial layers of cornea without adjacent conjunctivitis.

Dimmer F. Weber Eine der Keratitis Nurmnularis. *Mahestehende Hirnhutentzuendung Augenheilkd* 1905; 13: 621-635.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Diphtheria 371

General: Acute infectious disease caused by *Corynebacterium diphtheriae;* severity is dependent upon the amount of exotoxin absorbed prior to initiation of specific therapy.

Ocular: Conjunctivitis; xerophthalmia; keratitis; corneal ulcer; blepharitis; cellulitis of lid; meibomianitis; ptosis; dacryocystitis; cataract; central retinal artery occlusion; optic neuritis; accommodative spasm or paralysis; convergence paralysis; divergence paralysis; paralysis of third, fourth, or sixth nerve; paralysis of accommodation (in children); ocular motor nerve paresis; choroiditis; cranial neuropathies involving the trigeminal, vagus, and hypoglossal cranial nerves; myocarditis.

Clinical: Local inflammatory lesion, with effect on heart, kidneys, and nervous system.

Collins JE *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duke-Elder S, ed. *System of Ophthalmology*, vol. XV. St. Louis: CV Mosby, 1976:46.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Holmes RK. Diphtheria. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:623-627.

Miller NR, ed. Walsh and Hoyt's *Clinical Neuro-Ophthalmology*, vol. 5, part 1, 4th ed. Baltimore: Williams & Wilkins, 1995.

Dirofilariasis 372

General: Caused by parasite nematode *Dirofilaria.*

Ocular: Chemosis; tenonitis; proptosis; granuloma of conjunctiva, eyelid, orbit, and sclera; periocular soft tissue mass.

Clinical: Pulmonary symptoms; coughing; cardiac infection; "coin" lesions in the lung; pruritus, most commonly in subepithelial tissues of eyelids, fingers, cheeks, breast, abdomen, and conjunctivae (rarely); allergic reactions; hemoptysis.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kersten RC, et al. Periorbital dirofilariasis. *Ophthal Plast Reconstr Surg* 1994; 10:293-296.

Preshaw LE, et al. Conjunctival dirofilariasis in British Columbia. *Can J Ophthalmol* 1993; 28:343-345.

Romano A, et al. Human ocular dirofilariasis in Israel. *Isr J Med* Sci 1976; 12:208-214.

Soylu M, et al. Dirofilariasis: an uncommon parasitosis of the eye. *Br J Ophthalmol* 1993; 77:602-603.

Thomas D, et al. The Dirofilaria parasite in the orbit. *Am J Ophthalmol* 1976; 82:931-933.

Disseminated Intravascular Coagulation 373

General: Intravascular coagulation in small vessels throughout the body.

Ocular: Serous retinal detachment; choroidal, retinal, and vitreous hemorrhage, and hyphema; recurrent choroidal hemorrhagic detachment.

Clinical: Hypocoagulable, hemorrhagic state.

Allinson RW, et al. Recurrent hemorrhagic choroidal detachment associated with disseminated intravascular coagulation. *Ann Ophthalmol* 1992; 24:72-74.

Azar P, et al. Ocular findings in disseminated intravascular coagulation. *Am J Ophthalmol* 1974; 78:493-496.

Cogan DG. Ocular involvement in disseminated intravascular coagulopathy. *Arch Ophthalmol* 1975; 93:1-8.

Martin VA. Disseminated intravascular coagulopathy. *Trans Ophthalmol Soc U K* 1978; 98:506-507.

Disseminated Lupus Erythematosus (Systemic Lupus Erythematosus; Lupus Erythematosus; Kaposi-Libman-Sack Syndrome) 374

General: Possible etiology includes viral infections and genetic predisposition; immunologic abnormalities.

Ocular: Keratitis; keratoconjunctivitis sicca; corneal ulcer; optic nerve atrophy; optic neuritis; papilledema; arteritis; central retinal vein occlusion; retinal detachment; microaneurysm; scleritis; uveitis; ptosis; conjunctivitis; paralysis of third nerve; homonymous hemianopsia; multifocal microinfarcts; mydriasis; nystagmus; proptosis; orbital myositis; pseudoretinitis pigmentosa; photophobia.

Clinical: Polyarthritis; morning stiffuess; fever; malaise; fatigue; polyserositis; renal disease; central nervous system disease; anemia; leukopenia; maculopapular rash in a "butterfly" distribution over malar region; alopecia.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Gold DH, et al. Ocular findings in systemic lupus erythematosus. *Br J Ophthalmol* 1972; 56:800-804.

Disseminated Sclerosis (Multiple Sclerosis) 375

General: Disseminated demyelination affecting white matter of the brain, spinal cord, and optic nerves; etiology unknown.

Ocular: Nystagmus; ptosis; myokymia; optic atrophy; papillitis; optic neuritis; anisocoria; Argyll Robertson pupil; Marcus Gunn pupil; hippus, decreased or absent papillary reaction to light; periphlebitis; visual field defects; gaze palsy; paralysis of third or sixth nerve; uveitis; oscillopsia; Uhthoff symptom (reduction of visual acuity with exercise or ocular hyperthermia); pars planitis; retinal venous sheathing; retinitis; granulomatous uveitis.

Clinical: Incoordination; paresthesia; spasticity; tic douloureux; urinary frequency and infections; progressive disability; paralysis; death.

Francois J. Ocular manifestation in demyelinating diseases. *Adv Ophthalmol* 1979; 39:1-36.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hutchinson WM. Acute optic neuritis and the prognosis for multiple sclerosis. *J Neurol Neurosurg Psychiatry* 1976; 39:283-289.

Lim JI, et al. Anterior granulomatous uveitis in patients with multiple sclerosis. *Ophthalmology* 1991; 98:142-145.

Lucarelli MJ, et al. Immunopathologic features of retinal lesions in multiple sclerosis. *Ophthalmology* 1991; 98: 1652-1656.

Maninowski SM, et al. The association of HLA-B8, B51, DR2, and multiple sclerosis in pars planitis. *Ophthalmology* 1993; 100:1199-1205.

Tabira T, et al. Multiple sclerosis cerebrospinal fluid produces myelin lesions in tadpole optic nerves. *N Engl J Med* 1976; 295:644-649.

Wikstrom J, et al. Optic neuritis as an initial symptom in multiple sclerosis. *Acta Neurol Scand* 1980; 61:178-185.

Williams, KE, Johnson LN: Neuroretinitis in patients with multiple sclerosis. *Ophthalmology* 2004; 111: 335-341.

Distichiasis (Distichiasis with Congenital Anomalies of the Heart and Peripheral Vasculature) 376

General: Autosomal dominant.

Ocular: Double rows of eyelashes; congenital ectropion; absence of meibomian glands; replacement of dense collagenous tissue of the tarsal plates by loose areolar tissue.

Clinical: Congenital heart defects; ventricular septal defects; stress-induced asystole; visible varicosities; chronic venous disease of the legs; sinus bradycardia.

Goldstein S, et al. Distichiasis, congenital heart defects, and mixed peripheral vascular anomalies. *Am J Med Genet* 1985; 20:283-294.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Diverticulosis of Bowel, Hernia, Retinal Detachment 377

General: Autosomal recessive.

Ocular: Severe myopia; esotropia; retinal detachment.

Clinical: Femoral or inguinal hernias; diverticula of bowel or bladder.

Clunie GJ, Mason JM. Visceral diverticula and the Marfan syndrome. *Br J Surg* 1962:50:51-52.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Dollinger-Bielschowsky Syndrome (Jansky-Bielschowsky Syndrome; Infantile Amaurotic Familial Idiocy [Late]; Infantile Ganglioside Lipidosis [Late]; Bielschowsky-Jansky Disease) 378

General: Late infantile form of neuronal ceroid lipofuscinosis; onset age 2 to 5 years; autosomal recessive; other neuronal ceroid lipofuscinoses include infantile (Santavuori-Haltia), juvenile (Spiel-Meyer-Sjögren), and adult (Kufs-Hallervorden).

Ocular: Optic nerve atrophy; macular pigmentation.

Clinical: Cerebroretinal degeneration; cerebellar ataxia; defective hearing; convulsions; spasticity; contractures; progressive mental deterioration.

Bateman JB, Philippart M. Ocular features of the Hagberg-Santavuori syndrome. *Am J Ophthalmol* 1986; 102: 262-271.

Bielschowsky M. Uber Spatintatile Familiare Amaurotische Idiotie Mit Klein Hirn-symptomen. *Dtsch* Z *Nervenh* 1914; 50:7-29.

Brod RD, et al. Diagnosis of neuronal ceroid lipofuscinosis by ultrastructural examination of peripheral blood lymphocytes. *Arch Ophthalmol* 1987; 105:1388-1393.

Dollinger A. Zur Klinik der Infantiler Form der Familioren Amaurotischen Idiotie (Tay-Sachs). Einige neue Symptome. Ein Beitrag zu den von Magnus beschrie-benen Tonischer. Hals-und Labyrinth Reflex. Z *Kinderh* 1919; 22: 167-194.

Goebel HH, et al. An ultrastructural study of the retina in the Jansky-Bielschowsky type of neuronal ceroid lipofuscinosis. *Am J Ophthalmol* 1977; 83:70-79.

Dominant Optic Atrophy Syndrome (Dominant Optic Atrophy, Deafness, Ptosis, Ophthalmoplegia, Dystaxia, and Myopathy) 379

General: Autosomal dominant disorder; ptosis, ophthalmoplegia, dystaxia, and nonspecific myopathy occur in midlife; optic atrophy and hearing loss occur in early life; autosomal dominant inheritance.

Ocular: Ptosis; ophthalmoplegia; progressive optic atrophy; abnormal electroretinogra-phy; diplopia; ocular myopathy; nystagmus; focal temporal excavation of optic disk; dyschromatopsia (blue-yellow); myopia; temporal pallor of the optic nerve.

Clinical: Sensorineural hearing loss; myopathy; dystaxia.

Del Porto G, et al. Clinical heterogeneity of dominant optic atrophy: the contribution of visual function investigations to diagnosis. *Graefes Arch Clin Exp Ophthalmol* 1994; 232:717-727.

Eliott D, et al. Visual prognosis in autosomal dominant optic atrophy. *Am J Ophthalmol* 1993; 115:360-367.

Grehn F, et al. Dominant optic atrophy with sensorineural hearing loss. *Ophthalmic Paediatr Genet* 1982;1 : 77-88.

Treft RL, et al. Dominant optic atrophy, deafness, ptosis, ophthalmoplegia, dystaxia, and myopathy. *Ophthalmology* 1984; 91:908-915.

Donohue Syndrome (Leprechaunism) 380

General: Etiology unknown; possibly autosomal recessive; prevalent in females; present at birth; possible correlation between Donohue syndrome and insulin receptor gene defects.

Ocular: Hypertelorism.

Clinical: Failure to thrive; mental retardation; sexual precocity; hirsutism; broad nose; hypertrophic nipples; hypertrophy of external genitals; intrauterine growth retardation; extreme insulin resistance hyperkeratosis; precocious tooth eruption.

Donohue WL, et al. Dysendocrinism. *J Pediatr* 1948; 32:739-748.

Goodman PA, et al. Growth factor receptor regulation in the Minnl leprechaun: defects in both insulin receptor and epidermal growth factor receptor gene expression. *Metab Clin Exp* 1992; 41:504-509.

Krook A, et al. Homozygous nonsense mutation in the insulin receptor gene in infant with leprechaunism. *Lancet* 1993; 342:277-278.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Dorsal Midbrain Syndrome 381

General: Caused by lesions of the posterior commissure located in the dorsal midbrain, pineal tumors, shunt malfunction, or hydrocephalus; less common causes include midbrain hemorrhage or infection, hypoxia, multiple sclerosis, trauma, lipid storage disease, Wilson disease, Whipple disease, syphilis, and tuberculosis.

Ocular: Loss of upward gaze; lid retraction; light-near dissociation; impaired convergence and divergence; convergence-retraction nystagmus.

Clinical: Patients tend to adopt abnormal head postures to fixate or maintain binocularity.

Balch RW, Kurman JM, Yee RD. Dorsal midbrain syndrome: *Clinical* and oculographic Findings. *Neurology* 1985; 75: 54-60.

Brazis PW 10 Clinical pearls for dorsal midbrain syndrome. *Ophthalmol Times* January 2000:9.

Buckley SA, Elston JS. Surgical treatment of supranuclear and internuclear ocular motility disorders. *Eye* 1997; 11[Pt 3]:377-380.

Double Whammy Syndrome (Voluntary Propulsion of the Eyes) 382

General: Ability to displace the globe forward actively while retracting the upper and lower lids behind the equator of the eyeball.

Ocular: Voluntary dislocation of either eye separately or of both simultaneously.

Friedenwald H. Luxation and avulsion of eye ball during birth. *Am J Ophthalmol* 1918; 1:9.

Walsh TJ, Gilman M. Voluntary propulsion of the eyes. *Am J Ophthalmol* 1969; 67:583.

Down Syndrome (Mongolism; Trisomy G; Trisomy 21 Syndrome; Mongoloid Idiocy) 383

General: Trisomy of chromosome 21.

Ocular: Hypertelorism; epicanthus; blepharitis; ectropion; nystagmus; esotropia; high myopia (30%); hyperopia; color blindness; yellow spots on the iris; hypoplasia of the iris; blepharoconjunctivitis; lens opacities (50%); keratoconus (may be acute); corneal hydrops; corneal ectasia; corneal edema; leukoma; lateral displacement of canaliculi and puncta; megaloblepharon; euryblepharon; decreased accommodation; Leber congenital amaurosis.

Clinical: Mental retardation; skeletal abnormalities; overextension of joints; deformed and low-set ears; short fifth finger; transverse palmar crease; fissured tongue; heart anomalies.

Coates DK, McCreery KB, Plager DA, et al. Nasolacrimal outlaw drainage anomalies in Down's Syndrome. *Ophthalmology* 2003; 110: 1437-1441.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Down JLH. Observations on ethnic classifications of idiots. *Clin Lect Rep Lond Hosp* 1866; 111:259.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Hayasaka S, et al. Leber congenital amaurosis in an infant with Down syndrome. *Ann Ophthalmol* 1992; 24:250-252.

Markowitz GD, et al. Congenital euryblepharon and nasolacrimal anomalies in a patient with Down syndrome. *J Pediatr Ophthalmol Strabismus* 1994; 31:330-331.

Woodhouse JM, et al. Reduced accommodation in children with Down syndrome. *Invest Ophthalmol Vis Sci* 1993; 34:2382-2387.

Doyne Honeycomb Choroiditis (Dominant Orbruch Membrane Drusen; Hutchinson-Tays Central Guttate Choroiditis; Holthouse-Batten Superficial Choroiditis; Malattia-Leventinese Syndrome) 384

General: Autosomal dominant; represents early manifestation of senile macular degeneration; both sexes affected; onset in advanced age; patients present with drusen at an early age (second to third year of life) with near-normal visual acuity in childhood.

Ocular: Drusen with multiple yellow lesions becoming calcified and presenting crystalline appearance.

Clinical: None.

Lewis H, et al. Retinal degeneration of pigment epithelium. *Ophthalmology* 1985; 92:1485-1495.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Reichel E, Sandberg MA. Hereditary macular degeneration. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994: 1254.

Dracontiasis (Dracunculosis; Guinea Worm Infection; Dracunculiasis) 385

General: Caused by *Dracunculus medinensis;* affects connective and subcutaneous tissues.

Ocular: Conjunctivitis; proptosis; nematode present in the conjunctiva, eyelid, globe, and orbit.

Clinical: Itching; urticaria; small blisters; tetany; septicemia; arthritis; paraplegia; constrictive pericarditis; urogenital involvement.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kale OO. Clinical evaluation of drugs for dracontiasis. *Trop Doct* 1977; 7:15-16.

Kothari ML, et al. Management of dracontiasis. *Ind J Surg* 1970; 32:479-484.

Dragged-Fovea Diplopia Syndrome 386

General: Binocular double vision with macular conditions such as epiretinal membrane, choroidal neovascular membranes, localized retinal detachment or paramacular scars dragging the fovea

Ocular: Maculopathy: metamorphopsia: vascular wrinkling; central diplopia in the presence of peripheral fusion

Clinical: Lights on-off test with 20/70 to 20/100 is diagnostic

DePool, ME, Campbell JP, Broome SO, et al. The dragged-fovea diplopia syndrome. *Ophthalmology* 2005; 112: 1455-1462.

Drummond Syndrome (Idiopathic Hypercalcemia; Blue Diaper Syndrome) 387

General: Autosomal recessive; manifests itself in infancy; defective intestinal transport of tryptophan, which oxidizes to indigo blue and stains the diaper blue.

Ocular: Sclerosis of optic foramina (occasionally); prominent epicanthal folds; nystagmus; strabismus; peripheral retinal atrophy; papilledema; optic atrophy; microcornea; hypoplasia of the optic nerve; abnormal eye movements.

Clinical: Dwarfism; osteosclerosis; craniostenosis; depressed bridge of nose; "elfin-like" face; mental retardation; anorexia; vomiting; constipation.

Chen Y, et al. The ocular abnormalities of blue diaper syndrome. *Metab Pediatr Syst Ophthalmol* 1991; 14:73-75.

Drummond KN, et al. The blue diaper syndrome: familial hypercalcemia with nephrocalcinosis and indicanuria: a new familial disease with definition of the metabolic abnormality. *Am J Med* 1964; 37:928.

Harley RD, et al. Idiopathic hypercalcemia of infancy: optic atrophy and other ocular changes. *Trans Am Acad Ophthalmol Otolaryngol* 1965; 69:977.

Duane Syndrome (Retraction Syndrome; Stilling Syndrome; Turk-Stilling Syndrome) 388

General: Autosomal dominant; more frequent in females; manifestations in infancy; was thought to be secondary to fibrosis of the lateral rectus muscle or abnormal check ligaments; now established to be due to congenital aberrant innervation affecting third and seventh cranial nerves.

Ocular: Narrowing of palpebral fissure on adduction, widening on abduction; primary global retraction; deficiency of medial and lateral recti motility; limitation of abduction in affected eye usually is complete; retraction of the globe with attempted adduction varies from 1 to 10 mm; convergence insufficiency; heterochromia irides; left eye is more frequently involved.

Clinical: Associated Klippel-Feil syndrome; malformation of face, ears, and teeth.

Cross HE, Pfaffenbach DD. Duane's retraction syndrome and associated congenital malformations. *Am J Ophthalmol* 1972; 73:442.

Duane A. Congenital deficiency of abduction, associated with impairment of adduction, retraction movements, contractions of the palpebral fissure and oblique movements of the eye. *Arch Ophthalmol* 1905; 34:133.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Holtz SJ. Congenital ocular anomalies associated with Duane's retraction syndrome, the nevus Ota, and axial anisometropia. *Am J Ophthalmol* 1974; 77:729.

Kim, JH, Hwang JM: Presence of the abducens nerve according to the type of Duane's retraction syndrome. *Ophthalmology* 2005; 112: 109-113.

Dubin-Johnson Syndrome (Chronic Idiopathic Jaundice; Rotor Syndrome) 389

General: Onset in infancy, but may be present at birth; autosomal dominant; liver cells are unable to excrete conjugated bilirubin; manifestations similar to Rotor syndrome, except that in the latter there is no melanin pigment present in the liver cells.

Ocular: Jaundice of sclera and conjunctiva in infancy.

Clinical: Abdominal pain right hypochondrium; nausea; vomiting; diarrhea; anorexia; weakness; hepatomegaly.

Dubin IN, Johnson FB. Chronic idiopathic jaundice with unidentified pigment in liver cells: new clinico-pathologic entity with report of 12 cases. *Medicine* 1954; 33:155.

Rosenthal P. Neonatal Dubin-Johnson syndrome. *J Pediatr Gastroenterol Nutr* 1994; 19:255.

Wolkoff AW, et al. Inheritance of the Dubin-Johnson syndrome. *N Engl J Med* 1973; 288:113.

Dubowitz Syndrome (Dwarfism-Eczema-Peculiar Facies) 390

General: Affects both sexes; congenital; may be autosomal recessive inheritance.

Ocular: Hypertelorism; lateral telecanthus; palpebral ptosis; short palpebral tissues.

Clinical: Eczema; sparse hair; cleft palate; hypospadia; microcephaly; low birth weight; mild mental retardation; characteristic face; short stature; spontaneous keloids; intrauterine growth retardation.

Berthold F, Fuhrmann W; Lampert F. Fatal aplastic anemia in a child with features of Dubowitz syndrome. *Eur J Pediatr* 1987; 146:605-607.

Dubowitz V. Familial low birth weight dwarfism with unusual facies and a skin eruption. *J Med Genet* 1969; 2:12-17.

Hansen KE, et al. Dubowitz syndrome: long-term follow-up of an original patient. *Am J Med Genet* 1995; 155:161-164.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* Philadelphia: JB Lippincott, 1981:236-237.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Paradisi M, et al. Dubowitz syndrome with keloidal lesions. *Clin Exp Dermatol* 1994; 119:425-427.

Soyer AD, McConnell JR. Progressive scoliosis in Dubowitz syndrome. *Spine* 1995; 120:2335-2337.

Winter RM. Dubowitz syndrome. *J Med Genet* 1986; 23:11-13.

Duck-Bill Lips and Ptosis 391

General: Autosomal dominant.

Ocular: Ptosis; strabismus; hypertelorism.

Clinical: Short philtrum; duck-bill lips; low-set ears; broad forehead; slightly anteverted nose and flat nasal bridge; slightly wide-spaced teeth and high-arched palate; slightly receding chin; slightly wide-set nipples; two phalanges in both fifth fingers; impaired speech.

Char F. Peculiar facies with short philtrum, duck-bill lips, ptosis and low-set ear: a new syndrome. *Birth Defects* 1978; 14:303-305.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Duplication 14q Syndrome 392

General: Chromosomal 14q duplication syndrome.

Ocular: Hypertelorism; sparse eyelashes and eyebrows; slanted palpebral fissures; ocular colobomata.

Clinical: Postnatal growth retardation; mental retardation; hypotonia; microcephaly; nasal dysmorphism; tented lip; micrognathia; posteriorly rotated ears; minor skeletal anomalies.

Onwochei BC, et al. Ocular colobomata: major review. *Surv Ophthalmol* 2000; 45:175-194.

Sklower SL, et al. Distal duplication 14q: report of three cases and further delineation of the syndrome. *Hum Genet* 1984; 68:159-164.

Dyschondroplasia Syndrome (Ollier Syndrome; Enchondromatosis) 393

General: Chondrodysplasia in which ossification in the epiphyseal region is delayed or absent, with resulting continuation of excessive hypertrophic cartilage formation; dyschondroplasia associated with hemangiomas is referred to as Maffucci syndrome (see Maffucci Syndrome).

Ocular: Narrowing of the optic foramen and supraorbital fissure; ophthalmoplegia; optic atrophy, retinal pigmentation.

Clinical: Joint deformities with functional disturbances; coxa vara or valga; scoliosis; facial asymmetry; unilateral bone involvement with resulting shortening of the extremity; intracranial gliomas; intracavernous chondrosarcoma; clival chondroma.

Brazier DJ, et al. Intracavernous chondrosarcoma associated with Ollier's disease. *Br J Ophthalmol* 1993; 77: 599-600.

Chang S, Prados MD. Identical twins with Ollier's disease and intracranial gliomas: case report. *Neurosurgery* 1994; 34:903-906.

Duke-Elder S, ed. *System of Ophthalmology, vol. III, part 2.* St. Louis: CV Mosby, 1963:1068.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Ghogawala Z, et al. Clival chondroma in a child with Ollier's disease. Case report. *Pediatr Neurosurg* 1991; 17:53-56.

Ollier M. Sur une Nouvelle Affection: la Dyschondroplasie. *Rev Chir (Paris)* 1900; 21:396.

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Eales Disease (Periphlebitis) 394

General: Common; young adults.

Ocular: Sheathing of peripheral veins; hemorrhage in new vessels and later retinal detachment; retinal vascular tortuosity; microaneurysms of retina; postneovascularization of vitreous; internuclear ophthalmoplegia.

Clinical: Epilepsy and hemiplegia have been reported; chronic encephalitis; ulcerative colitis; central nervous infarction.

Atabay C, et al. Eales disease with internuclear ophthalmoplegia. *Ann Ophthalmol* 1992; 24:267-269.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Elliott A. J. 30-year observation of patients with Eales' disease. *Am J Ophthalmol* 1975; 80:404.

Katz B, et al. Eales' disease with central nervous infarction. *Ann Ophthalmol* 1991; 23:460-463.

Pomonis E, et al. Report of Eales' disease and ulcerative colitis in the same patient. *Am J Gastroenterol* 1992; 87: 1531-1532.

Weber F, Conrad B. Chronic encephalitis and Eales disease. *J Neurol* 1993; 240:299-391.

East-West Syndrome 395

General: Caused when an intraocular lens is placed so that the edge of the optic, a positioning hole, and the components of the loop-optic junction are well within the papillary aperture.

Ocular: Glare; halos; monocular diplopia; irregular pupil.

Clinical: None.

Brems RN, et al. Posterior chamber intraocular lenses in a series of 75 autopsy eyes. *J Cataract Refract Surg* 1986; 12:368-375.

Rosner M, et al. Optical aberrations from a well-centered intraocular lens implant. Am J Ophthalmol 1986; 101: 117-118.

Eaton-Lambert Syndrome (Myasthenic Syndrome; Ocular Myoclonus Syndrome; Myoclonic Syndrome) 396

General: Males; over 40 years of age; intrathoracic tumor; myasthenia-like condition less likely to have ocular manifestations; positive association with small cell cancer of the lung; underlying autoimmune basis.

Ocular: Decreased amplitude of version in all directions of gaze; ocular myoclonus; corneal abrasion; decreased corneal sensitivity; conjunctival injection; miotic pupil.

Clinical: Weakness; fatigue; peripheral paresthesia; dryness of mouth.

Cogan D. Myasthenia gravis. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:2489.

Eaton LM, Lambert EM. Electromyography and electric stimulation of nerves in diseases of the motor units; observations on myasthenic syndrome associated with malignant tumors. *J Am Med Assoc* 1957; 163:1117.

Fetell MR, Younger DS. Neurologic paraneoplastic syndromes. In: Rowland LP, ed. *Merritt's Textbook of Neurology,* 9th ed. Baltimore: Williams & Wilkins, 1995:935-936.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Penn AS. Lambert-Eaton syndrome. In: Rowland LP, ed. *Merritt's Textbook of Neurology*. 9th ed. Baltimore: Williams & Wilkins, 1995:761-763.

Sanders DB, Howard JF. Disorders of neuromuscular transmission. In: Bradley WG, et al. *Neurology in Clinical practice.* 2nd ed. Boston: Butterworth-Heinemann, 1995:1998-1999.

Eclampsia and Preeclampsia (Toxemia of Pregnancy; Preeclampsia) 397

General: Disorders of cells in glomeruli of kidneys that occur during gestation or shortly after delivery.

Ocular: Cortical blindness; nystagmus; mydriasis; absolute pupillary paralysis; ptosis; choroidal detachment; retinal detachment; cotton wool exudates; optic atrophy; retinal hemorrhages; petechial hemorrhages and focal edema in the occipital cortex.

Clinical: Hypertension; edema; proteinuria; convulsions; coma; death; cardiac failure; weight gain.

Barron WM. Hypertension: preeclampsia-eclampsia. In: Barron WM, Lindheimer MD, eds. *Medical Disorders During Pregnancy*. 2nd ed. St. Louis: Mosby, 1994:1-36.

Cunningham FG, et al. Blindness associated with preeclampsia and eclampsia. *Am J Obstet Gynecol* l995; 172[4 Pt 1]:1291-1298.

Donaldson JO. Neurological problems of pregnancy. In: Bradley WG, ed. *Neurology in Clinical practice*, 2nd ed. Boston: Butterworth-Heinemann, 1995:2069-2075.

Fastenberg DM, et al. Choroidal vascular changes ill toxemia of pregnancy. *Am J Ophthalmol* 1980; 89:362-368.

Gandhi J, et al. Blindness and retinal changes with preeclampsia toxemia. *NY State J Med* 1978; 78:1930-1932.

Halsey JH. Neurology of pregnancy. In: Rowland LP, ed. *Merritt's Textbook of Neurology*. 9th ed. Baltimore: Williams & Wilkins, 1995:962-966.

Hara N, et al. Postpartum eclampsia associated with cortical blindness. *Int J Gynaecol Obstet* 1994; 47:287-*288.*

Oliver M, Uchenik D. Bilateral exudative retinal detachment in eclampsia without hypertensive retinopathy. *Am J Ophthalmol* 1980; 90:792-796.

Shukla BK, Prasad GN. Retinal changes in eclampsia and preeclampsia. *J Ind Med Assoc* 1976; 66:8-10.

Ectopia Lentis with Ectopia of Pupil (Ectopia Lentis et Pupillae) 398

General: Autosomal recessive.

Ocular: Lens and pupil displaced in opposite directions; bilateral cataracts; acute intermittent intraocular pressure crises; persistent pupillary membrane; poor pupillary dilation.

Clinical: None.

Colley A, et al. Ectopia lentis et pupillae: the genetics aspects and differential diagnosis. *J Med Genet* 1991; 28: 791-794.

Cruysberg JR, Pinckers A. Ectopia lentis et pupillae syndrome in three generations. *Br J Ophthalmol* 1995; 79: 135-138.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Townes PL. Ectopia lentis et pupillae. *Arch Ophthalmol* 1976; 94: 1126-1128.

Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome (EEC Syndrome) 399

General: Autosomal dominant; low penetrance; variable expressivity.

Ocular: Dacryocystitis; photophobia; corneal ulceration; blepharophimosis; atresia or absence of lacrimal puncta; strabismus; decreased visual acuity.

Clinical: Cleft lip; cleft palate; abnormalities of the urinary tract, hands, feet, and nail hypoplasia, granulomatous perlèche; scalp dermatitis.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Mawhorter LG, et al. Keratopathy in a family with the ectrodactyly-ectodermal dysplasia-clefting syndrome. *Ophthalmology* 1985; 92:1427-1431.

Trueb RM, et al. Scalp dermatitis, distinctive hair abnormalities and atopic disease in the ectrodactyly-ectodermal dysplasia-clefting syndrome. *Br J Dermatol* 1995; 132:621-625.

Ehlers-Danlos Syndrome (Fibrodysplasia Elastica Generalisata; Cutis Hyperelastica; Meekeren-Ehlers-Danlos Syndrome; Indian Rubber Man Syndrome; Cutis Laxa) 400

General: Present at birth; autosomal dominant; two groups: (i) cutaneous and (ii) articular; syndrome is one of three primary disorders of elastic tissue (other two are pseudoxanthoma elasticum [Grönblad-Strandberg syndrome] and senile elastosis); inherited disorder of collagen biosynthesis.

Ocular: Hyperelasticity of palpebral skin; easy eversion of upper lid; ptosis; epicanthal folds; hypotony of extraocular muscles; strabismus; microcornea; thinning of cornea with keratoconus; thinning of sclera (blue sclera); subluxation of lens; angioid streaks; chorioretinal hemorrhages; retinitis proliferans with secondary detachment; macular degeneration; myopia; ruptured globe after minor trauma; limbus-to-limbus corneal thinning; acute hydrops; cornea plana; keratoglobus.

Clinical: Cutaneous manifestations include thin, atrophic, fragile skin, cutaneous hyperelasticity, and pseudomolluscoid tumors; articular manifestations include excessive articular laxity and luxations; hypermobile joints.

Ainsworth SR, Aulicino PL. A survey of patients with Ehlers-Danlos syndrome. *Clin Orthop Rel Res* 1993; 286: 250-256.

Cameron JA. Corneal abnormalities in Ehlers-Danlos syndrome type VI. *Cornea* 1993; 12:54-59.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Danlos H. Un Cas de Cutis Laxa avec Tumeurs par Contusion Chronique des Caudes et des Genoux. *Bull Soc Fr Dermatol Syph* 1908; 19:70.

Ehlers E. Cutis Laxa, Neigung zu Hoemorrhagien in der Haut, Lockerung mehrerer Artikulationen. *Dermatol Zr* 1901; 8:173.

Farley MK, et al. Spontaneous carotid-cavernous fistula and the Ehlers-Danlos syndromes. *Am Acad Ophthalmol* 1983; 90:1337-1341.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Pesudovs K: Orbscan mapping in Ehlers-Danlos syndrome. *J Cataract Refract Surg* 2004; 30: 1795-1798.

Prockop DJ, Kuivaniemi H, Tromp G. Heritable disorders of connective tissue. Ehlers-Danlos syndrome. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:2113-2114.

18p- Syndrome (18p Deletion Syndrome) 401

General: Chromosome 18p deletion syndrome.

Ocular: Hypertelorism, epicanthus, horizontal palpebral fissures.

Clinical: Microcephaly, round face, broad-based nose, "carp mouth," microretrognathic; pterygium colli; dysplastic and low-set ears; clinodactyly; failure to grow; muscular hypotony; mental retardation; hypoplastic male genitalia.

Gocke H, Muradow I, Stein W The fetal phenotype of the 18p- syndrome. Report of a male fetus at twenty-one weeks. *Ann Genet* 1988; 31:60-64.

Zumel RM, et al. The 18p- syndrome. Report of five cases. *Ann Genet* 1989; 32:160-163.

18q- Syndrome (18q Deletion Syndrome) 402

General: Chromosome 18q deletion syndrome.

Ocular: Macular "fibrosis"; Optic disk abnormalities with tractional retinal detachment, retinal degeneration, and tilting of the optic disk.

Clinical: Microcephaly; short stature; hypotonia; hypothyroidism; diabetes mellitus; short neck; sensorineural hearing loss; sensorimotor axonal neuropathy; mild-to-moderate mental retardation; chronic arthritis; seizures.

Gordon MF, et al. Dystonia in a patient with deletion of 18q. *Mov Disord* 1995; 10:496-499.

Hansen US, Herline T. Chronic arthritis in a boy with 18q- syndrome. *J Rheumatol* 1994; 21:1958-1959.

Smith A, Caradus V, Henry JG. Translocation 46X6 t(17;18)(q25;q21) in a mentally retarded boy with progressive eye abnormalities. *Clin Genet* 1979; 16:156-162.

Eldridge Syndrome 403

General: Autosomal recessive inheritance; also may involve an enzyme deficiency.

Ocular: Myopia, onset 4 to 6 years of age; increased retinal translucency; temporal crescents; mild electroretinographic abnormalities.

Clinical: Severe myopia; sensorineural hearing loss; low intelligence; mild renal disease.

Eldridge RC, et al. Cochlear deafness, myopia and intellectual impairment in an Amish family. *Arch Otolaryngol* 1968; 88:49-54.

Ohlsson L. Congenital renal disease, deafness and myopia in one family. *Arch Med Scand* 1963; 174:77-83.

Electrical Injury 404

General: Electric current passes through the body; voltage ranging from 100 to 200 million volts may cause electrical burns.

Ocular: Choroidal atrophy; corneal perforation; necrosis of cornea or lids; blepharospasm; anterior or posterior subcapsular cataracts and vacuoles; optic neuritis; optic nerve atrophy; retinal edema; retinal hemorrhage; pigmentary degeneration; retinal holes; anterior uveitis; hyphema; hypotony; glaucoma; night blindness; nystagmus; paralysis of extraocular muscles; visual field defects; dilation of retinal veins.

Clinical: Skin burns; injury to cardiovascular, central nervous, and musculoskeletal systems; tissue necrosis; vascular injury.

Browne BJ, Gaasch WR. Electrical injuries and lightning. *Emerg Med Clin North Am* 1992; 10:211-229.

Danielsen L, et al. Diagnosis of electrical skill injuries. A review and description of a case. *Am J Forensic Med Pathol* 1991; 12:222-226.

Fraunfelder FT, Hanna C. Electric cataracts. I. Sequential changes, unusual and prognostic findings. *Arch Ophthalmol* 1972; 87:179-183.

Hanna C, Fraunfelder FT. Electric cataracts. II. Ultrastructural lens changes. *Arch Ophthalmol* 1972; 87:184-191.

11q- Syndrome 405

General: Chromosome II deletion syndrome.

Ocular: Telecanthus/hypertelorism; rarely, congenital glaucoma, cyclopia.

Clinical: Psychomotor retardation, trigonocephaly, broad depressed nasal bridge, micrognathia, low-set abnormal ears, cardiac anomalies, hand and foot anomalies, renal agenesis, anal atresia, supratentorial white matter abnormality on computed tomography or magnetic resonance imaging; microphallus; holoprosencephaly; female preponderance.

Helmuth RA, Weaver DD, Wills ER. Holoprosencephaly, ear abnormalities, congenital heart defect, and microphallus in a patient with 11q-mosaicism. *Am J Med Genet* 1989; 32:178-181.

Ishida Y, et al. The 11q- syndrome with mosaic partial deletion of 11q. *Acta Paediatr Jpn* 1992; 34:592-596.

Leegte B, et al. 11q- syndrome: three cases and a review of the literature. *Genet Couns* 1999; 10:305-313.

Ellis-Van Creveld Syndrome (Chondroectodermal Dysplasia) 406

General: Autosomal recessive inheritance; occurs in the Amish; associated with *de novo* chromosomal abnormality: deletion of 12 (p11.21p12.2).

Ocular: Esotropia; iris coloboma; congenital cataract.

Clinical: Bilateral polydactyly; short and plump limbs; genu valgum; talipes (equinovarus, calcaneovalgus); thoracic constriction; fusion of middle part of upper lip to maxillary gingival margin; dental anomalies: number, shape, spacing; congenital heart defect in about 50% of patients; dystrophic fingernails; genital anomalies; mild mental retardation; short stature; hypoplastic hair and skin; oligodontia; small thoracic cage; hypoplastic pelvis; cone-shaped epiphyses of hands.

Ellis RWB, van Creveld S. A syndrome characterized by polydactyly, chondro-dysplasia and congenital morbus cordis. *Arch Dis Child* 1940; 15:65.

Nagai T, et al. Del (12) (p11.21p12.2) associated with an asphyxiating thoracic dystrophy or chondroectodermal dysplasia-like syndrome. *Am J Med Genet* 1995; 55:16-18.

Wynne-Davies R. Genetics and malformations of the hand. *Hand* 1971; 3:184-192.

Elschnig Syndrome (Elschnig Syndrome II) 407

General: Present from birth; etiology unknown.

Ocular: Elongation of lid fissure with downward displacement of the lateral angle; ectropion of lower lid.

Clinical: Cleft lip and palate may occur.

Elschnig A. Zur Kenntnis der Anomalien der Lidspalten Form. *Klin Monatsbl Augenheilkd* 1912; 17-30.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981:253.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Elschnig Syndrome I (Meibomian Conjunctivitis) 408

General: Chronic inflammations; characteristic foamy secretion; benign.

Ocular: Conjunctivitis; foamy secretion; ocular irritation; photophobia; minimal visual impairment.

Clinical: Hyperplasia of tarsal glands.

Elschnig A. Belt ray Artiologie und Therapie der chronischen Conjunctivitis. *Dtsch Med Wochenschr* 1908; 34: 1133-1135.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Empty Sella Syndrome 409

General: Further progression of ocular findings and symptoms after treatment of pituitary tumors.

Ocular: Reduced visual acuity and possible blindness; hemianopsia; quadranopsia; irregular field defects; central scotoma; pale optic disks; central retinal vein occlusion.

Clinical: Acromegalic features and other general systemic manifestations depend on the type of the primary tumor the patient had and are not part of the "empty sella syndrome" that is responsible for progression of ocular pathologic condition.

Battaglia Parodi M, et al. Primary empty sella syndrome and central retinal vein occlusion. *Ophthalmologica* 1995; 209:106-108.

Colby MY Jr, Kearns TP. Radiation therapy of pituitary adenomas with associated visual impairment. *Mayo Clin Proc* 1962; 37:15-24.

Dahlstrom R, Acers TE. Chiasmatic arachnoiditis and empty sella: report and discussion of a case. *Ann Ophthalmol* 1975; 7:73.

Pollock SC, Bromberg BS. Visual loss in a patient with primary empty sella. *Arch Ophthalmol* 1987; 105:1487-1488.

Encephalitis, Acute 410

General: In approximately 0.1% to 0.2% of patients having rubeola (measles), an acute encephalitis is seen within 1 week after the onset of the rash; a case of immunosuppressive encephalitis can present with focal seizures leading to progressive obtundation.

Ocular: Papillitis; optic atrophy; ocular motor palsies; nystagmus; optic neuritis or neuroretinitis.

Clinical: Rise in temperature; drowsiness; irritability; meningismus; vomiting and headache; stupor; convulsions; coma.

Chen RE, et al. Immunosuppressive measles encephalitis. *Pediatr Neurol* 1994; 10:325-327.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Francois J. Ocular manifestations in demyelinating diseases. *Adv Ophthalmol* 1979; 39:1-36.

Endocarditis 411

General: Common in people with arteriosclerosis; subacute form leads to pyemia; bacterial endocarditis without heart murmur seen in intravenous drug users.

Ocular: Retinal hemorrhages; conjunctival petechiae; choroiditis; Roth spots; spastic mydriasis; optic neuritis; central retinal artery occlusion; choroidal abscess, subretinal neovascularization.

Clinical: Heart murmur; fever; intracranial aneurysm; cerebral hemorrhage; subarachnoid hemorrhage.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Coll GE, Lewis H. Metastatic choroidal abscess and choroidal neovascular membrane associated with Staphylococcus aureus endocarditis in a heroin user. *Retina* 1994; 14:256-259.

Etienne J, et al. Actinobacillus endocarditis. *Med J Aust* 1986; 144:54-55.

Munier F, Othenin-Girard P. Subretinal neovascularization secondary to choroidal septic metastasis from acute bacterial endocarditis. *Retina* 1992; 12:108-112.

Okada AA, et al. Endogenous bacterial endophthalmitis. Report of a ten-year retrospective study. *Ophthalmology* 1994; 101:832-838.

Engelmann Syndrome (Osteopathia Hyperostotica [Scleroticans] Multiplex Infantilis; Diaphyseal Dysplasia; Camurati-Engelmann Disease; Hereditary Multiple Diaphyseal Sclerosis; Juvenile Paget Disease) 412

General: Etiology unknown; progressive resorption and deposits of bone with thickening of periosteum and changes of cortex as evident by diagnostic x-ray studies in the intermediate portion of the long bones.

Ocular: Exophthalmos; hypertelorism (secondary); ptosis; lagophthalmos; lateral rectus palsy; convergence insufficiency; epiphora; cataract; tortuous retinal vessels; papilledema; optic atrophy.

Clinical: Pain in extremities; poorly developed musculature; waddling gait; delayed ambulation; scaly skin; delayed dentition; deafness; hypogonadism; pain in both legs; aching in the forearms; episodic temporofrontal and occipital headache.

Brodrick JD. Luxation of the globe in Engelmann's disease. *Am J Ophthalmol* 1977; 83:870-873.

DeVits A, et al. Progressive diaphyseal dysplasia (Camurati-Engelmann's disease). Improvement of clinical signs and of bone scintigraphy during pregnancy. *Clin Nucl Med* 1994; 19:104-107.

Engelmann G. Ein Fall von Osteopathia Hyperostotica (Scleroticans) Infantilis. *Fortschr Geb Roentgen Strahl* 1929; 39:1101.

Morse PH, et al. Ocular findings in hereditary diaphyseal dysplasia (Engelmann's disease). *Am J Ophthalmol* 1969; 68:100.

Fraunfelder FT, Roy FH, eds. *Current Ocular Therapy.* 5th ed. Philadelphia: WB Saunders, 2000:247-248.

Enterobiasis (Oxyuriasis; Pinworm; Seatworm) 413

General: Intestinal infection caused by *Enterobius vermicularis;* worm's head attached to cecal mucosa, appendix, or parts of bowel; worms travel anal canal and deposit eggs on perianal skin; eggs infective for 10 to 20 days; airborne transmission; common in children; extraintestinal pinworm infection has been reported.

Ocular: Palpebral edema; blepharitis; keratoconjunctivitis; macular edema.

Clinical: Pruritus; eczema; pyogenic infection; vaginal discharge; chronic granulomatous salpingitis; endometritis.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Duke-Elder S, Scott GI, eds. *System of Ophthalmology. vol. XIII.* St. Louis: CV Mosby, 1971.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology,* 4th ed. Baltimore: Williams & Wilkins, 1987.

Russell LJ. The pinworm, Enterobius vermicularis. *Prim Care Clin Office Pract* 1991; 18:13-24.

Tornieporth NG, et al. Ectopic enterobiasis: a case report and review. *J Infect* 1992; 24:87-90.

Wiebe BM. Appendicitis and Enterobius vermicularis. *Scand J Gastroenterol* 1991; 26:336-338.

Epiblepharon 414

General: Autosomal dominant; occurs predominantly in Chinese individuals.

Ocular: Epicanthus; epiblepharon of upper and lower lids.

Clinical: None.

Hu DN. Ophthalmic genetics in China. *Ophthalmic Paediatr Genet* 1983; 2:39-45.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Epicanthus 415

General: Autosomal dominant.

Ocular: Epicanthus; epiblepharon of upper and lower lids; ptosis.

Clinical: None.

Hu DN. Ophthalmic genetics in China. *Ophthalmic Paediatr Genet* 1983; 2:39-45.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Epidemic Keratoconjunctivitis 416

General: Highly communicable; adenovirus types 8 and 19; usually bilateral; epidemic keratoconjunctivitis has been reported worldwide associated with 11 virus serotypes, with serotypes 8, 11, and 19 being the most common responsible ones.

Ocular: Follicular or membranous conjunctivitis; chemosis; subconjunctival hemorrhages; corneal opacity; punctate epithelial keratitis; corneal ulcer; blepharospasm; lid edema; serous discharge; uveitis; epiphora.

Clinical: Submaxillary and cervical lymphadenopathy.

Fraunfelder FT, Roy FH, eds. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000:93.

Abel R Jr. Adenovirus keratoconjunctivitis and new approaches to prophylaxis. *Ann Ophthalmol* 1977; 9:13.

Murrah W. Epidemic keratoconjunctivitis. *Ann Ophthalmol* 1988; 20:36.

Pavan-Langston D. Viral disease of the cornea and external eye. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. I* Philadelphia: WB Saunders, 1994:150.

Sprague JB, et al. Epidemic keratoconjunctivitis: a severe industrial outbreak due to adenovirus type 8. *N Engl J Med* 1973 ;289: 1341-1346.

Epidermal Nevus Syndrome (Ichthyosis Hystrix) 417

General: One or a combination of the following epidermal nevi described as nevus unius lateris, ichthyosis hystrix, linear nevus sebaceous, or congenital acanthosis nigricans; autosomal dominant.

Ocular: Blepharoptosis and fibroma on bulbar conjunctiva; antimongoloid eyelid fissures; eyelid colobomata; horizontal and rotary nystagmus; esotropia; conjunctival tumors; corneal opacities; corectopia and colobomata of the iris.

Clinical: Somatic anomalies involving the skeletal and central nervous system; anomalies of bone formation; atrophy; ankylosis; vitamin D-resistant rickets; bone cysts; mental retardation; cortical atrophy; hydrocephalus; focal and grand mal epilepsy; cerebrovascular tumors; cortical blindness.

Benjamin SN, Allen HF. Classification for limbal dermoid choristomas and branchial arch anomalies. *Arch Ophthalmol* 1972; 87:305-314.

Burch JV, et al. Ichthyosis hystrix (epidermal nevus syndrome) and Coat's disease. *Am J Ophthalmol* 1980; 89:25-30.

Fenichel GM. Linear nevus sebaceous syndrome. In: Fenichel GM, ed. *Clinical pediatric neurology,* 2nd ed. Philadelphia: WB Saunders, 1993:370-371.

Goldberg LH, Collins SA, Siegel DM. The epidermal nevus syndrome: case report and review. *Pediatr Dermatol* 1987; 4:27-33.

Greenberg BM, Pess GM, May JW Jr. Macrodactyly and the epidermal nevus syndrome. *J Hand Surg [Am]* 1987; 12[5 Pt 1]:730-733.

Mullaney PB, Weatherhead RG: Epidermal nevus syndrome associated with a complex choristoma and a bilateral choroidal osteoma. *Arch Ophthalmol* 1996; 114: 1292-1291.

Epilepsy, Light-Sensitive 418

General: Autosomal dominant.

Ocular: Photic stimulation of 15 to 30 flashes per second, inducing epileptic seizure.

Clinical: Spastic paraparesis; mental retardation; light sensitivity.

Davidson S, Watson CW. Hereditary light sensitive epilepsy. *Neurology* 1956; 6:235-261.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Epiphyseal Dysplasia of Femoral Heads, Myopia, Deafness 419

General: Autosomal recessive.

Ocular: Severe myopia.

Clinical: Femoral epiphyseal dysplasia; deafness.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Pfeiffer RA, et al. Epiphyseal dysplasia of the femoral head, severe myopia, and perceptive hearing loss in 3 brothers. *Clin Genet* 1973; 4:141-144.

Epiphyseal Dysplasia, Microcephaly, and Nystagmus 420

General: Autosomal recessive.

Ocular: Nystagmus; retinitis pigmentosa.

Clinical: Epiphyseal dysplasia; microcephaly; short stature.

Lowry RB, et al. Epiphyseal dysplasia, microcephaly, nystagmus and retinitis pigmentosa. *Am J Med Genet* 1989; 33: 341-345.

Lowry RB, Wood BJ. Syndrome of epiphyseal dysplasia, short stature, microcephaly, and nystagmus. *Clin Genet* 1975; 8:269-274.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Epiphyseal Dysplasia, Multiple, with Myopia and Conductive Deafness 421

General: Autosomal dominant; onset early adulthood of severe osteoarthritis of hips; deficiency of distal tibial ossification seen in children results in sloping end of tibia in adulthood.

Ocular: Progressive myopia; retinal thinning; cataracts.

Clinical: Osteoarthritis of hips; short stature; brachydactyly, hyperextensibility of fingers; widening of joint spaces; conductive deafness.

Beighton P, et al. Dominant inheritance of multiple epiphyseal dysplasia, myopia, and deafness. *Clin Genet* 1978; 14:173-177.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Episkopi Blindness 422

General: Sex-linked; confined to male members of Greek Cypriot family group, most of whom live in Episkopi.

Ocular: Microphthalmia; corneal opacities; transverse corneal band; iritis; cataract; retrolental opacities; retinitis pigmentosa; Leber optic atrophy; amaurosis.

Clinical: None.

Roy FH. *Ocular Differential Diagnosis*, 7th ed. Philadelphia: Lippincott Williams & Wilkins, 2002.

Taylor PJ, et al. Episkopi blindness: hereditary blindness in a Greek Cypriot family. *Br J Ophthalmol* 1959; 43:340-344.

Epithelial Erosion Syndrome (Metaherpetic Keratitis; Kaufman Syndrome; Franceschetti Dystrophy; Posttraumatic Keratitis) 423

General: Most likely caused by herpes simplex virus; previous corneal trauma or autosomal dominant.

Ocular: Recurrent erosions of the corneal epithelium, usually seen within weeks or months after herpes simplex infection of the cornea; "loose" epithelium is removed from the underlying Bowman membrane mechanically by lid blinking; defects are irregular in shape and stain positively with fluorescein dye; underlying corneal stroma usually shows some edema; pain upon opening eyes in morning.

Clinical: Mild fever; occasionally herpetic skin lesions.

Aitken DA, et al. Ultrastructural study of the corneal epithelium in the recurrent erosion syndrome. *Br J Ophthalmol* 1995; 79:282-289.

Azar DT, Scally A, Hannush SB,et al. Epithelial-defect-masquerade syndrome after laser in situ keratomileusis. *J Cataract Refract Surg* 2003; 29: 2358-2365

Gunderson T. Herpes corneae: with special reference to its treatment with strong solutions of iodine. *Arch Ophthalmol* 1936; 15:225.

Huang AJ, Tseng SC. Corneal epithelial wound healing in the absence of limbal epithelium. *Invest Ophthalmol Vis Sci* 1991; 32:96-105.

Kaufman HE. Epithelial erosion syndrome: metaherpetic keratitis. *Am J Ophthalmol 1964; 57:983.*

Erb-Goldflam Syndrome (Erb II Syndrome; Hoppe-Goldflam Disease; Pseudoparalytic Syndrome; Myasthenia Gravis) 424

General: Occurs at any age; more frequent between ages 20 and 40 years; more females affected than males; progressive; spontaneous; symptoms improve or resolve with rest in early stages of disease (see Myasthenia Gravis, Neonatal or Infantile); caused by autoantibodies against the acetylcholine receptor at the neuromuscular junction, leading to abnormal fatigability and weakness of skeletal muscle.

Ocular: Transient diplopia; ptosis of upper eyelids.

Clinical: Excessive fatigability of musculature; symptoms appear and increase as day progresses; expressionless face; sagging jaw; difficulty in chewing and talking; nasal regurgitation.

Erb W. Zur Casuistick der Bulbaren La hmungen. *Arch Psychiatr Vervenkr* 1879; 9:325-350.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Goldflam S. Vebereinen Scheinbar Keilbaren Bulbarparalytischem Symptom Complex mit Betheiligung der Extremitaten. *Dtsch* Z *Nerven* 1983; 4:312-352.

Kim JH, Hwang JM, Hwang YS, et al. Chilhood ocular myasthenia gravis. *Ophthalmology* 2003; 110: 1458-1462.

Lepore FE. Pupillary dysfunction in myasthenia gravis. *Ann Neurol* 1979; 6:29-33.

Sommer N, et al. Ocular myasthenia gravis. A critical review of *Clinical* and pathophysiological aspects. *Doc Ophthalmol* 1993; 84:309-333.

Erosion Syndrome 425

General: Caused by imperfect adherence of corneal epithelium due to abnormalities in basement membrane; abnormalities may be inherent, induced by trauma, or both; disease not vision threatening.

Ocular: Disabling episodes of pain; stabbing pain in the eye like that from a foreign body, most frequent on awakening; lesser forms of irritation; blurring of vision; drying associated with a stinging in the eyes; irregular astigmatism; corneal findings include anterior membrane dystrophies (epithelial and subepithelial dot, map, or fingerprint-type changes).

Clinical: None.

Aitken DA, et al. Ultrastructural study of the corneal epithelium in the recurrent erosion syndrome. *Br J Ophthalmol* 1995; 79:282-289.

Brown N, Bron A. Recurrent erosion of the cornea. *Br J Ophthalmol* 1976; 60:84.

Langston RH, et al. Soft lens therapy for recurrent erosion syndrome. *Ann Ophthalmol* 1978; 10:875-878.

Erythema Nodosum (Dermatitis Contusiformis) 426

General: Young females; hypersensitive reaction secondary to viral, bacterial, and fungal infections; duration 2 to 4 weeks; recurrences possible.

Ocular: Subcutaneous nodules involving lids; keratitis; uveitis.

Clinical: Painful nodules on surface of thighs, arms, and face; fever; malaise; red lesions that progress to bruiselike and disappear in a few days to 3 weeks; cervical lymphadenopathy; exquisitely tender, erythematous nodules distributed symmetrically on the extensor surfaces of the lower extremities.

Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine*, 11th ed. New York: McGraw-Hill, 1987.

Fox MD, Schwartz RA. Erythema nodosum. *Am Fam Physician* 1992; 46:818-822.

Novick NL, et al. Erythema dyschromicum perstans. *Int J Dermatol* 1986; 25:322-323.

Escherichia Coli 427

General: Gram-negative rod found in the gastrointestinal tract; urinary tract is the usual portal of entry.

Ocular: Uveitis; hyphema; hypopyon; gas bubbles in anterior chamber; purulent conjunctivitis; keratitis; corneal edema; panophthalmitis; endophthalmitis; glaucoma.

Clinical: Diarrhea; gastroenteritis; dehydration.

Fraunfelder FT, Roy FH.: *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Krachmer JH, Purcell JJ.: Bacterial corneal ulcers in cosmetic soft contact lens wearers. *Arch Ophthalmol* 1978; 96: 57-61.

Lissner GS, Romano PE. Pneumatosis oculi and spontaneous hyphema in association with pneumatosis intestinalis. *Am J Ophthalmol* 1979; 88:708-713.

Parl SB, et al. Endogenous endophthalmitis caused by Escherichia coli. *Ann Ophthalmol* 1993; 25:95-99.

Espildora-Luque Syndrome (Ophthalmic Sylvian Syndrome) 428

General: Embolism of the ophthalmic artery with refectory spasm of the middle cerebral artery.

Ocular: Unilateral blindness (caused by ophthalmic artery embolism).

Clinical: Temporary hemiplegia contralateral side of amaurosis (caused by reflex spasm of the middle cerebral artery).

Espildora-Luque C. Ophthalmic sylvian syndrome. *Arch Oft Hisp Am* 1934; 34:616; *Am J Ophthalmol* 1935; 18: 402(abst).

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Ethan Syndrome, Primary 429

General: Congenital, esotropia, head turn, and nystagmus coexistent with nystagmus compensation and nystagmus blockage syndrome (see Nystagmus Compensation Syndrome; Nystagmus Blockage Syndrome).

Ocular: Esotropia; esophoria; nystagmus; amblyopia; orthophoria.

Clinical: Head turn and chin elevation to compensate for nystagmus.

Isenberg SJ, Yee RD. The Ethan syndrome. *Ann Ophthalmol* 1986; 18:358-361.

Robb RM. Periodic alternation of null point in congenital nystagmus. *Arch Ophthalmol* 1972; 87:169-172.

Ethan Syndrome, Secondary 430

General: Classic nystagmus blockage syndrome, but after strabismus surgery development of head turn with straight eyes and appearance of nystagmus compensation syndrome (see Nystagmus Blockage Syndrome; Nystagmus Compensation Syndrome).

Ocular: Esotropia; nystagmus; orthophoria; amblyopia; nystagmus increased in abduction.

Clinical: Abnormal head position.

Isenberg SJ, Yee RD. The Ethan syndrome. *Ann Ophthalmol* 1986; 18:358-361.

von Noorden GK. The nystagmus compensation (blockage) syndrome. *Am J Ophthalmol* 1976; 82:287.

Ewing Sarcoma (Ewing Syndrome) 431

General: Highly metastatic round cell tumor of bone; most commonly involves long or trunk bones; metastasizes at high rate; usually occurs between ages 10 and 25 years; seen more frequently in males than in females.

Ocular: Exophthalmos; orbital hemorrhages; orbital necrosis; commonly found as the second malignancy in patients with hereditary retinoblastoma.

Clinical: Lytic bone destruction; pain; edema; slight fever.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology*, 4th ed. Baltimore: Williams & Wilkins, 1987.

Smith LM, Donaldson SS. Incidence and management of secondary malignancies in patients with retinoblastoma and Ewing's sarcoma. *Oncology* 1991; 5:135-141.

Exfoliation Syndrome (Capsular Exfoliation Syndrome) 432

General: Only in men older than 60 years.

Ocular: Iridodonesis; rubeosis iridis; cataract; phacodonesis; dislocated lens; corneal dystrophy; choroidal sclerosis; primary optic atrophy; lens capsule exfoliation; lower endothelial cell density.

Clinical: None.

Eagle RC Jr, et al. The basement membrane exfoliation syndrome. *Arch Ophthalmol* 1979; 97:510-515.

Layden WE, Shaffer RN. The exfoliation syndrome. *Trans Am Acad Ophthalmol Otolaryngol* 1974; 78:326.

Puska P. Lens opacity in unilateral exfoliation syndrome with or without glaucoma. *Acta Ophthalmol* 1994; 72:290-296.

Seland JH, Chylack LT. Cataracts in the exfoliation syndrome. *Trans Ophthalmol Soc UK* 1982; 102:375-379.

Sood GL, et al. Capsular exfoliation syndrome. *Br J Ophthalmol* 1973; 57:120.

Stefaniotou M, et al. The cornea in exfoliation syndrome. *Doc Ophthalmol* 1992; 80:329-333.

Extreme Hydrocephalus Syndrome (Kleeblattschädel Syndrome; Cloverleaf Skull Syndrome; Hydrocephalus; Chondrodystrophicus Congenita) 433

General: Secondary obstruction of cerebrospinal fluid circulation caused by some primary disease such as maternal rubella, Rh incompatibility, or hydramnion; Arnold- Chiari syndrome has similar associated findings; almost all affected children are born dead.

Ocular: Exophthalmos with downward placement and downward rotation of the globes; propulsion of globes; upper lid retraction and lower lids covering almost half of the downwardly rotated cornea; nystagmus; strabismus; exposure keratitis; optic nerve atrophy.

Clinical: Extreme hydrocephalus; low-set ears; thin and spastic extremities with digital anomalies; convulsions; spina bifida.

Holtermuller K, Wiedemann HR. Kleeblattschadel syndrome. *Med Wschr* 1960; 14:439.

Resnick DK, et al. Surgical management of the cloverleaf skull deformity. *Pediatr Neurosurg* 1995; 22:29-37.

Rogers GL, et al. The management of the Kleeblattschadel syndrome. *Ann Ophthalmol* 1981; 13: 1173-1175.

Shiroyama Y, et al. The relationship of cloverleaf skull syndrome to hydrocephalus. *Child Nerv Syst* 1991; 7:382-385.

Walters EC, et al. Cloverleaf skull syndrome. *Am J Ophthalmol* 1973; 76:716.

Eyebrow Whorl 434

General: Autosomal dominant.

Ocular: Whorl in the hair of the eyebrow; myopia, telecanthus, hypertelorism.

Clinical: Deafness, proteinuria.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Virchow H. Stellung der Haare im Brauenkopf. Z *Ohtnol* 1912; 44:402-403.

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Fabry Disease (Angiokeratoma Corporis Diffusum Syndrome; Diffuse Angiokeratosis; Fabry-Anderson Syndrome; Glycosphingolipid Lipidosis; Glycosphingolipidosis) 435

General: Lipoid storage disorder; X-linked recessive inheritance; lack of α-galactosidase A enzyme.

Ocular: Swelling of eyelids; varicosities of palpebral and bulbar conjunctiva; corneal dystrophy; corneal opacities; increased tortuosity of retinal vessels and aneurismal dilatations; cornea verticillata; cataract; central retinal artery occlusion; internuclear paralysis of extraocular muscles; papilledema; tortuosity and caliber irregularity of conjunctival vessels; characteristic cream-colored whorllike opacity in deep part of corneal epithelium; posterior cataract; occasional edema of optic disk and reitina.

Clinical: Angiokeratoma of the skin with small, grouped papular lesions mainly over the scrotum, thighs, buttocks, sacral area, umbilical area, and lips; elevated blood pressure; disturbance in sweat secretion; pain in arms and legs; enlarged heart; albuminuria.

Andersen MV, et al. Central retinal artery occlusion in a patient with Fabry's disease documented by scanning laser ophthalmoscopy. *Acta Ophthalmol* 1994; 72:635-638.

Clarke JTR, et al. Enzyme replacement therapy by renal allotransplantation in Fabry's disease. *N Engl J Med* 1972; 287:1215-1218.

Ho PC, Feman SS. Internuclear ophthalmoplegia in Fabry's disease. *Ann Ophthalmol* 1981; 13:949-951.

Fraunfelder FT, Roy FH, eds. *Current Ocular Therapy.* 4th ed. Philadelphia: WB Saunders, 2000:180.

Weingeist TA, Blodi FC. Fabry's disease: ocular findings in a female carrier. *Arch Ophthalmol* 1971; 85:169.

Facio-Oculo-Acoustico-Renal Syndrome 436

General: Autosomal recessive.

Ocular: Congenital myopia; undeveloped filtration angle; persistent pupillary remnant membrane; hypertelorism; dysplasia carthonum; antimongoloid obliquity of palpebral fissure.

Clinical: Large head; sensorineural hearing loss; proteinuria; epiphyseal dysplasia of the femoral heads.

Fraser GR. The causes of profound deafness in childhood. A study of 3535 individuals with severe auditory handicaps present at birth or of childhood onset. Baltimore: The Johns Hopkins University Press, 1976.

Holmes LB, Schepens LC. Syndrome of facial anomalies, telecanthus and deafness. J Pediatr 1972; 81:552-555.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Murdock JL, Mengel MC. An unusual eye-ear syndrome with renal anomaly. *Birth Defects* 1971; 7:136.

Facio-Scapulo-Humeral Muscular Dystrophy (FSH Muscular Dystrophy) 437

General: Autosomal dominant disorder; onset varies from infancy to old age; severity varies from scarcely detectable to incapacitating; recessive inheritance has been reported.

Ocular: Retinal telangiectasis; macular lesion; macular edema; retinal sea fans.

Clinical: Deafness; wasting of shoulder girdle, upper deltoid, pectoralis, biceps, and triceps; difficulty whistling, drinking through straws, and playing wind instruments; foot dragging; mental retardation.

Brouwer OF, et al. Hearing loss in facioscapulohumeral muscular dystrophy. *Neurology* 1991; 41:1878-1881.

Gurwin EB, et al. Retinal telangiectasis in facioscapulohumeral muscular dystrophy with deafness. *Arch Ophthalmol* 1985; 103:1695-1700.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Meyerson MD, Lewis EK. Facioscapulohumeral muscular dystrophy and accompanying hearing loss. *Arch Otolaryngol* 1984; 110:261-266.

Falciform Detachment 438

General: Autosomal dominant or recessive; preperinatally acquired; characterized by ocular signs only; falciform detachment and congenital total detachment may alternate in affected siblings; falciform detachment and folds; retina projects as a wedge-shaped fold from the posterior pole of eye into the vitreous, occasionally as far anterior as the lens; less typical fold flattens and tapers out in the midperiphery of the retina.

Ocular: Falciform folds; retinal detachment; retrolental fibroplasia.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Warburg M. Heterogeneity of congenital retinal non-attachment falciform folds and retinal dysplasia: a guide to genetic counseling. *Hum Hered* 1976; 26:137-148.

Falciform Detachment with Microphthalmia and Microcephaly 439

General: Rare; both sexes affected; etiology unknown.

Ocular: Microphthalmia; congenital cataract; corneal opacities; falciform folds; glaucoma; buphthalmos; congenital detachment of retina; vitreous hemorrhages; persistent hyperplastic primary vitreous; retinal neovascularization; retinal dysplasia.

Clinical: Microcephaly; vestigial cerebellum; cleft palate; micrognathia; hydrocephalus.

Jarmas AL. Microcephaly, microphthalmia, falciform retinal folds and blindness. *Am J Dis Child* 1981; 135:930-933.

Masuda Y. Two cases of ablatio falciforms congenita and two other cases of ocular congenital anomalies, which appeared in a pedigree with consanguineous marriages. *J Clin Ophthalmol Tokyo* 1962; 16:325.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Warburg M. Heterogeneity of congenital retinal non-attachment falciform folds and retinal dysplasia: a guide to genetic counseling. *Hum Hered* 1976; 26:137-148.

Young ID, et al. Microcephaly, microphthalmos and retinal folds: report of a family. *J Med Genet* 1987; 24: 172-184.

Falciform Folds with Obesity, Nontoxic Goiter, Hypogenitalism, and Cryptorchidism 440

General: Etiology unknown; isolated rare cases.

Ocular: Falciform folds; vitreous opacity.

Clinical: Obesity; nontoxic goiter; hypogenitalism; cryptorchidism.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998,

Warburg M. Heterogeneity of congenital retinal non-attachment falciform folds and retinal dysplasia: a guide to genetic counseling. *Hum Hered* 1976; 26:137-148.

Familial Histiocytic Dermatoarthritis Syndrome 441

General: Autosomal dominant; manifestations in childhood or adolescence; progressive.

Ocular: Glaucoma; bilateral uveitis; complicated cataract.

Clinical: Multiple histiocytic cutaneous nodules (face, ears, upper and lower extremities); subcutaneous plaques apparent on palpation and thickened, lichenified skin; arthropathy with symmetrical destructive arthritis mainly of hands and wrists but also observed on feet and elbows; possible hearing loss; cardiac and skeletal muscle failure; severe synovitis with arthritis mutilans.

Campbell DA, Edwards NL. Multicentric reticulohistiocytosis: systemic macrophage disorder, *Baillieres Clin Rheumatol* 1991; 5:301-319.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Sauden Y. Diagnosis of peripheral arthropathies. *Radiol Clin Biol* 1974; 43:283.

Zayid J, Farraj S. Familial histiocytic dermatoarthritis: a new syndrome. *Am J Med* 1973; 54:793.

Familial Hypogonadism Syndrome 442

General: Defect in testosterone biosynthesis.

Ocular: Progressive visual loss to complete blindness beginning shortly after birth; cataract; retinal degeneration.

Clinical: Partial deafness (neural type); obesity; shortness of stature; normal virilization.

Jenner MR, et al. Hormonal changes in puberty. IV. Plasma estradiol, LH and FSH in prepubertal children, pubertal females, and in precocious puberty, premature thelarche, hypogonadism, and in a child with a feminizing ovarian tumor. *J Clin Endocrinol Metab* 1972; 34:521.

Lubinsky MS. Cataracts and testicular failure in 3 brothers. *Am J Med Genet* 1983; 16:149-152.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Weinstein RL, et al. Familial syndrome of primary testicular insufficiency with normal virilization, blindness, deafness and metabolic abnormalities. *N Engl J Med* 1969; 281:969.

Familial Juvenile Nephronophthisis (Medullary Cystic Disease) 443

General: Number of closely related renal disorders are associated with tapetoretinal degeneration; cause unknown.

Ocular: Retinitis pigmentosa; night blindness; progressive constriction of peripheral fields; retinal arterioles narrowed; yellow pigment deposits present throughout retina; macular degeneration.

Clinical: Renal disorders; polydipsia; polyuria.

Cohen AH, Hoyer JR. Nephrophthisis: a primary tubular basement membrane defect. *Lab Invest J* 1986; 564-572.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Godel V, et al. Congenital Leber amaurosis, keratoconus, and mental retardation in familial juvenile nephronophthisis. *J Pediatr Ophthalmol Strabismus* 1978; 15:89-91.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Familial Mediterranean Fever 444

General: Recessive inherited polyserositis; progressive, fatal complications are renal failure and amyloidosis.

Ocular: Episcleritis; uveitis; colloid bodies; optic neuritis.

Clinical: Peritonitis; pleuritis; arthritis; fever; skin rash; renal failure; amyloidosis; recurrent attacks of fever and polyserositis of unknown origin.

Lossos A, et al. Optic neuritis associated with familial Mediterranean fever. *J Clin Neuro-Ophthalmol* 1993; 13: 141-143.

Scharf J, et al. Episcleritis associated with familial Mediterranean fever. *Am J Ophthalmol* 1985; 100:337-338.

Yazici H, Pazarli H. Eye involvement in a patient with familial Mediterranean fever. *J Rheumatol* 1982; 9:4.

Fanconi Syndrome (Toni-Fanconi Syndrome; Amino Diabetes; Hypochloremic-Glycosuric Osteonephropathy Syndrome; De Toni-Fanconi Syndrome) 445

General: Autosomal recessive inheritance; hematologic manifestations mainly in young patients; in adults the syndrome resembles milkman syndrome with disorder of calcium and phosphorus metabolism; chronic organic acidosis in Fanconi syndrome due to an inborn error of protein metabolism.

Ocular: Massive retinal hemorrhage may be present secondary to blood dyscrasia; bilateral anterior uveitis.

Clinical: Ecchymoses and mucous membrane hemorrhages; skin hyperpigmentation; osteomalacia; pseudo fractures; deformities of radius and absence of thumbs; hypophosphatemia.

Boniuk M, Hill LL. Ocular manifestations of the Toni-Fanconi syndrome with cystine storage disease. *South Med J* 1966; 59:33.

Fanconi G. Die Nicht Diabetischen Glykosurien und Hyperglykamien des Alteren Kindes. *Jb Kinderheilkd* 1931; 133: 257.

Igarashi T, et al. Acute tubulointerstitial nephritis with uveitis syndrome presenting as multiple tubular dysfunction including Fanconi's syndrome. *Pediatr Nephrol* 1992; 6:547-549.

Fanconi-Turler Syndrome (Familial Ataxic Diplegia; Ataxic Diplegia) 446

General: Aberration of the third cranial nerve (supranuclear type); ataxic diplegia is cerebellar ataxia with spastic pareses mainly of lower extremities; affects both sexes; onset at birth.

Ocular: Nystagmus; uncoordinated eye movements; dysmetria.

Clinical: Cerebellar ataxia; mental deficiency; spastic pareses.

Fanconi G, Turler U. Kongenitale Kleinhimatrophie mit Supranuklearen Storungen der Motilitat der Augenrnuskeln. *Helv Paediatr Acta* 1951; 6:475.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Pfeiffer RA, et al. Nosology of congenital non-progressive cerebellar ataxia. Report on six cases in three families. *Neuropaediatrie* 1974; 5:91.

Farber Syndrome (Farber Lipogranulomatosis; Disseminated Lipogranulomatosis) 447

General: Autosomal recessive inheritance; onset shortly after birth; rare; ceramidase deficiency.

Ocular: Parafoveal edema with mild cherry-red spot; grayness of the macula; diffuse fine pigmentary changes in the fundus.

Clinical: Progressive hoarseness; swelling of extremities; nodular and granulomatous infiltrations of periarticular and subcutaneous tissue; mild lymphadenopathy; fever attacks; dysphonia and dyspnea; irritability; ceramidase deficiency associated with storage of ceramide in body tissues.

Cogan DG, et al. Retinopathy in a case of Farber's lipogranulomatosis. *Arch Ophthalmol* 1966; 75:752.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Farber S, et al. Lipogranulomatosis: a new lipo-glycoprotein storage disease. *Mt Sinai Hosp Bull* 1957; 24:816.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology. vol. V.* Philadelphia: WB Saunders, 1994:2778.

Fat Adherence Syndrome 448

General: Presence of a scar or adhesion that originates in extraconal fat and extends through Tenon capsule to attach to the muscle insertion or sclera; seen following retinal surgery.

Ocular: Persistent acquired restrictive strabismus after retinal surgery; diplopia.

Clinical: None.

Mets MB, et al. Ocular deviation after retinal detachment surgery. *Am J Ophthalmol* 1985; 99:667-672.

Parks MM. Causes of the adhesive syndrome. *Symposium on Strabismus; Transactions of the New Orleans Academy of Ophthalmology*. St. Louis: CV Mosby, 1978:269-279.

Wright KW. The fat adherence syndrome and strabismus after retina surgery. *Ophthalmology* 1986; 93:411-415.

Favre-Racouchot Syndrome (Nodular Elastoidosis) 449

General: Reaction to sun; permanent, slowly progressing condition; occurs in people chronically exposed to sun; usually apparent in fourth or fifth decade.

Ocular: Yellowish thickening of skin, with comedones and follicular cysts in and around orbit.

Clinical: Elastotic degeneration of skin; raised yellow patches and numerous comedones; orifices enlarged; citrine skin; cutis rhomboidalis nuchae; elastoma Dubreuilh.

Favre M, Racouchot JL. Elasteidose Cutanee Nodulaire a Kystes et a Comedons. *Ann Dermatol Symp* 1951; 78: 681-702.

Ritchie EB, Williams HN. Degenerative collagenous plaques of the hands. *Arch Dermatol* 1966; 93:202-203.

Feer Syndrome (Swift-Feer Syndrome; Infantile Acrodynia; Acrodynia; Pink Disease) 450

General: Etiology unknown, possibly allergic reaction to mercury or infection; onset in early childhood; both sexes equally affected.

Ocular: Proptosis; lacrimation; pronounced photophobia; severe conjunctival itching; conjunctival injection with occasional marked signs of inflammation; severe keratitis; mild optic neuritis; mydriasis.

Clinical: Restlessness; irritability; continuous profuse sweating; muscle hypotony; tachycardia; exanthema of palms and soles with exfoliation of large skin flaps; stomatitis; sleeplessness; cyanosis of fingers, toes, and nose; loss of teeth; rectal prolapse; muscle hypotonia; hypertension; hypertrichosis; gangrene of fingers.

Duke-Elder S, ed. *Systems of Ophthalmology, vol. VIII, part* 2. St. Louis: CV Mosby, 1976.

Feer E. Eine Eigenartige Neurose des Vegatativen Systems Beim Kleinkinde, *Ergebn Inn Med Kinderh* 1923; 24:100.

Felty Syndrome (Chauffard-Still Syndrome; Primary Splenic Neutropenia with Arthritis; Rheumatoid Arthritis with Hypersplenism; Still-Chauffard Syndrome; Uveitis-Rheumatoid Arthritis Syndrome) 451

General: Etiology not fully understood, possibly infection or allergy; onset in middle-aged patients or children; prognosis poor; collagen disorder; occasionally can occur without articular disease.

Ocular: Decreased tear formation; scleromalacia perforans; keratoconjunctivitis; chronic anterior uveitis; scleritis; vitreous opacities; macular edema; choroidal inflammation; papillitis; keratitic precipitates; band-shaped keratopathy.

Clinical: Rheumatoid arthritis; splenomegaly; leukopenia; anemia (mild); oral lesion with ulcers and atrophy.

Crosby WH. What to treat in Felty's syndrome. *JAMA* 1973; 225:1114.

Davis M. Endogenous uveitis in children: associated band-shaped keratopathy and rheumatoid arthritis. *Arch Ophthalmol* 1953; 50:443.

Felty AR. Chronic arthritis in the adult associated with splenomegaly and leukopenia. *Bull Johns Hopkins Hosp* 1924; 35:16.

Pau H. *Differential diagnosis of eye disease*. New York: Thieme, 1987.

Reddy CV, Foster CS. Adult rheumatoid arthritis. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:2889.

Fetal Alcohol Syndrome 452

General: Dysgenesis in children born to alcoholic mothers; both sexes affected; onset from birth.

Ocular: Antimongoloid slant of lid fissures; lateral displacement of inner canthi; ptosis; epicanthi; strabismus; myopia; optic nerve hypoplasia; diffuse corneal clouding; iridocorneal abnormalities with central corneal edema; lens opacification; motility disorders.

Clinical: Growth retardation; delayed development (physical and intellectual); maxillary hypoplasia; micrognathia; large, low-set ears; abnormal motor function; irritability; microcephaly; cerebral nervous system dysfunctions; abnormal philtrum; flattened nasal bridge; cardiovascular defects; thin upper lip.

Chan T, et al. Ocular manifestations in fetal alcohol syndrome. *Br J Ophthalmol* 1991; 75:524-526.

Cook CS, et al. Fetal alcohol syndrome. *Arch Ophthalmol* 1987; 105:1576.

Edward DP, et al. Diffuse corneal clouding in siblings with fetal alcohol syndrome. *Am J Ophthalmol* 1993; 115: 484-493.

Tenbrinck MS, Buchin SY. Fetal alcohol syndrome. *JAMA* 1975; 232:1144.

Ulleland CN. The offspring of alcoholic mothers. *Ann N Y Acad Sci* 1972; 197:167.

Fetal Anticonvulsant Syndrome 453

General: Congenital after maternal ingestion fo sodium valproate, carbamazepine and phenytoin.

Ocular: Myopia; astigmatism; strabismus; anisometropia; optic nerve hypoplasia

Clinical: Craniofacial dysmorphisms vary with type of abnormality

Glover SJ, Quinn AG, Barter P, et al. Ophthalmic findings in fetal anticonvulsant syndrome. *Ophthalmology* 2002; 109:942-947

Hoyt CS, Billson FA: Maternal anticonvulsants and optic nerve hypoplasia. *Br J Ophthalmol* 1978; 62: 3-6.

Waller PH, Genstler DE, George CC, et al. Multiple systemic and periocular malformations associated with the fetal hydantoin syndrome. *Ann Ophthalmol* 1978; 10: 1568-72

Fetal Hydantoin Syndrome 454

General: Syndrome due to an epoxide hydrolase 1, microsomal, arene oxide detoxification defect.

Ocular: Nystagmus.

Clinical: *In vitro* testing for the defect correlates most highly with congenital heart disease, cleft lip/palate, microcephaly, and major genitourinary, eye, and limb defects; hypersensitivity to phenytoin has occurred.

Buehler BA, et al. Prenatal prediction of risk of the fetal hydantoin syndrome. *N Engl J Med* 1990; 322:1567-1571.

Gennis MA, et al. Familial occurrence of hypersensitivity to phenytoin. *Am J Med* 1991; 91:631-634.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Fibrinoid Syndrome 455

General: From 2 to 14 days postvitrectomy, white-gray criss-cross layers of fibrin appear on the surface of the retina and immediately behind the plane of the iris; occurs only in patients with diabetes mellitus and usually in those requiring insulin; seen more frequently in people who have been diabetic for 15 years or more.

Ocular: Fibrin material interlaced on the surface of the retina and behind the iris; retinal detachment; neovascular glaucoma; rubeosis irides.

Clinical: Diabetes mellitus.

Ho T, et al. Vitrectomy in the management of diabetic eye disease. *Surv Ophthalmol* 1992; 37:190-202.

Schepens CL. Clinical and research aspects of subtotal open-sky vitrectomy: 439VII Edward Jackson Memorial Lecture. *Am J Ophthalmol* 1981; 92:143-171.

Sebestzen JG. Fibrinoid syndrome: a severe complication of vitrectomy surgery in diabetes. *Ann Ophthalmol* 1982; 14:853-856.

Fibrosarcoma 456

General: Malignant tumor of fibrous connective tissue; most commonly seen in persons 30 to 70 years old; frequent metastases to lung; true fibrosarcoma has a tendency to occur in children (better prognosis); most fibrosarcomas now would be classified as malignant fibrous histiocytomas.

Ocular: Paralysis of extraocular muscles; proptosis; orbital edema; erosion of orbital bony walls; increased intraorbital pressure; metastases to choroid/orbit.

Clinical: Tumors of mesenchymal soft tissues of the extremities, especially in the knee region; progressive pain; edema; tumors of the sinuses; tumors of the lungs.

Henderson JW. *Orbital tumors,* 2nd ed. New York: Brian C. Decker, 1980:183-188.

Jakobiec FA, Tannenbaum M. The ultrastructure of orbital fibrosarcoma. *Am J Ophthalmol* 1974; 77:899-9l7.

Roy FH. Fibrosarcoma. In: Fraunfelder FT, Roy FH, eds. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000:337.

Winer JM, et al. Juvenile fibrosarcoma of the orbit and eyelid: a study of 5 cases. *Arch Ophthalmol* 1983; 101:253.

Fish Odor Syndrome (Trimethylaminuria) 457

General: Metabolic syndrome characterized by a strong body odor of rotting fish; trimethylamine levels elevated; it appears that the enzyme flavin-containing monooxygenase is defective in this disorder; possibly autosomal recessive pattern of inheritance.

Ocular: Hypertelorism, cortical blindness.

Clinical: Hydrocephalus; mental retardation; unusual facies; short stature; skeletal abnormalities; cryptorchidism; hyperextensible skin.

Chen H, Aiello F. Trimethylaminuria in a girl with Prader-Willi syndrome and del(15)(q11q13). *Am J Med Genet* 1993; 45:335-339.

Humbert JR, et al. The fish odor syndrome. *Lancet* 1970; 2:770-771.

Shelley ED, Shelley WB. The fish odor syndrome. *JAMA* 1984; 251:253-256.

Fisher Syndrome (Ophthalmoplegia Ataxia Areflexia Syndrome; Miller-Fisher Syndrome) 458

General: Acute idiopathic polyneuritis; prognosis good; complete recovery over several weeks (variant of Guillain-Barrée Syndrome; see Guillain-Barré Syndrome).

Ocular: Moderate ptosis; complete external and almost complete internal ophthalmoplegia; diplopia; sluggish pupil reaction to light; may present without total ophthalmoplegia.

Clinical: Dizziness; severe ataxia; loss of tendon reflexes; chest pains; difficulties in chewing; diminished or absent sense of vibration; upper respiratory tract infection preceding this syndrome.

Fisher M. An unusual variant of acute idiopathic polyneuritis (syndrome of ophthalmoplegia, ataxia and areflexia). *N Engl J Med* 1956; 255:57.

Igarashi Y, et al. Fisher's syndrome without total ophthalmoplegia. *Ophthalmologica* 1992; 205:163-167.

Swick HM. Pseudo internuclear ophthalmoplegia in acute idiopathic polyneuritis (Fisher syndrome). *Am J Ophthalmol* 1974:77:725.

Fish-Eye Disease (Corneal Opacities-Dyslipoproteinemia) 459

General: Etiology unknown; rare; described in Swedish family; currently considered a unique dyslipoproteinemia.

Ocular: Visual impairment; marked corneal opacities.

Clinical: Very-low-density triglycerides and cholesterol raised.

Carlson LA, Philipson B. Fish-eye disease. A new familial condition with massive corneal opacities and dyslipoproteinemia. *Lancet* 1979; 2:922-924.

Koster H, et al. A fish-eye disease-like familial condition with massive corneal clouding and dyslipoproteinemia. Report of *Clinical,* histologic, electron microscopic, and biochemical features. *Cornea* 1992; 11:452-464.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Fleck Retina of Kandori Syndrome (Kandori Syndrome) 460

General: Possibly hereditary; onset young age; focal disturbance of the retinal pigment epithelium (RPE); affects both sexes; toxic causes also considered.

Ocular: Relatively large, irregular, yellowish flecks, sharply border-lined without pigmentation underneath retinal vessels and usually in the midperiphery; poor dark adaptation; normal photopic electroretinographic response; delay in generation of the scotopic response.

Clinical: None.

Bullock JD, Albert DM. Flecked retina. *Arch Ophthalmol* 1975; 93:26.

Carr RE. Abnormalities of cone and rod function. In: Ryan SJ, ed. *Retina, vol. II,* 2nd ed. St. Louis: Mosby, 1994: 512-513.

Kandori F. Very rare cases of congenital non-progressive nightblindness with fleck retina. *Jpn J Ophthalmol* 1959; 13:394.

Floppy Eyelid Syndrome 461

General: Origin unknown; more common in males; overweight; X-chromosome-linked inheritance pattern or possible hormonal influence; has been postulated that the degenerative changes in the tarsus may result from the combination of local pressure-induced lid ischemia and systemic hypoventilation.

Ocular: Easily everted, floppy upper eyelid and papillary conjunctivitis of the upper palpebral conjunctiva; upper eyelid everts during sleep, resulting in irritation, papillary conjunctivitis, and conjunctival keratinization; most distinct feature is rubbery, malleable upper tarsus; keratoconus; punctate keratopathy; blepharoptosis; lash ptosis.

Clinical: Obesity; sleep apnea.

Culbertson WW, Ostler HB. The floppy eyelid syndrome. *Am J Ophthalmol* 1981; 92:568-575.

Culbertson WW, Tsenc SC. Corneal disorders in floppy eyelid syndrome. *Cornea* 1994; 13:33-42.

Donnenfeld ED, et al. Keratoconus associated with floppy eyelid syndrome. *Ophthalmology* 1991; 98:1674-1678.

Goldberg R, et al. Floppy eyelid syndrome. *Am J Ophthalmol* 1986; 102:376-381.

Katz J, Kaufman HE. Corneal exposure during sleep (nocturnal lagophthalmos). *Arch Ophthalmol* 1977; 95:449.

Moore MB, et al. Floppy eyelid syndrome. *Ophthalmology* 1986; 93:184-188.

Netland PA, et al. Histopathologic features of the floppy eyelid syndrome. Involvement of tarsal elastin. *Ophthalmology* 1994; 101:174-181.

Floppy Iris Syndrome 462

General: Flomax (tamsulosin) is causative agent during cataract. Flomax is commonly prescribed for benign prostate hypertrophy.

Ocular: Floppy iris

Clinical: Benign prostate hypertrophy; noted during cataract surgery

Chang DF, Campbell JR: Intraoperative floppy iris syndrome associated with tamsulosin. *J Cataract Refract Surg* 2005; 31: 664-73

Gurbaxani A, Packard R: Intracameral phenylephrine to prevent floppy iris syndrome during cataract surgery in patients on tamsulosin

Schrehardt US, Stojkovic M, Hofmann-Rummelt C, et al. the pathogenesis of floppy eyelid syndrome. *Ophthalmology* 2005; 112: 694-704

Flynn-Aird Syndrome 463

General: May be basic hereditary enzyme deficiency, probably autosomal dominant; no sex predilection apparent.

Ocular: Severe myopia; bilateral cataracts; retinitis pigmentosa; total blindness; onset of visual difficulties in first or second decade of life.

Clinical: Hearing loss; joint stiffness; muscular wasting; kyphoscoliosis.

Flynn P, Aird RB. A neuroectodermal syndrome of dominant inheritance. *J Neurol Sci* 1965; 2:161-182.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Regenbogen LS, Coscas GJ. *Oculo-auditory syndromes.* New York: Masson, 1985:99-103.

Foix Syndrome (Cavernous Sinus Syndrome; Hypophyseal-Sphenoidal Syndrome; Cavernous Sinus Neuralgia Syndrome; Godtfredsen Syndrome; Cavernous Sinus-Nasopharyngeal Tumor Syndrome; Cavernous Sinus Thrombosis) 464

General: Causes include tumor of lateral sinus wall or sphenoid bone, intracranial aneurysm, cavernous and lateral sinus thrombosis, or lesions; multiple myeloma; may result from infarctions or cancer or be idiopathic.

Ocular: Proptosis; severe ocular and periorbital pain; lid edema; paresis or paralysis of cranial nerves III, IV,V, and VI ; corneal anesthesia; optic atrophy.

Clinical: Postauricular edema; trigeminal neuralgia; deviation of the tongue toward paralyzed side; patients usually have prominent manifestations of sepsis and paranasal sinus; local skin infections are the most common cause.

Foix C. Syndrome de la Paroi Externe du Sinus Caverneux. *Rev Neurol* 1922; 37/38:827.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Newman NJ. Third-, fourth, and sixth-nerve lesions and the cavernous sinus. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994:2444.

Rucker CW. The causes of paralysis of the third, fourth and sixth cranial nerves. *Am J Ophthalmol* 1966; 61:1293.

Tveteras K, et al. Septic cavernous and lateral sinus thrombosis. Modem diagnostic and therapeutic principles. *J Laryngol Otol* 1988; 102:877.

Folling Syndrome (Phenylketonuria; Phenylpyruvic Oligophrenia; Ikiotia Phenylketonuria Syndrome) 465

General: Rare; autosomal recessive; phenylalanine cannot be converted to tyrosine; poor prognosis without early diet therapy; both sexes affected.

Ocular: Blue sclera; severe photophobia; corneal opacities; cataracts (controversial); partial ocular albinism; macular atrophy.

Clinical: Phenylketonuria; oligophrenia; partial albinism; muscle hypertonicity; hyper-reflexia of tendons; epilepsy; microcephaly; mousy odor of habitus; fair skin.

Dornguth S, et al. Golgi-Kopsch silver study of the brain of a patient with untreated phenylketonuria, seizures, and cortical blindness. *Am J Med Genet* 1992; 44:443-448.

Foiling A. Uber Ausscheidung von Phenylbrenztraubensaure in den Ham als Stoffwechselanomalie in Verbindung mit Imbezillitat. *Ztschr Physiol Chem* 1934; 227:169.

Pitt DB, O'Day J. Phenylketonuria does not cause cataracts. *Eur J Pediatr* 1991; 150:661-664.

Zeligman I, et al. Macular anetoderma, secondary to lichen sclerosus et atrophicus, acanthosis nigricans and apocrine hydrocystoma in a man with phenylketonuria. *Birth Defects* 1971; 7:256-258.

Foot-in-the-Wound Syndrome 466

General: Occurs when the anterior chamber intraocular lens haptic is within the wound.

Ocular: Haptic in the wound.

Clinical: None.

Hessburg PC. Complications noted in PMMA IOL haptics: fracture, pupillary capture named problems. *Ophthal Times* August 21, 1983.

Foramen Lacerum Syndrome (Aneurysm of Internal Carotid Artery Syndrome) 467

General: Most commonly caused by congenital aneurysm involving the intradural portion of the carotid artery.

Ocular: Periorbital pain; ptosis; oculomotor paralysis with ptosis, diplopia, and internal ophthalmoplegia; cranial nerves IV and VI may be involved; homonymous hemianopia (occasionally); loss of pupillary reflexes for light and accommodation; papilledema; optic atrophy.

Clinical: Meningism; mental disturbances; unilateral frontal or orbital headache; migraine attacks.

Dailey EJ, et al. Evaluation of ocular signs and symptoms in cerebral aneurysms. *Arch Ophthalmol* 1964; 71:463.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Misra M, et al. Giant aneurysm of internal carotid artery presenting features of retrobulbar neuritis. *Ind J Ophthalmol* 1992; 39:28-29.

Forsius-Eriksson Syndrome (Aland Disease) 468

General: Associated with the natives of the Aland Islands; sex-linked inheritance; consanguinity versus mutant gene; affects males only; it has been considered a variety of incomplete congenital stationary night blindness.

Ocular: Microphthalmos; irregular latent nystagmus; myopia; astigmatism; dyschromatopsia; tapetoretinal degeneration; primary foveal hypoplasia or dysplasia; nystagmus.

Clinical: Prematurity; impaired hearing; mental retardation; epilepsy.

Forsius H, Eriksson AW, Ein Neues Augensyndrom mit X-chromosomaler Transmission. Eine Sippe mit Fundusal-binismus, Foveahypoplasie, Nystagmus, Myopic, Astigmatismus und Dyschromatopsie. *Klin Monatsbl Augenheilkd* 1964; 144:447.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*. 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Vliet AGM, et al. Nystagmographical studies in Aland eye disease. *Acta Ophthalmol* 1973; 51:782.

Weleber RG, et al. Aland Island disease (Forsius-Eriksson syndrome) associated with contiguous gene syndrome at Xp21: similarity to incomplete congenital stationary night blindness. *Arch Ophthalmol* 1989; 107:1170.

Foster Kennedy Syndrome (Basal-Frontal Syndrome; Gowers-Paton-Kennedy Syndrome) 469

General: Caused by tumor in base of frontal lobe or sphenoidal meningioma.

Ocular: Central scotoma may be present on side of optic atrophy; enlarged blind spot and peripheral contraction of field (opposite eye); homolateral descending optic atrophy due to compression of the ipsilateral optic nerve at the optic foramen; contralateral papilledema due to increased intracranial pressure; ipsilateral proptosis.

Clinical: Anosmia; headache; dizziness; vomiting; memory loss; psychic changes; also may be caused by an olfactory groove tumor (usually a meningioma) or pituitary adenoma.

Banerjee T, Meagher JN. Foster Kennedy syndrome, aqueductal stenosis and empty sella. *Am Surg* 1974; 40:552.

Kennedy F. Retrobulbar neuritis as an exact diagnostic sign of certain tumors and abscesses in frontal lobes. *Am J Med Sci* 1911; 142:355.

Ruben S, et al. Pituitary adenoma presenting as the Foster-Kennedy syndrome. *Br J Ophthalmol* 1992; 76:117-119.

Yildizhan A. A case of Foster Kennedy syndrome without frontal lobe or anterior cranial fossa involvement. *Neurosurg Rev* 1992; 15:139-142.

4 a Syndrome (Adrenocortical insufficiency associated with achalasia, alacrima, autonomic and other neurological abnormalities) 470

General: Associated with autonomic and other neurologic abnormalities

Ocular: Alacrima

Clinical: Adreno-coritcal infufficiency; achalsia

Davidoff E, Friedman AH. Congenital alacrima. *Surv Ophthalmol* 1997; 22: 113-119

Gazarian M, Cowell CT, Bonney M, et al. The 4a syndrome: adrenocortal insufficiency associated with achalasia, alacrima, autonomic and other neurological abnormalities. *Eur J Pediat* 1995; 154: 18-23.

4q- Syndrome (4q Deletion Syndrome) 471

General: Chromosome 4q deletion syndrome.

Ocular: Hypertelorism, epicanthal folds.

Clinical: Depressed nasal bridge; short nasal septum with upturned nose, cleft lip and palate; micrognathia; low-set malformed ears; short neck; distally placed nipples; sacral dimple; hypospadias; dysplastic nails; overriding toes; simian creases; hypoplasia of gall bladder; cardiac defects; mental retardation.

Robertson SP, O'Day K, Banker A. The 4q- syndrome: delineating of the minimal critical region to within band 4q31. *Clin Genet* 1998; 53:70-73.

Townes PL, White M, DiMarzo SV. 4q- syndrome. *Am J Dis Child* 1979; 133:383-385.

Yu CW, et al. Terminal deletion of the long arm of chromosome 4. Report of a case of 46,XY,del(4)(q31) and review of 4q- syndrome. *Ann Genet* 1981; 24:158-161.

Foveal Hypoplasia and Presenile Cataract Syndrome (O'Donnell-Pappas Syndrome) 472

General: Autosomal dominant.

Ocular: Foveal hypoplasia; nystagmus; presenile cataract; peripheral corneal pannus.

Clinical: None.

O'Donnell FE, Pappas HR. Autosomal dominant foveal hypoplasia and presenile cataracts: a new syndrome. *Arch Ophthalmol* 1982; 100:279-281.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Foville Syndrome (Foville Peduncular Syndrome) 473

General: Pontine area tumor, hemorrhage, tuberculoma, multiple sclerosis, or unilateral obstruction of paramedian branches may cause clinical manifestations.

Ocular: Paralysis of cranial nerve VI; paralysis of conjugate movement to the side of the lesion; abduction or horizontal gaze deficit.

Clinical: Peripheral facial palsy; contralateral hemiplegia; headache; ipsilateral: facial weakness, loss of taste, facial analgesia, Homer syndrome, and deafness.

Bedi HK. Clinical tuberculoma of pons presenting as Foville's syndrome. *J Ind Med Assoc* 1973; 61:184.

Foville ALF. Note sur une Paralysie Peu Connue des Certains Muscles de l'Oeil, et Sa Liaison avec Quelques Points de l'Anatomie et la Physiologic de la Protuberance Annulaire. *Bull Soc Anat (Paris)* 1858; 33:393.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Newman NJ. Third, fourth, and sixth nerve lesions and the cavernous sinus. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994:2458.

Fragile X Syndrome 474

General: X-linked recessive; primarily affects males.

Ocular: Strabismus; nystagmus; high myopia; adult-onset glaucoma; blepharospasm; congenital optic atrophy; hyperopia; astigmatism; cataract; ptosis; corneal dystrophy.

Clinical: Mental retardation; dysmorphism; epilepsy; macroorchidism.

Harley RD, ed. *Pediatric Ophthalmology,* 4th ed. Philadelphia: WB Saunders, 1988.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Reiss AL, et al. Neuroanatomy of fragile X syndrome: the posterior fossa. *Ann Neurol* l991; 29:26-32.

Storm RL, et al. Ophthalmologic findings in the fragile X syndrome. *Arch Ophthalmol* 1987; 105:1099-1102.

Franceschetti Disease (Fundus Flavimaculatus) 475

General: Affects both sexes; onset between ages 10 and 25 years; autosomal recessive; genetic linkage analysis has assigned the disease locus to chromosome 1p21-p13.

Ocular: Irregular yellowish deposit in and around the macula lutea forming a garland; impaired central vision with intact peripheral retinal function; bilateral retinal dystrophy; progressive subretinal fibrosis; chorioretinal punched-out spots in the posterior pole and midperiphery of the retina.

Clinical: None.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* Philadelphia: JB Lippincott, 1981.

Parodi MB. Progressive subretinal fibrosis in fundus flavimaculatus. *Acta Ophthalmol* 1994; 72:260-264.

Franceschetti Syndrome (Franceschetti-Zwahlen-K1ein Syndrome; Treacher Collins Syndrome; Mandibulofacial Dysostosis; Mandibulofacial Syndrome; Eyelid-Malar-Mandible Syndrome; Oculovertebral Syndrome; Berry Syndrome; Franceschetti-Zwahlen Syndrome; Zwahlen Syndrome; Bilateral Facial Agenesis; Berry-Franceschetti-K1ein Syndrome; Franceschetti-K1ein Syndrome; Franceschetti Syndrome (II); Treacher Collins-Franceschetti Syndrome; Weyers-Thier Syndrome) 476

General: Irregular dominant inheritance; Weyers-Thier syndrome has similar features, except it is a unilateral variant; prevalent in Caucasians.

Ocular: Microphthalmia; oblique position of eyes with lateral downward slope of palpebral fissures; temporal lower lid coloboma; lack of cilia on middle third of lower lid; iris coloboma; underdeveloped orbicularis oculi muscle; cataract; optic disk hypoplasia.

Clinical: Fishlike face with sunken cheek bones, receding chin, and large, wide mouth; absent or malformed external ears with auricular appendages; high palate and possible harelip; hypoplastic zygomatic arch with absence of normal malar eminences; prolonged hairline on the cheek; deafness; micrognathia; glossoptosis; cleft palate.

Berry GA. Note of congenital defect (coloboma) of lower lid. *Lond Ophthalmol Hosp Rep* 1889; 12:255-277.

Collins ET. Cases with symmetrical congenital notches in the outer part of each lid and defective development of the malar bones. *Trans Ophthalmol Soc UK* 1900; 20:190.

Franceschetti A, Klein D. Mandibulo-facial dysostosis: new hereditary syndrome. *Acta Ophthalmol* 1949; 27:143.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Jabs EW, et al. Chromosomal deletion 4p15.32-p14 in a Treacher Collins syndrome patient: exclusion of the disease locus from and mapping of anonymous DNA sequences to this region. *Genomics* 1991; 11:188-192.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Rogers BO. The surgical treatment of mandibulofacial dysostosis (Berry syndrome; Treacher Collins syndrome; Franceschetti-Zwahlen-Klein syndrome). *Clin Plast Surg 1976; 3:653-666.*

Franceschetti Syndrome (I) 477

General: Inborn disease occurring at birth; etiology unknown.

Ocular: Deep punctate dystrophy of cornea with ichthyosis dystrophia (punctiformis profunda corneae with congenital ichthyosis).

Clinical: Dry, scaly skin; follicular hyperkeratotic lesions; atopic dermatitis; pruritus.

Jablonski S. *Eponymic syndrome and diseases.* Philadelphia: WB Saunders, 1969.

Stewart WD, et al. *Dermatology*, 4th ed. St. Louis: CV Mosby, 1978.

Franceschetti-Thier Syndrome 478

General: Autosomal recessive inheritance.

Ocular: Corneal dystrophy.

Clinical: Multiple lipomas; mental retardation.

Franceschetti A, et al. Hornhautdystrophien bei Genodermatosen unter besonderen Berucksichtigung der Palmoplantarkeratosen. *Graefes Arch Clin Exp Ophthalmol* 1961; 162:610-670.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981:294.

Francois (1) Dystrophy (Francois-Neetens Syndrome; Central Cloudy Dystrophy; Cloudy Central Corneal Dystrophy) 479

General: Autosomal dominant; etiology unknown; not progressive; isolated keratocytes contain elevated amounts of glycosaminoglycans and lipids.

Ocular: Bilateral dystrophy of central third of cornea; snowflake patches covering the pupil; lesions show no definite structure or limits; more dense near Descemet membrane and becoming less toward the anterior surface toward the periphery; associated with central cloudy dystrophy; keratoconus; limbal dermoid; pseudoxanthoma elasticum; lenticular opacities; reduced corneal sensation.

Francois J. Une Novelle Dystrophie Heredo-familiale de la Cornee. *J Genet Hum* 1956; 5:189-196.

Jablonsk S. *Eponymic syndromes and diseases.* Philadelphia: WB Saunders, 1969:110.

Merin S. Corneal dystrophies affecting the stroma. Cloudy central cornea dystrophy. In: Merin S, ed. *Inherited eye diseases.* New York: Marcel Dekker, 1991:22-41.

Pavan-Langston D. Cornea and external diseases. Central cloudy and parenchymatous dystrophy. In: Pavan-Langston D, ed. *Manual of ocular diagnosis and therapy*, 4th ed. Boston: Little, Brown and Company, 1995:108.

Smolin G. Corneal dystrophies and degenerations. In: Smolin G, Thoft RA, eds. *The Cornea*. Boston: Little, Brown and Company, 1994:516.

Francois (2) Dystrophy (Francois-Evens Syndrome; Speckled Corneal Dystrophy) 480

General: Etiology unknown; congenital; nonprogressive; autosomal dominant, but sporadic cases have been reported.

Ocular: Corneal dystrophy characterized by minute punctate opacities found in all layers of the cornea; varies in size, form, and degree of opacity but is identical in both eyes; anterior limiting membrane is always intact.

Bron AJ. The corneal dystrophies. *Curr Opin Ophthalmol* 1990; 1:333.

Francois J. *Heredity in Ophthalmology.* St. Louis: CV Mosby, 1961.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Francois Syndrome (2) (Dystrophia Dermachondrocornealis Familiaris) 481

General: Autosomal recessive.

Ocular: Central superficial corneal dystrophy with subepithelial opacities.

Clinical: Distal osteochondral dystrophy of the extremities; cutaneous xanthomas.

Caputo R, et al. Dermatochondrocorneal dystrophy (Francois' syndrome): report of a case. *Arch Dermatol* 1988; 124: 424-428.

Francois J. Dystrophic Dermo-Chondro-Corneenne Familiale. *Ann Ocul (Paris)* 1949; 182:409-442.

Jablonski S. *Eponymic syndromes and diseases.* Philadelphia: WB Saunders, 1969.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Frankl-Hochwart Syndrome (Pineal-Neurologic-Ophthalmic Syndrome) 482

General: Pineal tumor, usually in early adulthood; poor prognosis.

Ocular: Limitation of upward gaze; concentric field constriction; papilledema; lack of pupillary reaction; nystagmus.

Clinical: Bilateral deafness; ataxia; weakness; headache; vomiting; polydipsia; polyphagia; convulsions; facial paralysis; tremor; Romberg sign; hypertonia; tendon hyperreflexia; Babinski sign.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB

Lippincott, 1981.

Von Frankl-Hochwart L. Uber Diagnose der Zirbeldrusen-tumoren. *Dtsch Zr Nervenheilkd* 1910; 37:455; 1910; 38:309.

Freeman-Sheldon Syndrome (Cranio-Carpo-Tarsal Dysplasia; Whistling Face Syndrome) 483

General: Rare; autosomal dominant and recessive inheritance as well as sporadic cases (genetic heterogeneity).

Ocular: Eyes deeply sunken (enophthalmos); hypertelorism; blepharophimosis; ptosis; antimongoloid slanting of lid fissures; esotropia.

Clinical: Small nose with narrow nostrils and long philtrum; alae nasi often bent, simulating colobomas; nasolabial folds present only near the nose; microstomia; high-arched palate and small mandible; flexion contractures of fingers; excessive bulging of central part of cheeks when whistling.

Freeman EA, Sheldon JH. Cranio-carpotarsal dystrophy: an undescribed congenital malformation. *Arch Dis Child* 1938; 13:277.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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O'Keefe M, et al. Ocular abnormalities in the Freeman-Sheldon syndrome. *Am J Ophthalmol* 1986; 102:346-348.

Wang TR, Lin SJ. Further evidence for genetic heterogeneity of whistling face or Freeman-Sheldon syndrome in a Chinese family. *Am J Genet* 1987; 28:471-475.

Weinstein S, Gorlin RJ. Cranio-carpotarsal dystrophy or the whistling face syndrome. *Am J Dis Child* 1969; 117:427.

Frenkel Syndrome (Ocular Contusion Syndrome; Anterior Segment Traumatic Syndrome) 484

General: Minor blunt trauma to the anterior segment of the globe.

Ocular: Sluggish pupil reaction; traumatic mydriasis; iris dialysis; heavy pigment deposits on the vitreous surface; subluxation of the lens; transient posterior cortical lens opacities; permanent anterior or posterior capsular opacities; coronary opacities; late anterior cortical rosette; late total traumatic cataract; Vossius ring following hyphema; peripheral pigment disturbance resembling atypical retinitis pigmentosa; macular edema; retinal detachment.

Clinical: None.

Frenkel H. Sur la Valeur Medico-Legale du Syndrome Traumatique du Segment Anterieur. *Arch Ophthalmol* 1931; 48:5.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Friedreich Ataxia (Spinocerebellar Ataxia) 485

General: Etiology unknown, either autosomal recessive or dominant; progressive; incapacitating by age 20 years; death from secondary diseases or cardiac failure; prevalent in males.

Ocular: Nystagmus; optic atrophy; there is a form of Friedreich ataxia associated with congenital glaucoma.

Clinical: Kyphoscoliosis; tremor; dysmetria; asynergia; slow ataxic speech; paresthesias; Babinski sign; headache; retarded growth; mental retardation; polyuria; polydipsia; deformity of feet (onset in first year of life); clumsy gait and difficult to turn arms, head, and trunk; deafness.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Combarros O. Association of an ataxia indistinguishable from Friedreich's ataxia and congenital glaucoma in a family: a new syndrome. *J Med Genet* 1988; 25:44-46.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*. 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Fröhlich Syndrome (Dystrophia Adiposogenitalis) 486

General: Caused by chromophobe adenoma of pituitary, Rathke pouch tumors; craniopharyngiomas; suprasellar tumors; encephalitis; trauma; more frequent in Jewish families; manifestations occur in childhood, often during puberty.

Ocular: Bitemporal hemianopsia; impaired scotopic vision; papilledema; optic nerve atrophy (with increased intracranial pressure).

Clinical: Adiposity; genital hypoplasia; in females, menstruation fails to appear or may cease in postpubertal period; in males, voice remains high-pitched, undescended testes, absent facial hair, and feminine pubic line; possible retarded growth; polyuria; polydipsia.

Frohlich A. Ein Fall von Tumor der Hypophysis Cerebri ohne Akromegalie, Wien. *Kim Rdsch* 1901; 15:883,906.

Landau J, Bromberg YM. Impaired scotopic vision in adiposo-genital dystrophy. *Br J Ophthalmol* 1955; 38:155.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology,* 4th ed. Baltimore: Williams & Wilkins, 1987.

Frontometaphyseal Dysplasia (FMD) 487

General: Sex-linked; rare; bony dysplasia.

Ocular: Strabismus; supraorbital deformity; hyperopia; hypertelorism; prominent supraorbital ridges.

Clinical: Agenesis of frontal sinuses; underdevelopment of mandible; metaphysis of tubular bones; deafness; hirsutism; teeth abnormalities; bony overgrowth at base of nose resulting in nasal obstruction and mouth breathing; facial nerve paralysis; normal intelligence.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*. 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Reardon W, et al. Sibs with mental retardation, supraorbital sclerosis, and metaphyseal dysplasia: frontometaphyseal dysplasia, craniometaphyseal dysplasia, or a new syndrome? *J Med Genet* 1991; 28:622-626.

Stem SD, et al. The ocular and cosmetic problems in frontometaphyseal dysplasia. *J Pediatr Ophthalmol* 1972;9: 151-160.

Frontonasal Dysplasia Syndrome (Median Cleft Face Syndrome) 488

General: Congenital disorder without genetic background; condition may present a variety of facial malformations, depending on the stage of embryonic development at which interference occurs.

Ocular: Hypertelorism; anophthalmia or microphthalmia; significant refractive errors; strabismus; nystagmus; eyelid ptosis; optic nerve hypoplasia; optic nerve colobomas; cataract; corneal dermoid; inflammatory retinopathy.

Clinical: Broad nasal root may be associated with median nasal groove and cleft of nose and/or upper lip; cleft of ala nasi (unilateral or bilateral); V-shaped hair prolongation into forehead.

Kinsey JA, Streeten BW: Ocular abnormalities in the medial cleft face syndrome. *Am J Ophthalmol* 1977; 83:261.

Roarty JD, et al. Ocular manifestations of frontonasal dysplasia. *Plast Reconstr Surg* 1994; 93:25-30.

Sedano HO, et al. Frontonasal dysplasia. *J Pediatr* 1970; 76:906.

Weaver D, Bellinger D. Bifid nose associated with midline cleft of the upper lip: case report. *Arch Otolaryngol* 1946; 44:480.

Fuchs (1) Syndrome (Heterochromic Cyclitis Syndrome) 489

General: Etiology unknown; mild infective cyclitis is the most likely cause; etiology remains unclear, although it is likely to be autoimmune; positive epidemiologic association with ocular toxoplasmosis has been investigated.

Ocular: Secondary glaucoma; unilateral hypochromic heterochromia; painless cyclitis with absence of synechiae and little or no ciliary injection; secondary cataract; vitreous opacities; small white discrete keratic precipitates with fine filaments between the precipitates; corneal epithelium may be slightly edematous; peripheral choroiditis occasionally; keratoconus.

Clinical: Occasional dysraphia of the cervical cord.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fuchs E. Uber Komplikationen der Heterochromie. *Z Augenheilkd* 1906; 15:191.

Rutzen AR, Raizman MB. Fuchs' heterochromic iridocyclitis. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. I.* Philadelphia: WB Saunders, 1994:503.

Smith RE, O'Connor GR. Cataract extraction in Fuchs syndrome. *Arch Ophthalmol* 1974; 91:39.

Fuchs-Lyell Syndrome (Debré-Lamy-Lyell Syndrome; Toxic Epidermal Necrolysis) 490

General: Allergic reaction with severe manifestations; similar to Fuchs-Salzmann- Terrien syndrome (see Fuchs-Salzmann-Terrien Syndrome); may result as a reaction to *Staphylococcus aureus* toxin in children or associated with certain medications, including penicillin, sulfa, nonsteroidal antiinflammatory agents, and allopurinol.

Ocular: Obstruction of nasolacrimal duct; cicatricial changes in conjunctiva and cornea; conjunctivitis; symblepharon; corneal ulceration and possible perforation.

Clinical: Inflammation of mucous membrane with ulcerations; general epidermolysis; cicatricial changes, especially of orifices.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Lyell A. Toxic epidermal necrolysis: eruption resembling scalding of skin. *Dr J Dermatol* 1956; 68:355.

Reich H, Junemann G. Das Debre-Lamy-Lyell-Syndrom. *Klin Monatsbl Augenheilkd* 1970; 157:358.

Westly ED, Wechsler HL. Toxic epidermal necrolysis. *Arch Dermatol* 1984; 120:721.

Fuchs-Salzmann- Terrien Syndrome 491

General: Features similar to those of Fuchs-Lyell syndrome; both are based on drug allergies (antibiotics, sulfonamides, arsenic preparations) (see Fuchs-Lyell Syndrome).

Ocular: Features of Salzmann nodular dystrophy; superficial punctate keratitis; marginal degeneration of the cornea; intraocular hemorrhages; choroidal hemorrhages.

Clinical: Allergic cutaneous lesions with erythema and degrees of exfoliative dermatitis.

Duke-Elder S, ed. *System of Ophthalmology, vol. VIII, part* 2. St. Louis: CV Mosby, 1965.

Fuchs E. Uber knotchenformige Hornhauttrubung. *Graefes Arch Clin Exp Ophthalmol* 1902; 53:423.

Fusobacterium 492

General: Gram-negative; normal inhabitant of mouth and respiratory, intestinal, and urogenital tracts; usually secondary to an underlying disease, surgical procedure, or therapy that impairs the defense of the host; non-spore-forming, nonmotile Gram-negative anaerobic bacilli,

Ocular: Conjunctivitis; dacryocystitis; orbital abscess; orbital cellulitis; corneal ulcer; tenonitis; lid edema; panophthalmitis; gangrene of conjunctiva; cavernous sinus thrombosis; cranial nerve palsy.

Clinical: Brain abscess; pneumonia; liver abscess; endocarditis; sepsis; tissue necrosis.

Bennett KW, Eley A. Fusobacteria: new taxonomy and related diseases. *J Med Microbiol* 1993; 39:246-254.

Burns RP, et al. Unilateral conjunctivitis and canaliculitis due to fusospirochetal infection. *Arch Ophthalmol* 1958; 59:235-242.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5. *part 1,* 4th ed. Baltimore: Williams &Wilkins, 1995.

Weinberg RJ, et al. Fusobacterium in presumed actinomyces canaliculitis. *Am J Ophthalmol* 1977; 84:371-374.

G

G Syndrome (Hypertelorism Esophageal Abnormality and Hypospadias; Hypospadias-Dysphagia Syndrome) 493

General: Neuromuscular defect; autosomal dominant; prevalent in males; males more severely affected (see BBB Syndrome).

Ocular: Retinitis pigmentosa; hypertelorism; narrow palpebral fissures; epicanthal folds; telecanthus.

Clinical: Defect of esophagus; hoarseness; hypospadias; cryptorchidism; imperforate anus; defect of lingual frenulum; deafness; mild mental retardation; dysphagia; anosmia; swallowing difficulties; nasal bridge broad and flat; stridor; aspiration; prominent forehead; cleft lip and palate; laryngotracheal esophageal clefts.

Allanson JE. G syndrome: an unusual family. *Am J Med Genet* 1988; 31:637-642.

Arya S, et al. The G syndrome: additional observations. *Am J Med Genet* 1980; 5:321-324.

Bershof JF, et al. G syndrome: a review of the literature and a case report. *J Craniomaxillofac Surg* 1992; 20:24-27.

Cote GB, et al. The G syndrome of dysphagia, ocular hypertelorism and hypospadias. *Clin Genet* 1981; 19:473-478.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Gaisbock Syndrome (Emotional Polycythemia; Stress Erythrocytosis) 494

General: Prevalent in men, heavy smokers; associated with emotional tension and stress.

Ocular: Afferent pupillary defect; conjunctivitis; central retinal vein occlusion; cystoid macular edema; peripapillary retinal hemorrhage; glaucoma.

Clinical: Obesity; hypertension; stress; vascular disease; plethora.

Avins LR, Krummenacher TK. Venous stasis retinopathy and Gaisbock's syndrome. *Am J Ophthalmol* 1988; 105:420-421.

Gaisbock F. Die Bedeutung des Blutdruckmessung fur die arztlichen Praxis. *Dtsch Arch Klin Med* 1905; 83:363.

Galactosyl Ceramide Lipidosis (Krabbe [1] Syndrome; Infantile Globoid Cell Leukodystrophy; Krabbe Disease; Globoid Cell Leukodystrophy) 495

General: Defect in metabolism of galactocerebroside; genetically determined demyelinating disease that is fatal in early childhood; both sexes affected; onset usually in first year of life; ambiguous onset; autosomal recessive; onset at age 4 to 6 months, although some late-onset cases have been reported; diagnosis is made after identification of "globoid cells" in brain tissue.

Ocular: Photophobia; cortical blindness; optic atrophy; nystagmus.

Clinical: Hypersensitivity to external stimuli; rigidity; vomiting; seizures; episodic fever; mental retardation; death.

Brownstein S, et al. Optic nerve in globoid leukodystrophy (Krabbe's disease). Ultrastructural changes. *Arch Ophthalmol* 1978; 96:864-870.

Fiumara A, et al. Late onset globoid cell leukodystrophy: report on 7 new patients. *Child Nerv Syst* 1990; 6:194-197.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Krabbe KH. A new familial infantile form of brain sclerosis. *Brain* 1916; 39:74.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Gangliosidosis GMI Type 1 (Generalized Gangliosidosis [Infantile]; Norman-Landing Syndrome; Pseudo-Hurler Lipoidosis) 496

General: Absence of A, B, and C isoenzymes of β-galactosidase visceral tissue and mucopolysaccharides in visceral tissues; both sexes affected; autosomal recessive; onset from birth; death from age 6 months to 2 years; defect has been localized to chromosome 3 (3p12-3p13).

Ocular: Macular cherry-red spots; optic disk pallor; nystagmus; esotropia; corneal clouding; retinal artery tortuosity and narrowing; retinitis pigmentosa; macular cherry-red spot found in 50% of patients with this disorder.

Clinical: Cerebral degeneration combined with visceromegaly and skeletal dysplasia; mental and motor retardation; seizures; deafness; spastic quadriplegia; feeding difficulties; recurrent bronchopneumonia; broad nose; frontal bossing; prominent maxilla; hepatosplenomegaly.

Landing BH, et al. Familial neurovisceral lipidosis. *Am J Dis Child* 1964; 108:503-582.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Norman RM, et al. Tay-Sachs disease with visceral involvement and its relation to gargoylism. *Arch Dis Child* 1964; 39:634-640.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV* Philadelphia: WB Saunders, 1994:2777.

Gangliosidosis GM1 Type 2 (Juvenile Gangliosidosis) 497

General: Absence of Band C isoenzymes of β-galactosidase results in neural and visceral deposition of gangliosides and visceral deposition of mucopolysaccharides; autosomal recessive; defective hexosaminidase A; defect localized to chromosome 15 (15q22-15q25.1).

Ocular: Optic atrophy; pigmentary retinopathy; strabismus; macular cherry-red spot; late optic atrophy.

Clinical: Cerebral degeneration; skeletal changes; visceromegaly; psychomotor deterioration; death between ages 3 and 4 years; abnormal acousticomotor reaction; hypotonia; variable hepatosplenomegaly; abnormal hexosaminidases A and B; defect localized to chromosome 5(5q13).

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Harley RD, ed. Pediatric *Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. Principles *and Practice* of *Ophthalmology*, vol. IV. Philadelphia: WB Saunders, 1994:2777.

Ganser Syndrome (Pseudodementia; Nonsense Syndrome; Prison Psychosis Syndrome) 498

General: Found in prisoners and patients with schizophrenia; disparity between the person's complaints and mental alertness.

Ocular: Patient pretends that he or she cannot see or read; no objective findings on examination of visual function.

Clinical: Patient pretends not to know how to do simple things previously familiar to him or her (not to know his or her age, how to spell or read, etc.); mild degree of mental deficiency; amnesia; analgesia; confusion; lethargy; apathetic indifference; headache.

Ganser SJ. Uber Einen Eigenartigen Hysterischen Dammerzustand. *Arch Psychiatr Nervenkrankh* 1898; 30:633.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Gansslen Syndrome (Familial Hemolytic Icterus; Hematologic-Metabolic Bone Disorder) 499

General: Autosomal dominant inheritance; occurs mainly in Caucasians.

Ocular: Hypertelorism; microphthalmos; epicanthus; narrowing of palpebral fissure; lid hemorrhages; myopia; dyschromatopsia; hypochromic heterochromia; scleral icterus; conjunctival hemorrhages; retinal pallor and edema in advanced stages; dilated retinal arteries and veins; round retinal hemorrhages in deeper retinal layers; retinal exudates and macular star.

Clinical: Splenomegaly; hemolytic crises; dental deformities; brachydactyly; polydactyly; congenital hip luxation; oxycephaly; deformities of the outer ear and otosclerosis.

Gansslen M. Uber Hamolytischen Ikterus. *Dtsch Arch Klin Med* 1922; 40:210

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*. 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

GAPO Syndrome (Growth Retardation, Alopecia, Pseudoanodontia, Optic Atrophy Syndrome) 500

General: Autosomal recessive.

Ocular: Progressive optic atrophy; glaucoma; keratoconus.

Clinical: Growth retardation; alopecia; pseudoanodontia; frontal bossing; high forehead; midfacial hypoplasia; wide-open anterior fontanelle; retarded bone age; premature aged appearance; hypogonadism; hepatomegaly; muscular body build.

Gagliardi ART, et al. GAPO syndrome: report of three affected brothers. *Am J Med Genet* 1984; 19:217-223.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Phadke SR, et al. GAPO syndrome in a child without dermal hyaline deposit. *Am J Med Genet* 1994; 51:191-193.

Sayli BS, Gul D. GAPO syndrome in three relatives in a Turkish kindred. *Am J Med Genet* 1993; 47:342-345.

Good Acuity Plus Photosensitivity (GAPP). Track Related Iridiocyclitis and Scleritis (TRISC), Transient Light Sensitivity (TLS) 501

General: Use of intralase to perform LASIK

Ocular: Photophobia; glare; uveitis; iridocyclitis; scleritis

Clinical: Associated with refractive surgery and the use of Intralase technology ; starts 6 to 8 weeks postoperatively and resolves by four to five months.

Binder P: New ocular syndrome surfaces with Intralase. Ophthal News 2004; Aug 12-14

Garcin Syndrome (Half-Base Syndrome; Schmincke Tumor-Unilateral Cranial Paralysis) 502

General: Causes include tumors of nasopharynx, rapidly progressing growth of a sarcoma of base of skull, meningitis, and cranial polyneuritis; cranial nerves VIII to XII are most frequently involved.

Ocular: Ptosis (unilateral); unilateral external ophthalmoplegia; papilledema.

Clinical: Difficulties in swallowing; impairment of speech; hearing defect; respiratory difficulties; sensory disturbances; hoarseness.

Bruce JN, Fetell MR. Tumors of the skull and cranial nerves. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1995:320-329.

Chakrabarti AK. Nasopharyngeal carcinoma with multiple cranial nerve palsies. *Practitioner* 1974; 212: 103.

Garcin R. Le Syndrome Paralytique Unilateral Global des Nerfs Craniens. Doctoral Thesis, University of Paris, 1927.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hochberg F, Pruitt A. Neoplastic diseases of the central nervous system: tumors of the skull base. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:2267.

Gardner Syndrome 503

General: Autosomal dominant; both sexes affected; average onset age 20 years.

Ocular: Exophthalmos; congenital hypertrophy of RPE; multiple lesions of the eye; bilateral occurrence; orbital osteoma; highly pleomorphic pigmentation; unilateral or bilateral retinal lesions; pilomatrixoma-like epidermal cysts; presence of pigmented fundus lesions appears to cluster within families.

Clinical: Intestinal polyps; dermoid tumors; neurofibrous osteomatosis; colon cancer; supernumerary teeth.

Blair NP, Trempe CL. Hypertrophy of the retinal pigment epithelium associated with Gardner's syndrome. *Am J Ophthalmol* 1980; 90:661-667.

Cooper PH. Pilomatrixoma-like changes in the epidermal cyst of Gardner's syndrome. *J Am Acad Dermatol* 1983; 8:639.

Gardner EJ, Richards RC. Multiple cutaneous and subcutaneous lesions occurring simultaneously with hereditary polyposis and osteomatosis. *Am J Hum Genet* 1953; 5: 139.

Heinemann MH, et al. Familial polyposis coli: the spectrum of ocular and other extracolonic manifestations. *Graefes Arch Clin Exp Ophthalmol* 1991; 229:213-218.

Lewis RA, et al. The Gardner syndrome. *Ophthalmology* 1984; 91:916-925.

Whitson WE, et al. Orbital osteoma in Gardner's syndrome. *Am J Ophthalmol* 1986; 101:236-241.

Gastrocutaneous Syndrome 504

General: Combination of symptoms including peptic ulcer, hiatal hernia, multiple lentigines, cafe-au-lait spots, hypertelorism, myopia, acute or chronic; occurs in people with stress who smoke and use ulcerogenic drugs.

Ocular: Hypertelorism; myopia; nystagmus.

Clinical: Upper abdomen pain 1 to 2 hours after eating; pain at night, when gastric secretion is at its peak; nausea; vomiting; excessive salivation.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Gaucher Syndrome (Glucocerebroside Storage Disease; Glucosyl Ceramide Lipidosis; Cerebroside Lipidosis) 505

General: Storage of glucocerebroside in the reticuloendothelial system; autosomal recessive; occurs frequently in Jewish families; onset at any age; onset usually sudden in the infantile form; disease belongs to group of lipid storage disturbances such as ganglioside (Tay-Sachs), sphingomyelin (Niemann-Pick), and ceramide trihexoside (Fabry) (see Tay-Sachs Syndrome; Niemann-Pick Syndrome; Fabry Syndrome); caused by glucosylceramide β-glucosidase (glucocerebrosidase) deficiency; psychomotor deterioration apparent before age 6 months.

Ocular: Strabismus; brown-yellowish, wedge-shaped pinguecula; corneal clouding; oculomotor paralysis; gaze palsies.

Clinical: Infantile form: generalized hypertonia, opisthotonus, dysphagia, vomiting, laryngeal spasm, dyspnea; chronic form: hepatosplenomegaly, lymphadenopathy, mild-to moderate anemia, yellowish-brown patchy skin pigmentation.

Cogan DG, et al. Fundal abnormalities of Gaucher's disease. *Arch Ophthalmol* 1980; 98:2202-2203.

Gaucher PCE. De l'Epithelioma Primitif de la Rate, Hypertrophie Idiopathique de la Rate sans Leucemie. Thesis, University of Paris, 1882.

Petrohelos M, et al. Ocular manifestations of Gaucher's disease. *Am J Ophthalmol* 1975; 80:1006-1010.

Sanders MD, Lake BD. Ocular movements in lipid storage disease. Reports of juvenile Gaucher disease and the ophthalmoplegic lipidosis. *Birth Defects* 1976; 12:535-542.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology. vol. IV* Philadelphia: WB Saunders, 1994.

Gelineau Syndrome (Narcoleptic Syndrome) 506

General: Etiology not well understood; causes include subthalamic lesions, multiple

sclerosis, and tumor of third ventricle; onset in adolescence or early adulthood; male preponderance (6:1); most characteristic sign is sudden attack of sleep that cannot be resisted and may last from only a few minutes to half an hour.

Ocular: Transient blurred vision; inability to read between attacks; bilateral diplopia; spontaneous eyelid closing; flickering vision.

Clinical: Diurnal attacks of short episodes of uncontrollable sleep (usually several times a day); cataplexy with decreased or absent muscle tone and paralysis caused by an upset emotional state; hallucinations.

Gelineau E. De La Narcolepsie. *Gaz Hop Paris* 1880; 53:626,635.

Keefe WP, et al. Ocular manifestations of narcolepsy. *Am J Ophthalmol* 1960; 49:953.

Norman ME, Dyer JA. Ophthalmic manifestations of narcolepsy. *Am J Ophthalmol* 1987; 103:81-86.

General Fibrosis Syndrome (Congenital Enophthalmos with Ocular Muscle Fibrosis and Ptosis; Congenital Fibrosis of the Inferior Rectus with Ptosis; Strabismus Fixus; Vertical Retraction Syndrome (Congenital Fibrosis Syndrome) 507

General: Present from birth; familial history; apparent autosomal dominant transmission; sex-linked recessive transmission also reported.

Ocular: Ptosis; enophthalmos; disk hypoplasia; astigmatism; esotropia; exotropia; hypotropia; nystagmus; visual loss; positive forced duction test; may be associated with Marcus Gunn jaw-winking and synergistic divergence in attempted right gaze.

Clinical: None.

Brodsky MC, et al. Neural misdirection in congenital ocular fibrosis syndrome: implications and pathogenesis. *J Pediatr Ophthalmol Strabismus* 1989; 26:159.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Harley RD, et al. Congenital fibrosis of the extraocular muscles. *J Pediatr Ophthalmol Strabismus* 1978; 15:346-358.

Kim, JH, Hwang JM: Hypoplastic oculomotor nerve in congenital fibrosis syndrome and synergistic divergence with magnetic resonance imaging. *Ophthalmology* 2005; 112: 728-732

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol.* 2. 4th ed. Baltimore: Williams & Wilkins, 1985.

Yazdani A, Traboulsi EI: Classification and surgical management of patients with familial and sporadic forms of congenital fibrosis of the extraocular muscles. *Ophthalmology* 2004; 111: 1035-1042.

Gerlier Disease (Kubisagari; Le Tourniquet; Paralytic Vertigo) 508

General: Cranial nerve dysfunction suggests a basal lesion such as epidemic paralyzed vertigo; infective agent suspected, small Gram-negative coccus from spinal fluid; contact with cows or horses in warm summer months in contaminated stables; pathology unknown; possible brainstem lesion.

Ocular: During attack: hyperemia of fundus oculi, fundus normal, and eyesight normal; between attacks: ptosis, dimness of vision, vertigo, and diplopia.

Clinical: Attack of palsies precipitated by severe exertion, bright light, warmth, hunger, or looking at a moving object (optokinetic irritation); attacks last approximately 10 minutes and may follow each other at very short intervals; mild form not incapacitating; severe form incapacitating; pains in back of neck; nodding of head during attack; hyperreflexia at intervals; attack of temporary palsy muscles, levator palpebrae superior, muscles of back of neck, extension limbs, face, pharynx, and larynx.

Gerlier E. Une Epidemie de Vertige Paralysant. *Rev Med Suisse Romande* 1887; 7:5-29.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

German Syndrome (Fetal Trimethadione; Tridione) 509

General: Both sexes affected; prenatal onset; occurs when trimethadione or paramethadione is taken during pregnancy.

Ocular: Eyebrows up-slanted.

Clinical: Mild brachycephaly; short upturned nose; prominent forehead; cleft lip and palate; micrognathia; ear abnormalities; tetralogy of Fallot; genital abnormalities; hypospadias; hypertrophic clitoris; abnormalities of skin, gastrointestinal, renal, and skeletal systems.

German J, et al. Trimethadione and human teratogenesis. *Teratology* 1970; 3:349-361.

Gerstmann Syndrome (Dominant Hemisphere Syndrome; Left Angular Gyrus Syndrome) 510

General: Most frequently caused by tumors of dominant hemisphere in which the angular gyrus is involved; may follow an ischemic posterior cerebellar artery (PCA) lesion affecting the inferior parietal lobule and resulting in jargon aphasia, dyscalculia, agraphia right-left confusion, constructional dyspraxia, and alexia.

Ocular: Homonymous hemianopsia; visual agnosia for colors.

Clinical: Agraphia (inability to write); finger agnosia; anosognosia; aciculae (difficulties in or inability to perform serial number projects); confusion (mainly between left and right).

Benton AL. Gerstmann's syndrome. *Arch Neurol* 1992; 49:445-447.

Caplan RL. Transient ischemia and brain and ocular infarction. In: Albert DM, Jakobiec FA eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994:2661.

Gerstmann J. Fingeragnosie und Isolierte Agraphie, ein neues Syndrom. Z *Neurol Psychiatr* 1927; 108:152.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Giant Fornix Syndrome 511

General: Enlarged upper fornix with buildup of mucopurulent debri and persistant discharge.

Ocular: Bacterial conjunctivitis; blepharospasm; enlarged fornix; secondary ptosis

Clinical: Recurrent chronic conjunctivitis

Rose GE: The giant fornix syndrome: an unrecognized cause of chronic, relapsing, grossly purulent conjunctivitis. *Ophthalmology* 2004; 111: 1539-45

Giant Papillary Conjunctivitis Syndrome 512

General: Commonly associated with contact lenses (hard and soft), foreign bodies, and ocular prosthesis; immunologic in origin.

Ocular: Ocular irritation; itching of the eye; decreased visual acuity; increased mucous production; papillary changes of the upper tarsal conjunctiva; contact lens coatings; may appear after a lens change from one style to another or by replacement of

the previous design; aging of a lens, particularly of a soft contact lens, may be associated; usually bilateral, although it can be markedly asymmetrical.

Allansmith MR, et al. Giant papillary conjunctivitis induced by hard or soft contact lens wear: quantitative histology. *Ophthalmology* 1978; 85:766-778.

Donshik PC. Giant papillary conjunctivitis. *Trans Am Ophthalmol Soc* 1994; 92:687-744.

Palmisano PC, et al. Causative factors in unilateral giant papillary conjunctivitis. *CLAO J* 1993; 19: 103-107.

Price MJ, et al. Tarsal conjunctival appearance in contact lens wearers. *Contact Lens* 1982; 8:16-22.

Giardiasis 513

General: Multiflagellate protozoan; parasite of human duodenum; encystation occurs in transit through colon.

Ocular: Uveitis; retinal hemorrhages; central serous retinopathy; palpebral edema.

Clinical: Nausea; flatulence; epigastric pain; abdominal cramps; diarrhea; weight loss; malabsorption.

Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Duke-Elder S, MacFaul PA, eds. *System of Ophthalmology, vol. XIII* St. Louis: CV Mosby, 1974.

Ryan SJ, ed. *Retina, vol. II*, 2nd ed. St. Louis: Mosby, 1994.

Gillum-Anderson Syndrome 514

General: Genetic defect responsible for weakness in the orbital connective tissue; proposed connective tissue defect of sclera, zonules, and levator aponeurosis.

Ocular: Dislocated lenses; high myopia; bilateral ptosis typical for levator disinsertions; ectopia lentis.

Gillum WN, Anderson RL. Dominantly inherited blepharoptosis, high myopia and ectopia lentis. *Arch Ophthalmol* 1982; 100:282-284.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Gitelman Syndrome 515

General: Autosomal recessive, renal tubulopathy characterized by hypokalemia, hypomagnesemia, metabolic alkalosis, hypocalciuria, and hypernatremia.

Ocular: Sclerochoroidal calcification.

Clinical: Patients may have arthralgias, seizures, episodes of tetany, muscular weakness, or paresthesia; also may be asymptomatic.

Al-Ghamdi SM, Cameron EC, Sutton RA. Magnesium deficiency: pathophysiologic and *Clinical* review. *Am J Kidney Dis* 1994; 24:737-752.

Bourcier T, et al. Sclerochoroidal calcification associated with Gitelman syndrome. *Am J Ophthalmol* 1999; 128: 767-768.

Cohen SY, Guyot-Sionnest M, Puech M. Choroidal neovascularization as a late complication of hyperparathyroidism. *Am J Ophthalmol* 1998; 126:320-322.

Gitelman HJ, Graham JB, Welt LG. A new familial disorder characterized by hypokalemia and hypomagnesemia. *Trans Assoc Am Physicians* 1966; 79:231-235.

Shields JA. Sclerochoroidal calcification in calcium pyrophosphate dihydrate deposition disease. *Arch Ophthalmol* 1997; 115:1077-1079.

Simon DB, Lifton RP. The molecular basis of inherited hypokalemic alkalosis: Bartter's and Gitelman's syndromes. *Am J Physiol* 1966; 271:F961-F966.

Glander Syndrome 516

General: Serious infection caused by *Malleomyces mallei;* no naturally acquired infections in United States since 1938; transmission from equine animals to man; caused by traumatic inoculations or inhalations; either acute, gangrenous, or chronic ulcerative; fatal usually in 7 to 10 days.

Ocular: Conjunctivitis; dacryocystitis; ulcerating granulomatous orbital lesions; photophobia; lacrimation.

Clinical: Systemic erythematous pustules; inhalation infection; fever; rigors; generalized myalgia lymphangitis; fatigue, headaches; pleuritic chest pain; diarrhea; lymphadenopathy; splenomegaly; mild leukocytosis; multiple subcutaneous and intramuscular abscesses (often arms and legs); visceral involvement, including pulmonary, pleural, skeletal, hepatic, splenic, meningeal, and intracranial.

Braunwald E, et al., eds. *Harrison's principles o/internal medicine,* 11th ed. New York: McGraw-Hill, 1987.

Howe C, et al. The pseudomaller group: a review. *J Infect Dis* 1971; 124:598.

Jackson AE, et al. Recrudescent melioidosis associated with diabetes ketoacidosis. *Arch Intern Med* 1972; 130:268.

Glaucoma, Congenital 517

General: Autosomal recessive; occurs more frequently in males; can occur isolated or associated with other systemic or ocular malformations (dysgenesis of iris, angle and peripheral cornea).

Ocular: Buphthalmos; corneal haze; glaucoma; epiphora.

Clinical: None.

Demenasis F. Further analysis of familial transmission of congenital glaucoma. *Am J Hum Genet* 1983; 35: 1156-1160.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Richt R, Shields B, Krupin T. The glaucomas. St. Louis: Mosby, 1989.

Glaucoma with Elevated Episcleral Venous Pressures 518

General: Autosomal dominant.

Ocular: Open-angle glaucoma; elevated episcleral venous pressure; dilated episcleral veins.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Minas TF, Podos SM. Familial glaucoma associated with elevated episcleral venous pressure. *Arch Ophthalmol* 1968; 80:202-208.

Glaucoma, Goniodysgenesis (Dysgenic Glaucoma; Steroid Glaucoma) 519

General: Autosomal dominant; use of topical dexamethasone under the two-allele system; three genotypes; high, intermediate, and low; more prominent in blacks; mechanism may be related to morphologic changes in the trabecular meshwork.

Ocular: Glaucoma; cataracts.

Clinical: None.

Armaly MF. The heritable nature of dexamethasone induced ocular hypertension. *Arch Ophthalmol* 1966; 75:32-35.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Glaucoma, Hereditary Juvenile 520

General: Autosomal dominant, present at birth; there has been linkage with chromosome 1q21q23; age at diagnosis is 5 to 30 years; positive family history; male-to-female ratio of 2:1.

Ocular: Dysgenesis of the iris and iridocorneal angle; glaucoma; hypoplasia of iris; dark-colored irides; myopia; smooth iris; prominent iris processes; grayish-pale color of trabecular meshwork.

Clinical: None.

Martin JP, Zorab EC. Familial glaucoma in nine generations of South Hampshire family. *Br J Ophthalmol* 1974; 58: 536-542.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Melamed S, Ashkenazi I. Juvenile onset open angle glaucoma. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. III.* Philadelphia: WB Saunders, 1994.

Glaucoma, Recessive Juvenile 521

General: Autosomal recessive; rare; normal parents and consanguineous marriages; asymptomatic and insidious onset.

Ocular: Buphthalmos; aching of eyes; colored halos around lights; elevated intraocular pressure; corneal epithelial edema; severe cupping and atrophy of optic nerve; constriction of visual fields.

Clinical: Headaches.

Beiguelman B, et al. Recessive juvenile glaucoma. *J Genet Hum* 1963; 12:53-54.

Epstein DL. *Chandler and Grant's glaucoma*, 3rd ed. Philadelphia: Lea & Febiger, 1986.

Glucagonoma Syndrome 522

General: Alpha-cell islet tumor of pancreas with retrobulbar neuritis first sign and necrolytic migratory erythema early signs.

Ocular: Central scotoma; retrobulbar neuritis.

Clinical: Necrolytic migratory erythema; diabetes; hypoaminoacidemia and hyperglucagonemia secondary to alpha-cell islet tumor of pancreas; anemia; glossitis; weight loss; angular stomatitis; onycholysis; diarrhea; monochromic monocytic anemia; recurrent venous thrombosis (associated with alpha-cell tumor).

Holmes A, et al. Reversal of a neurologic paraneoplastic syndrome with octreotide (Sandostatin) in a patient with glucagonoma. *Am J Med* 1991; 91:434-436.

Lambrecht ER, et al. Retrobulbar neuritis as first sign of glucagonoma syndrome. *Int Ophthalmol* 1987; 11:14-16.

Vandersteen PR, et al. Glucagonoma syndrome: a clinicopathologic immunocytochemical and ultrastructural study. *J Am Acad Dermatol* 1985; 12:1032-1039.

Goldberg Disease 523

General: Unclassified syndrome with features of mucopolysaccharidoses, sphingolipidoses, and mucolipidoses; deficiency of neuraminidase; located in chromosome 20q 13.1.

Ocular: Macular cherry-red spot; corneal clouding; cerebromacular degeneration.

Clinical: Dwarfism; gargoyle facies; mental retardation; seizures; hearing disorder.

Goldberg MF, et al. Macular cherry red spot, corneal clouding and beta galactosidase deficiency. *Arch Intern Med* 1971; 128:387.

Maumenee AE, Emery JM. An anatomic classification of diseases of the macula. *Am J Ophthalmol* 1972; 74: 594-599.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Thomas GH, et al. Neuraminidase deficiency in the original patient with the Goldberg syndrome. *Clin Genet* 1979; 16:323-330.

Goldenhar Syndrome (Oculo-Auriculo-Vertebral Dysplasia; Goldenhar-Gorlin Syndrome) 524

General: Most cases have been sporadic, but cases of autosomal dominant and recessive inheritance have been reported; male preponderance (60%); present at birth.

Ocular: Anophthalmia; colobomata of choroid, iris, and eyelid; antimongolian slant of lid fissure; epibulbar dermoid or lipodermoids of conjunctiva, cornea, and orbit; tilted optic disk; nerve hypoplasia; microphthalmia; macular heterotopia; tortuous retinal vessels.

Clinical: Frontal bulging of the skull; receding chin; malar hypoplasia; micrognathia and macrostomia; auricular appendices (single or multiple); multiple vertebral anomalies; preauricular fistulas; mental retardation.

Baum JL, Feingold M. Ocular aspects of Goldenhar's syndrome. *Am J Ophthalmol* 1973; 75:250.

Boles DJ. Goldenhar complex in discordant monozygotic twins: a case report and review of the literature. *Am J Med Genet* 1987; 28:103-109.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Goldenhar M. Associations des Malformations de l'Oeil et de l'Oreille, en Particulier la Syndrome Dermoide Epibulbaire-Appendices Auriculaires-Fistula Auris Congenita et Ses Relations avec la Dysostose Mandibulo-Faciale. *J Genet Hum* 1953; 1:243.

Margolis S, et al. Retinal and optic nerve findings in Goldenhar-Gorlin syndrome. *Ophthalmology* 1984; 91: 1327-1333.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Goldscheider Syndrome (Weber-Cockayne Syndrome; Epidermolysis Bullosa; Dominant Epidermolysis Bullosa Dystrophiea Albopapuloidea) 525

General: Rare; Weber-Cockayne syndrome, inherited as an autosomal dominant trait, is actually a milder form without scar formation, whereas Goldscheider syndrome, inherited either autosomal dominant or recessive, shows dystrophic changes with scarring; consanguinity frequent.

Ocular: Blepharitis; shrinkage of conjunctiva; pseudomembrane formation with symblepharon; conjunctivitis; bullous keratitis and subepithelial blisters lead to erosions with subsequent ulcerations and corneal opacities or even perforation; sclera may be similarly involved; lagophthalmos, cicatricial lacrimal stenosis; retinal detachment; cataract; pannus.

Clinical: Vesicular and bullous skin lesions and similar lesions of mucous membranes occur spontaneously or after mild trauma; keloid scars and contraction after healing are common in the dystrophic forms, whereas in the mild form the lesions heal without scarring but may leave some skin pigmentation; growth and mental retardation may be present in the group with recessive inheritance; stenosis of the larynx due to scarring may occur.

Adamis AP, et al. Anterior corneal disease of epidermolysis bullosa simplex. *Arch Ophthalmol* 1993; 111:499-502.

Aurora AL, et al. Ocular changes in epidermolysis bullosa letalis. *Am J Ophthalmol* 1975; 79:464.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Iwamoto M, et al. The ultrastructural defect in conjunctiva from a case of recessive dystrophic epidermolysis bullosa. *Arch Ophthalmol* 1991; 109:1382-1386.

Venugopal NS, Sam-Raj D. Ocular manifestations in bullous dermatoses. *Ind J Ophthalmol* 1977; 25:13-17.

Goltz Syndrome (Focal Dermal Hypoplasia Syndrome) 526

General: X-linked dominant inheritance; lethal in males; skin manifestations present at birth.

Ocular: Microphthalmia; strabismus; coloboma of iris and/or choroid; epiphora; blue sclera; nystagmus; anophthalmos; keratoconus.

Clinical: Skin atrophy and linear pigmentation; telangiectasias of trunk and extremities; superficial, localized fatty skin deposits; multiple papillomas of mucous membranes and periorificial skin (oral, genital, anal); anomalies of extremities with syndactyly, oligodactyly, adactyly; hypohidrosis; paper-thin nails may be present; spina bifida; hypoplasia of right clavicle; umbilical or inguinal hernia.

Goltz RW, et al. Focal dermal hypoplasia. *Arch Dermatol* 1962; 86:708.

Gorski JL. Father to daughter transmission of focal dermal hypoplasia associated with nonrandom X-inactivation: support for X-linked inheritance and paternal X-chromosome mosaicism. *Am J Med Genet* 1991; 40:332-337.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Thomas JV, et al. Ocular manifestations of focal dermal hypoplasia syndrome. *Arch Ophthalmol* 1977; 95: 1997-2001.

Willetts GS. Focal dermal hypoplasia. *Br J Ophthalmol* 1974; 58:620-624.

Gonorrhea 527

General: Caused by *Neisseria gonorrhoeae*, which is transmitted sexually.

Ocular: Conjunctivitis; eyelid edema; keratitis; uveitis.

Clinical: Pelvic inflammatory disease; arthritis; dermatitis; carditis; meningitis.

Dunlop EM, et al. Ophthalmia neonatorum due to beta-lactamase-producing gonococci. *Br Med J* 1980; 281:483.

Fekete T. Antimicrobial susceptibility testing of Neisseria gonorrhoeae and implications for epidemiology and therapy. *Clin Microbiol Rev* 1993; 6:22-33.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Friendly DS. Gonorrheal ophthalmia: reappearance of an old problem. *Trans Am Acad Ophthalmol Otolaryngol* 1970; 74:975-983.

Podgore JK, Holmes KK. Gonococcal infection with minimal or no inflammatory response. *JAMA* 1981; 246: 242-243.

Goodpasture Syndrome 528

General: Chronic, relapsing pulmonary hemosiderosis, often in association with fatal glomerulonephritis; rare; occurs in young males.

Ocular: Episcleritis; juxtapapillary subretinal neovascular membranes; superficial retinal hemorrhages; bilateral peripheral retinoschisis; hemorrhagic and/or exudative retinopathy; nonrhegmatogenous retinal detachment (rare).

Clinical: Cough with recurrent hemoptysis; dyspnea; pulmonary infiltrates; hypochromic iron deficiency anemia; glomerulonephritis; progressive renal failure; diffuse hemorrhagic inflammation of lung.

Bergrem H, et al. Goodpasture's syndrome: a report of seven patients including long-term follow-up of three who received a kidney transplant. *Am J Med* 1980; 68:54-58.

Hoscheit AM, et al. Nomhegmatogenous retinal detachment in Goodpasture's syndrome: a case report and discussion of the clinicopathologic entity. *J Am Optom Assoc* 1993; 64:563-567.

Jampol LM, et al. Ocular *Clinical* findings and basement membrane changes in Goodpasture's syndrome. *Am J Ophthalmol* 1975; 79:452.

Rowe PA, et al. Ophthalmic features of fourteen cases of Goodpasture's syndrome. *Nephron* 1994; 68:52-56.

Gopalan Syndrome (Burning Feet Syndrome; Nutritional Melalgia Syndrome) 529

General: Female preponderance; onset age 20 to 40 years; pantothenic acid deficiency; lack of nicotinic acid, and low-protein diet are contributory factors; found in malnourished populations, detainees in prison camps, and chronic alcoholics.

Ocular: Decreased vision; central or paracentral scotomata.

Clinical: Hyperalgesia; hyperesthesia; severe burning of palms and soles (more pronounced at night); excessive sweating; circulatory insufficiency; tachycardia; muscle atrophy.

Gopalan G. Abstract from *Indian Medical Gazette, Calcutta,* 1946; 81:22. The "burning feet" syndrome. *J Am Med Assoc* 1946; 131:1177.

Vernon S. Nutritional melalgia: a deficiency vascular disease. *J Am Med Assoc* 1950; 143:799.

Gorlin-Chaudhry-Moss Syndrome 530

General: Etiology unknown.

Ocular: Microphthalmia; hypertelorism; depressed supraorbital ridges; inability to open or close lids fully because of incomplete lid development; antimongoloid, oblique palpebral fissures; sparse eyelash development; lid defect (notching); horizontal nystagmus at extreme lateral gaze; limited upper gaze; astigmatism; marked hyperopia; corneal scars (possibly due to exposure keratitis); keratoconus.

Clinical: Craniofacial dysostosis; saddled appearance of upper face; high-arched, narrow palate; dental anomalies (size, number, position); hypertrichosis; hypoplasia of labia majora; patent ductus arteriosus; normal mental development; fatigue; frontal headache.

Cohen MM, Putnam TI. An 18p21 q translocation in patient with presumptive "monosomy G." *Am J Dis Child* 1972; 124:908.

Gorlin RJ. Craniofacial dysostosis, patent ductus arteriosus, hypertrichosis, hypoplasia of labia majora, dental and eye anomalies. *J Pediatr* 1960; 56:778.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Gout (Hyperuricemia) 531

General: Genetic disease of purine metabolism and renal excretion of uric acid.

Ocular: Conjunctivitis; episcleritis; posterior scleritis; ocular motor disturbances; iritis; band keratopathy; interpalpebral paralimbal nodules.

Clinical: Acute inflammatory arthritis; accumulation of sodium urate deposits; uric acid nephrolithiasis; renal failure; tophi in any body tissue; marked swelling of feet and ankles.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Coassin M, Piovanetti O, Stark WJ, et al. Urate deposition in the iris and anterior chamber. *Ophthalmology* 2006; 113: 462-465.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fishman RS. Band keratopathy in gout. *Arch Ophthalmol* 1966;75: 367.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gradenigro Syndrome (Temporal Syndrome; Lannois-Gradenigro Syndrome) 532

General: Caused by extradural abscess of the petrous portion of the temporal bone; good prognosis.

Ocular: Ipsilateral paralysis (cranial nerve VI); transient involvement of cranial nerves III and IV occasionally present; severe pain in area of ophthalmic branch (cranial nerve V); photophobia; lacrimation; reduced corneal sensitivity; optic nerve involvement occasionally present.

Clinical: Inner ear infection with deafness; mastoiditis; facial paresis possible; temperature may be elevated; meningeal signs possible; can occur rarely as a complication of otitis media.

De Graaf J, et al. Gradenigo's syndrome: a rare complication of otitis media. *Clin Neural Neurosurg* 1988; 90:237.

Gradenigro G. A special syndrome of endocranial otitic complications. *Ann Otol Rhinol Laryngol* 1904; 13:637.

Joffe WS. Clinical nerve disease. *Int Ophthalmol Clin* 1967; 7:823.

Graft Versus Host Disease 533

General: Major complication of bone marrow transplantation; donor T lymphocytes attack recipient's cells; targets are skin, liver, intestine, oral mucosa, conjunctiva, lacrimal gland, vaginal mucosa, and esophageal mucosa.

Ocular: Keratoconjunctivitis; photophobia; hemorrhagic conjunctivitis; proptosis; intraretinal hemorrhages; nerve palsy; herpes simplex/herpes zoster manifestations; uveitis; corneal epithelial denudement; conjunctival scarring; dry eye syndrome; corneal melt; dacryoadenitis; keratoconjunctivitis sicca; cataract; retinitis.

Clinical: Leukemia; aplastic anemia; exanthematous dermatitis; hepatitis; enteritis; scleroderma-like involvement of skin; chronic liver dysfunction; recurrent bacterial infections.

Foster CS. Immunologic disorders of the conjunctiva, cornea and sclera. In: Albert DM, Jakobiec FA, eds. Principles *and Practice* of *Ophthalmology*, vol. I. Philadelphia: WB Saunders, 1994.

Jack MK, et al. Ocular manifestations of graft versus host disease. Arch Ophthalmol 1983; 101:1080-1084.

Lawley TJ, et al. Sjogren-like syndrome and chronic graft versus host disease. Ann Intern Med 1977; 87:707-709.

Granuloma Annulare 534

General: Benign; self-limited dermatosis; etiology unknown but reported to follow insect bites, sun exposure, trauma, viral infection, and psoralen ultraviolet radiation therapy; hereditary predisposition; seen in children and young adults.

Ocular: Granuloma of lid; predilection for lateral upper lid and outer canthus; lesions are clinically similar to the subcutaneous nodules of rheumatoid arthritis.

Clinical: Skin lesions localized, generalized, subcutaneous, perforating, or arcuate dermal erythema; papules often arranged in a complete or half circle.

Fitzpatrick TB, Freedberg IM. *Dermatology* in *general medicine.* 3rd ed. New York: McGraw-Hill, 1987.

Rao NA, Font RL. Pseudorheumatoid nodules of the ocular adnexa. *Am J Ophthalmol* 1975; 79:471-478.

Muhlbauer QE. Granuloma Annulare. *J Am Acad Dermatol* 1980; 3:217.

Granuloma Faciale 535

General: Uncommon disease; etiology unknown; characterized by single or multiple cutaneous nodules usually occurring on the face; asymptomatic; most common in males; seen in whites, rarely in blacks and Japanese.

Ocular: Unusual eyelid nodules.

Clinical: Cutaneous nodules most often on face but may appear anywhere; lesions are soft, elevated, well-circumscribed nodules, from a few millimeters to several centimeters in size; extrafacial lesions are extremely rare but have been reported.

Fitzpatrick TB, Freedberg IM. *Dermatology* in *general medicine.* 3rd ed. New York: McGraw-Hill, 1987.

Jacyk WK. Facial granuloma in a patient treated with clofazimine. *Arch Dermatol* 1981; 117:597.

Konohana A. Extrafacial granuloma faciale. *J Dermatol* 1994; 21:680-682.

Granuloma Venereum 536

General: Donovania granulomatis; infective venereal disease; prevalent in black women; *Chlamydia trachomatis* is an intracellular bacterium lacking respiratory enzymes that has an affinity for mucosal epithelium; serotypes A through C have been epidemiologically associated with trachoma; serotypes E through K have been associated with genital infection and keratoconjunctivitis in sexually active adults and neonates; other serotypes have been associated with lymphogranuloma venereum and Reiter syndrome.

Ocular: Lid and orbit granulomas.

Clinical: Painless primary lesions; painful secondarily infected ulcers.

Coovadia YM, et al. Granuloma inguinale of the oral cavity. *S Afr Med J* 1985; 68:815-817.

Duke-Elder S, MacFaul PA. *System of Ophthalmology, vol. XIII* St. Louis: CV Mosby, 1974.

Malaty R. Chlamydia. In: Tabbara K, Hyundiuk RA, eds. *Infections of the eye.* Boston: Little, Brown and Company, 1986:107,114.

Gray Iris Syndrome 537

General: Excessive trauma of iris at time of lens implantation with loss of posterior iris pigment; originally had blue irides.

Ocular: Pigmentary glaucoma; gray iris; massive pigment deposits in chamber angle; nonfixated intraocular lens.

Clinical: None.

Huber C. The gray iris syndrome. *Arch Ophthalmol* 1984; 102:397-398.

Simcoe CW. An ounce of prevention. *Am Intraocular Implant Soc* 1978; 4:39-44.

Grayson-Wilbrandt Syndrome (Reis-Buecklers Syndrome; Corneal Dystrophy of Reis-Buecklers) 538

General: Onset at end of first decade; infrequent episodes of eye redness and pain; autosomal dominant trait; electron microscopy reveals peculiar curly material in the subepithelial fibrous tissue that parallels the distribution of attachment proteins.

Ocular: Corneal changes variable from a mottled scarring to gray macular opacities of the anterior limiting membrane of the cornea; strabismus.

Clinical: None.

Buecklers M. Ueber Eine Weltere Familiaere Hornhautdystrophie (Reis). *Klin Monatsbl Augenheilkd* 1949; 144: 386-397.

Grayson M, et al. Dystrophy of the anterior limiting membrane of the cornea (Reis-Buecklers type). *Am J Ophthalmol* 1966; 61:345-349.

Lohse E, et al. Reis-Bucklers corneal dystrophy: immunofluorescent and electron microscopic studies. *Cornea* 1989; 8:200.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pavan-Langston D. Reis-Buecklers dystrophy. In: Pavan-Langston D, ed. *Manual of ocular diagnosis and therapy*, 4th ed. Boston: Little, Brown and Company, 1995:106.

Greig Syndrome (Ocular Hypertelorism Syndrome; Hypertelorism; Primary Embryonic Hypertelorism; Hypertelorism Ocularis) 539

General: Condition is rare; sporadic or hereditary; autosomal dominant or sex linked; if not associated with mental deficiency, then adequate mental and physical development is found.

Ocular: Hypertelorism (wide spacing of orbits); enophthalmos; epicanthus; deformities of eyelids and brows; defects of the palpebral fissure; bilateral sixth nerve paralysis; esotropia; astigmatism; optic atrophy by tension on the optic nerve; strabismus.

Clinical: Skull may show mild malformations, including bitemporal eminences and decreased anteroposterior diameter; harelip; high-arched palate; cleft palate; broad and flat nasal root; mental impairment.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Fuller RW, Weber AN. Hypertelorism in association with sex chromatin abnormality in two siblings. *Am J Ment Defic* 1961-1962; 66:844.

Greig DM. Hypertelorism. A hitherto undifferentiated congenital-craniofacial deformity. *Edinburgh Med J* 1924; 31: 560-593.

Grönblad-Strandberg Syndrome (Systemic Elastodystrophy; Pseudoxanthoma Elasticum; Elastorrhexis; Darier-Grönblad-Strandberg Syndrome) 540

General: Autosomal recessive; female-to-male ratio of 2:1; inheritance is usually autosomal recessive, but it also has reported as autosomal dominant.

Ocular: "Angioid streaks" of the retina; macular hemorrhages and transudates not infrequent; choroidal sclerosis; retinal detachment; keratoconus; cataract; paralysis of extraocular muscles (secondary to vascular lesions of central nervous system); subluxation of lens; exophthalmos; optic atrophy; vitreous hemorrhages; Salmon spot multiple atrophic peripheral RPE lesions; reticular pigment dystrophy of the macula; optic disk drusen; multiple small crystalline bodies associated with atrophic RPE changes.

Clinical: Pseudoxanthoma elasticum with thickening, softening, and relaxation of the skin; skin changes are symmetrical in skin folds near large joints (axilla, elbow, inguinal region, lower abdomen, neck); flattening of the pulse curve and peripheral vascular disturbances; gastrointestinal hemorrhages.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Grand MG, et al. Angioid streaks associated with pseudoxanthoma elasticum in a 13-year old patient. *Ophthalmology* 1987; 94:197-200.

Guyer DR, Gragoudas ES, D'Amico DJ. Angioid streaks. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994.

Hull DS, Aaberg TM. Fluorescein study of a family with angioid streaks and pseudoxanthoma elasticum. *Br J Ophthalmol* 1974; 58:738-745.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Schilling JS, Blach RK. Prognosis and therapy of angioid streaks. *Trans Ophthalmol Soc U K* 1975; 95:301-306.

Grouped Pigmentation of the Macula 541

General: Autosomal recessive.

Ocular: Grouped pigmentation limited to foveal area; metamorphopsia; pigmented spots around a clear hole in the foveal area.

Clinical: None.

Forsius H, et al. A genetic study of three rare retinal disorders: dystrophia retinae dysacusis syndrome, X-chromosomal retinoschisis and grouped pigments of the retina. *Birth Defects* 1971; 3:83-98.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Renardel de Lavalette, et al. Familial congenital grouped pigmentation of the retina. *Am J Ophthalmol* 1991; 112: 406-409.

Gruner-Bertolotti Syndrome 542

General: Various causes, including pineal tumor, supranuclear lesions, thrombosis of anterior choroidal artery, aneurysm or tumor; combination of Parinaud and von Monakow syndromes (see Parinaud Syndrome; von Monakow Syndrome).

Ocular: Hemianopia; lid retraction; ptosis; extraocular muscle paralysis; papilledema.

Clinical: Vertigo; hemiplegia; sensory disturbances; brain tumors.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Parinaud H. Paralysie des mouvements associes des yeux. *Arch Neurol* 1883; 5:145-172.

Guillain-Barré Syndrome (Landry Paralysis; Acute Infectious Neuritis; Acute Polyradiculitis; Acute Febrile Polyneuritis; Acute Idiopathic Polyneuritis; Inflammatory Polyradiculoneuropathy; Landry-Guillain-Barré-Strohl Syndrome; Postinfectious Polyneuritis) 543

General: Etiology unknown; occurs from age 16 to 50 years; Fisher syndrome is a variant (see Fisher Syndrome).

Ocular: Facial nerve paralysis with paralytic ectropion of the lower eyelid; mild-to-complete external ophthalmoplegia; optic neuritis; papilledema; ptosis; anisocoria; nystagmus; dyschromatopsia; scotoma; bilateral tonic pupils.

Clinical: Polyneuritis involving facial peripheral motor nerves and spinal cord; facial diplegia; bladder incontinence; variable degrees of paralysis, usually beginning in lower extremities; tendon reflexes absent; involvement of respiratory muscles possible; paresthesia (symmetrical).

Anzai T, et al. Guillain-Barre syndrome with bilateral tonic pupils. *Int Med* 1994; 33:248-251.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Guillain GA. Sur un Syndrome de Radiculoneurite avec Hyperalbuminose du Liquide Cephalo-Rachidien sans Reaction Cellulaire. Remarques sur les Caracteres Cliniques et Graphiques des Reflexes Tendineux. *Bull Mem Sec Med Hop (Paris)* 1916; 40:1462.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Gyrate Atrophy (Ornithine Ketoacid Aminotransferase Deficiency) 544

General: Deficiency of the enzyme ornithine aminotransferase; autosomal recessive; chronic, progressive dystrophy; responsible human gene has been localized to chromosome 10.

Ocular: Chorioretinal atrophy; crystalline deposits associated with brown pigment in fundus; myopia; cataract; keratoconus; night blindness; constricted visual fields; axial hypermetropia; cobblestone-like peripheral lesions; blunting of ciliary processes; iris atrophy.

Clinical: Absence of enzyme ornithine ketoacid transaminase; elevated levels of amino acid ornithine in body fluids; seizures; abnormal electroencephalography; eosinophilic subsarcolemmal deposits are seen on muscle biopsy; massive cystinuria, and lysinuria; diabetes.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WE Saunders, 2000.

Khan MY, et al. Gyrate atrophy of the choroid and retina with hyperornithinaemia, cystinuria and lysinuria. Eye 1994; 8[Pt 3]:247-248.

Wilson DJ, et al. Ocular clinicopathologic study of gyrate atrophy. Am J Ophthalmol 1991; 111:24-33.

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Haemophilus Aegyptius (Koch-Weeks Bacillus) 545

General: Caused by gram-negative Koch-Weeks bacillus in warm-climate regions; characterized by a 24- to 48-hour incubation period; now classified as *Haemophilus influenzae* biotype III; *H influenzae* is divided into biotypes based on biochemical reactions (indole production, urease activity, ornithine decarboxylase activity) and into serotypes based on their capsular polysaccharides; common cause of purulent conjunctivitis and preseptal cellulitis in children.

Ocular: Conjunctivitis; corneal opacity; corneal ulcer; phlyctenular keratoconjunctivitis; keratitis; cellulitis of lid; pseudoptosis; uveitis; petechial subconjunctival hemorrhages.

Clinical: Coryza; systemic symptoms are rare.

Allen JH. Common external ocular infections. *Postgrad Med* 1969; 45:144-148.

Brinser JH. Ocular bacteriology. In: Tabbara KF, Hyndiuk RA, eds. *Infections of the eye.* Boston: Little, Brown and Company, 1986:115-150.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WE Saunders, 2000.

Goldsmith MO. Diseases of the conjunctiva. *Am Fam Physician* 1970; 2:100-108.

Haemophilus Influenzae 546

General: Gram-negative rod.

Ocular: Conjunctivitis; cellulitis; tenonitis; uveitis; vitreous opacity; pannus; corneal opacity.

Clinical: Pharyngitis; epiglottitis; laryngotracheitis; pneumonia; bronchitis; otitis media; meningitis; cellulitis; septic arthritis; sinusitis.

Cohen KL, McCarthy LR. Haemophilus influenzae ophthalmia neonatorum. *Arch Ophthalmol* 1980; 98:1214-1215.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Haddad NA, McPhail JJ. Mucopurulent conjunctivitis and Haemophilus. *Am J Ophthalmol* 1969; 68:35-42.

Londer L, Nelson DL. Orbital cellulitis due to Haemophilus influenzae. *Arch Ophthalmol* 1974; 91:89-91.

Moxon ER. Haemophilus influenzae. In: Mandell GL, Douglas RG Jr, Bennett JE, eds. *Principles and Practice of infectious diseases,* 3rd ed. New York: Churchill Livingstone, 1990: 1722-1729.

Moxon ER. Molecular basis of invasive Haemophilus Influenzae type b disease. *J Infect Dis* 1992; 165[Suppl 1]: 577-581.

Hajdu-Cheney Syndrome 547

General: Rare; autosomal dominant; disorder of bone metabolism

Ocular: Bilateral visual loss; choroidal folds; optic nerve head swelling; optic neuropathy; optic nerve meningocele

Clinical: Facial dysmorphism; progressive platybasia; syringomyelia; curvature of the spine; aplasis of facial sinuses

DiRocco F, Oi S: Spontaneous regression of syringomyelia in Hajdu-Cheney syndrome with severe platybasia. Case report. J Neurosurg 2005; 103: 194-197.

Golnik KC, Kersten RC: Optic nerve head swelling in the Hadju-Cheney syndrome. J Neurophthalmol. 1998; 18: 60-65.

Hallermann-Streiff Syndrome (Dyscephalic-Mandibulo-Oculo-Facial Syndrome; Oculo-Mandibulo-Dyscephaly; Ullrich-Fremery-Dohna Syndrome; Francois Dyscephalic Syndrome; Mandibulo-Oculo-Facial Dyscephaly Syndrome; Francois-Hallermann-Streiff Syndrome; Hallermann-Streiff-Francois Syndrome; Audry I Syndrome; Dohna Syndrome; Francois Syndrome (1); Dyscephaly- Teeth Abnormality-Dwarfism; Dyscephalia Oculomandibularis-Hypotrichosis; Mandibulo-Ocular Dyscephalia Hypotrichosis; Fremery-Dohna Syndrome; Oculo-Mandibulo-Facial Dyscephaly) 548

General: Rare; familial occurrence and consanguinity; males and females equally affected.

Ocular: Microphthalmos (bilateral); proptosis; nystagmus; strabismus; cataracts; bilateral optic atrophy; coloboma of optic disk, choroid, and iris; keratoglobus; microcornea; antimongoloid slant; iris atrophy; uveitis; blue sclera; persistent pupillary membrane; secondary glaucoma.

Clinical: Malformations of skull (brachycephaly), facial skeleton, and jaws; erupted teeth at birth; diminished hair growth; hyperextensibility of joints; short stature; skin atrophy; mental deficiency; predisposition to upper airway compromise; obstructive sleep apnea.

Francois J, Victoria-Troncoso V. Francois' dyscephalic syndrome and skin manifestations. *Ophthalmologica* 1981; 183:63-67.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hallermann W. Vogelgesicht und Cataracta Congenita. *Klin Monatsbl Augenheilkd* 1948; 113:315.

Ronen S, et al. The early management of baby with Hallermann-Streiff-Francois syndrome. *J Pediatr Ophthalmol Strabismus* 1979; 16:119-121.

Spaepen A, et al. Hallermann-Streiff syndrome: *Clinical* and psychological findings in children. Nosologic overlap with oculodentodigital dysplasia? *Am J Med Genet* 1991; 41:517-520.

Streiff EB. Dysmorphic Mandibulo-faciale (Tete d'Oiseau) et Alterations Oculaires. *Ophthalmologica (Basel)* 1950; 120:79.

Hallervorden-Spatz Syndrome (pigmentary Degeneration of Globus Pallidus; Progressive Pallidal Degeneration Syndrome 549

General: Etiology unknown; autosomal dominant; onset between age 7 and 9 years; globus pallidus and pars reticularis of the substantia nigra are involved with demyelinization and degenerative processes; perhaps a form of iron storage disease.

Ocular: Nystagmus; retinitis pigmentosa; optic nerve atrophy; degeneration of photoreceptors; retinal gliosis; narrowing and obliteration of blood vessels with perivascular cuffing; degeneration of retinal pigment epithelial cells.

Clinical: Slowly progressing spasticity and rigidity of the extremities; emotional disturbances (pseudobulbar type); dementia; clubfoot; dysphagia; athetosis; dysphonia; choreoathetosis; rigidity; seizures; pyramidal signs; generalized dystonia.

Geeraets WJ. *Ocular Syndromes.* Philadelphia: Lea & Febiger, 1976.

Hallervorden J, Spatz H. Eigenartige Erkrankung im Extrapyramidalen System mit Besonderer Beteiligung des Globus Pallidus und der Substantia Nigra: Ein Beitrag zu den Beziehungen Zwischen Diesen Beiden Zentren. Z *Neurol Psychiatr* 1922; 79:254.

Martin JJ, et al. Niemann-Pick disease (Crocker group A). Late onset and pigmentary degeneration resembling Hallervorden-Spatz syndrome. *Arch Neurol* 1972; 27:45.

Orrell RW, et al. Acanthocytosis, retinitis pigmentosa, and pallidal degeneration: a report of three patients, including the second reported case with hypoprebetalipoproteinemia (HARP syndrome). *Neurology* 1995; 45[3 Pt I]: 487-492.

Tripathi RC, et al. Clinicopathologic correlation and pathogenesis of ocular and central nervous system manifestations in Hallervorden-Spatz syndrome. *Acta Neuropathol* 1992; 83:113-119.

Hallgren Syndrome (Retinitis Pigmentosa-Deafness-Ataxia Syndrome; Usher Syndrome Type I) 550

General: Autosomal recessive inheritance.

Ocular: Horizontal nystagmus (10%); cataract; retinitis pigmentosa; retinal atrophy; narrow retinal vessels; optic atrophy; keratoconus.

Clinical: Congenital deafness (complete or at least severe auditory impairment); mental deficiency (25%); vestibulocerebellar ataxia (90%); schizophrenia-like symptoms (25%).

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hallgren B. Retinitis pigmentosa combined with congenital deafness with vestibulocerebellar ataxia and neural abnormality in a proportion of cases: a *Clinical* and geneticostatistical study. *Acta Psychiatr Scand Suppl* 1959-1960; 138:1-101.

Kaplan J, et al. Probable location of Usher type I gene on chromosome 14q by linkage with D14S13 (MLJ14 probe). *Cytogenet Cell Genet* 1991; 58:1988(abst).

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Nuutila A. Dystrophia retinae pigmentosa-dysacusis syndrome (DRD): a study of the Usher and Hallgren syndromes. *J Genet Hum* 1970; 18:57.

Hamman-Rich Syndrome (Alveolar Capillary Block Syndrome; Diffuse Pulmonary Fibrosis Syndrome; Rheumatoid Lung Syndrome) 551

General: Etiology unknown; insidious onset with progressive exertional dyspnea; association with rheumatoid arthritis or scleroderma; autosomal recessive; occurs between age 40 and 50 years.

Ocular: Xerophthalmia; keratomalacia; retinal venous congestion and engorgement; ischemic retinopathy; cystic macular changes.

Clinical: Cyanosis; dyspnea; cough; weight loss; clubbing of fingers; high sodium and chloride concentrations in sweat; heart failure.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hamman L, Rich RR. Acute diffuse interstitial fibrosis of lungs. *Bull Johns Hopkins Hosp* 1944; 74:177.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Spalter HF. Cystic fibrosis and the eye. *J Pediatr Ophthalmol* 1971; 8:6.

Haney-Falls Syndrome (Congenital Keratoconus Posticus Circumscriptus Syndrome) 552

General: Etiology unknown; autosomal dominant or recessive.

Ocular: Hypertelorism (mild); lateral canthi are displaced upward; myopic astigmatism; sharply localized posterior curvature of the cornea; corneal nebulae.

Clinical: Mental retardation; retarded growth; broad nose; brachydactyly; pterygium colli; barrel chest.

Haney WP, Falls HF. The occurrence of congenital keratoconus posticus circumscriptus in two siblings presenting a previously unrecognized syndrome. *Am J Ophthalmol* 1961; 52:53.

Jacobs HB. Posterior conical cornea. *Br J Ophthalmol* 1957; 41:31.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Hanhart Syndrome (Richner Syndrome; Recessive Keratosis Palmoplantaris; Pseudoherpetic Keratitis; Richner-Hanhart Syndrome; Tyrosinemia II; Tyrosinosis; Pseudodendritic Keratitis) 553

General: Autosomal recessive; consanguinity.

Ocular: Excess tearing; photophobia; dendritic lesions of the cornea with corneal sensitivity not affected; keratitis; papillary hypertrophy of conjunctiva; corneal haze; neovascularization of cornea; cataract; nystagmus.

Clinical: Dyskeratosis palmoplantaris; diffuse keratosis; dystrophy of nails; hypotrichosis; mental retardation (usually pronounced); sensorineural hearing loss.

Bardelli AM, et al. Familial tyrosinaemia with eye and skin lesions. Presentation of two cases. *Ophthalmologica* 1977; 175:5-9.

Bienfang DC, et al. The Richner-Hanhart syndrome: report of a case with associated tyrosinemia. *Arch Ophthalmol* 1976; 94: 1133-1137.

Burns RP, et al. Keratopathy in tyrosinemia. *Birth Defects* 1976; 12:169-180.

Chandra Sekhar HK. Hanhart's syndrome with special reference to temporal bone findings. *Ann Otol Rhinol Laryngol* 1987; 96:309-314.

Charlton KH, et al. Keratitis and systemic tyrosinemia. *Ophthalmology* 1981; 88:355-360.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Goldsmith LA, Reed J. Tyrosine-induced eye and skin lesions: a treatable genetic disease. *JAMA* 1976; 236:382-384.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Hansen Disease (Leprosy) 554

General: Communicable disease caused by *Mycobacterium leprae.*

Ocular: Keratitis; leukoma; pannus; corneal ulcer; uveitis; iris atrophy; dacryocystitis; anisocoria; multiple pupils; decreased or absent pupillary reaction to light; paralysis of seventh nerve; episcleritis; blepharospasm; lagophthalmos; madarosis; secondary glaucoma; decreased intraocular pressure; subconjunctival fibrosis; punctate epithelial keratopathy; posterior subcapsular cataract; corneal hypesthesia; prominent corneal nerves; iridocyclitis; foveal avascular keratitis; scleritis; interstitial keratitis; iris pearls; dry eye.

Clinical: Disease affects primarily the skin, mucous membrane, and peripheral nerves.

Binford CH, et al. Leprosy. *JAMA* 1982; 247:16.

Dana MR. Ocular manifestations of leprosy in a noninstitutionalized community in the United States. *Arch Ophthalmol* 1994; 112:626-629.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hornblass A. Ocular leprosy in South Vietnam. *Am J Ophthalmol* 1973; 75:478.

Schwab IR, et al. Leprosy in a trachomatous population. *Arch Ophthalmol* 1984; 102:240-244.

Sekhar GC, et al. Ocular manifestations of Hansen's disease. *Doc Ophthalmol* 1994; 87:211-221.

Shields JA, et al. Ocular findings in leprosy. *Am J Ophthalmol* 1974; 77:880-890.

Happy Puppet Syndrome (Puppet Children) 555

General: Etiology unknown; very rare form of infantile epilepsy.

Ocular: Optic atrophy; deficiency of choroidal pigment; lightly colored irides; Brushfield spots; retinal pigment epithelium abnormalities; heterotropia; blindness.

Clinical: Mental retardation; seizures; puppet-like ataxia; paroxysms of laughter; absent speech; microcephaly; horizontal occipital depression; brachycephaly; prognathism, abnormal electroencephalographic findings.

Angelman H. "Puppet" children: a report on three cases. *Dev Med Child Neurol* 1965; 7:681-688.

Bower BD, Jeavons PM. The "happy puppet" syndrome. *Arch Dis Child* 1967; 42:298-302.

Massey JY, Roy FH. Ocular manifestations of the happy puppet syndrome. *J Pediatr Ophthalmol* 1973; 10:282-284.

Harboyan Syndrome (Congenital Corneal Dystrophy and Sensorineural Hearing Loss; Maumenee Syndrome; Corneal Dystrophy, Congenital Hereditary Endothelial) 556

General: Autosomal recessive; both sexes affected; corneal edema present at birth; slow and progressive; both dominant and recessive forms of this disorder have been described.

Ocular: Bluish-white opacities of cornea with normal sensitivity and no vascularization; nystagmus; keratoconus.

Clinical: Sensorineural hearing loss with childhood onset.

Harboyan A, et al. Congenital corneal dystrophy, progressive sensorineural deafness in a family. *Arch Ophthalmol* 1971; 85:27-32.

Kirkness CM, et al. Congenital hereditary corneal edema of Maumenee: Its *Clinical* features, management and pathology. *Br J Ophthalmol* 1987; 71:130-144.

Maumenee AE. Congenital hereditary corneal dystrophy. *Am J Ophthalmol* 1960; 50:1114-1124.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Regenbogen LS, Coscas GJ. *Oculo-auditory syndromes.* New York: Masson, 1985.

Harlequin Syndrome (Bullous Ichthyosiform Erythroderma; Collodion Baby; Congenital Ichthyosis; Epidermolytic Hyperkeratosis; Ichthyosis; Ichthyosis Vulgaris; Lamellar Ichthyosis; Nonbullous Ichthyosiform Erythroderma; Xeroderma; X-Linked Ichthyosis) 557

General: Autosomal inherited disorder; affects both sexes; normal at birth; onset within first 7 days.

Ocular: Keratopathy; corneal scarring; keratitis; conjunctivitis; lagophthalmos; photophobia; ectropion; lid erythema; lacrimation.

Clinical: At birth, the skin surface is moist, red, and tender; within several days, thick verrucous scales form.

Chua CN, Ainsworth J. Ocular management of harlequin syndrome: photo essay. *Arch Ophthalmol* 2001; 119: 454-455.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Frost P. Disorders of cornification. In: Moschella SL, et al., eds. *Dermatology.* Philadelphia: WB Saunders, 1975: 1056-1084.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Orth DH, et al. Collodion baby with transient bilateral lid ectropion. Review of ocular manifestations of ichthyosis. *Arch Ophthalmol* 1974; 91:206-207.

Hartnup Syndrome (pellagra-Cerebellar Ataxia-Renal Aminoaciduria Syndrome; H Disease; Niacin Deficiency) 558

General: Recessive; inborn error in amino acid metabolism with abnormal metabolism of tryptophan; both sexes affected; presents from infancy.

Ocular: Ectropion; symblepharon; nystagmus; scleral ulcers; corneal leukoma; photophobia; diplopia during attacks.

Clinical: Dermatitis (similar to pellagra) with skin eruptions; progressive mental retardation; cerebellar ataxia.

McLaren DS. Cutaneous changes in nutritional disorders. In: Fitzpatrick TB, Eisen AZ, Wolff K, et al., eds. *Dermatology in General Medicine*, 3rd ed. New York: McGraw-Hill, 1987:1601-1613.

Scriver CR. Hartnup disease: a genetic modification of intestinal and renal transport of certain neutral alpha-amino acids. *N Engl J Med* 1965; 273:530.

Singh S, et al. Hartnup disease: a case report. *Ind Pediatr* 1978; 15:191-193.

Hays-Wells Syndrome (AEC Syndrome; Ankyloblepharon-Ectodermal Defects-Cleft Lip/Palate Syndrome) 559

General: Autosomal dominant disease, as initially described, but may exist as an autosomal recessive disorder.

Ocular: Ankyloblepharon, filiforme adnatum (fused eyelids).

Clinical: Coarse, wiry, sparse hair; dystrophic nails; slight hypohidrosis; scalp infections; hypodontia; maxillary hypoplasia; cleft lip and palate.

Greene SL, Michels VV, Doyle JA. Variable expression in ankyloblepharon-ectodermal defects-cleft lip and palate syndrome. *Am J Med Genet* 1987; 27:207-212.

Seres-Santamaria A, Arimany JL, Muniz F. Two sibs with cleft palate, ankyloblepharon, alveolar synechiae, and ectodermal defects: a new recessive syndrome? *J Med Genet* 1993; 30:793-795.

Shwayder TA, Lane AT, Miller ME. Hays-Wells syndrome. *Pediatr Dermatol* 1986; 3:399-402.

Headache Neurologic Defects and Cerebrospinal Fluid Lymphcytosis Syndrome 560

General: Age range 7-52 years of age; no gender bias

Ocular: Papillema; homonymous hemianopia; photopsias; sixth nerve palsy

Clinical: Headache; hemisensory defects; muscle weakness; aphasia; elevated intracranial pressure

Morrison DG, Phuah HK, Reddy AT, et al. Ophthalmologic involvement in the syndrome of headache, neurologic deficits, and cerebrospinal fluid lymphocytosis. *Ophthalmology* 2003; 110: 115-118

Head-Riddoch Syndrome 561

General: Occurs in quadriplegics; caused by distention of a viscus below the level of spinal cord lesion; seen most frequently in people with high cervical cord lesion; may follow catheter obstruction, fecal impaction, bladder calculi, urinary infection, or decubiti.

Ocular: Dilated pupils; blurred vision.

Clinical: Sweating; flushing; pilomotor activity; nasal stuffiness; headaches; generalized seizures; bradycardia; hypertension.

Cole TM, et al. Alterations of cardiovascular control of high spinal myelomalacia. *Arch Phys Med Rehabil* 1967; 48: 359-368.

Head H, Riddoch G. The automatic bladder, excessive sweating and some other reflex conditions in gross injuries of the spinal cord. *Brain* 1917; 40:188-263.

Heerfordt Syndrome (Uveoparotid Fever; Uveoparotitis; Uveoparotitic Paralysis) 562

General: Occurs in young adults, more frequently in females than in males; usual cause is sarcoidosis.

Ocular: Band keratopathy; keratoconjunctivitis sicca; uveitis; optic atrophy; papilledema; episcleritis; snowball opacity of vitreous; retinal vasculitis; proptosis; cataract; paralysis of seventh nerve; sarcoid nodules of eyelid, iris, ciliary body, choroid, and sclera; dacryoadenitis.

Clinical: Parotid gland swelling; facial paralysis; lymphadenopathy; splenomegaly; cutaneous nodules; facial nerve palsy.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Scadding JG. *Sarcoidosis.* London, Eyre & Spottiswoode, 1967.

Smellie H, Hoyle C. The natural history of pulmonary sarcoidosis. *Q J Med* 1960; 29:539-558.

Sutula FC. Tumors of the lacrimal gland and sac. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. I.* Philadelphia: WB Saunders. 1994.

Hemangioma 563

General: Can occur throughout the body, but particularly in the head; primary intraosseous orbital hemangiomas is rare; capillary hemangioma of the orbit and eyelids generally is unilateral.

Ocular: Hemangiomas of lids or orbit; ptosis; strabismus; amblyopia; proptosis; optic atrophy; hypermetropia; cavernous hemangiomas are the most common benign orbital tumors of adults.

Clinical: Ipsilateral hemangiomas of the brain and meninges.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Shields JA, et al. Classification and incidence of space occupying lesions of the orbit: a survey of 645 cases. Arch Ophthalmol 1984; 102:1606.

Hemeralopia 564

General: Autosomal dominant.

Ocular: Complete loss of the outer quadrant of visual field bilaterally; visual fields become progressively more constricted until blindness occurs; corneal ulcers; photoreceptor dysfunction.

Clinical: None.

Gehrs K, Tiedeman J. Hemeralopia in an older adult. *Surv Ophthalmol* 1992; 37:185-189.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Roy FH. *Ocular Differential Diagnosis*, 7th ed. Philadelphia: Lea & Febiger, 2003.

Hemifacial Hyperplasia with Strabismus (Bencze Syndrome) 565

General: Autosomal dominant; abnormal growth of facial skeleton and soft tissue and viscera; left side prominent.

Ocular: Strabismus; amblyopia.

Clinical: Facial asymmetry; submucous cleft palate.

Bencze J, et al. Dominant inheritance of hemifacial hyperplasia associated with strabismus. *Oral Surg* 1973; 35: 489-500.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Hemifacial Microsomia Syndrome (Unilateral Facial Agenesis; Otomandibular Dysostosis; Francois-Haustrate Syndrome) 566

General: No inheritance pattern; left side of face seems to be more frequently involved; facial asymmetry usually most obvious finding; both sexes affected; alteration of intrauterine environment is possible cause.

Ocular: Microphthalmos; congenital cystic ophthalmia; enophthalmos; strabismus; cataract; colobomata of iris, choroid, and retina.

Clinical: Microtia; macrostomia; failure of development of mandibular ramus and condyle; external auditory meatus may be absent; single or numerous ear tags; hypoplasia of facial muscles unilaterally; pulmonary agenesis (ipsilateral side); associated with Goldenhar syndrome.

Francois J, Haustrate L. Anomalies Colobomateuses du Globe Oculaire et Syndrome du Premier arc. *Ann Ocul* 1954; 187:340.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kobrynski L, et al. Trisomy 22 and facioauriculovertebral (Goldenhar) sequence. *Am J Med Genet* 1993; 46:68-71.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Hemimacrosomia Syndrome (Steiner Syndrome; Hemigigantism; Hemifacial or Unilateral Hypertrophy) 567

General: Occasionally hereditary, although true etiology is obscure; right side affected more frequently than left side; slight male preponderance.

Ocular: Dilated pupil on the affected side; eccentric pupillary location; hypochromic heterochromia.

Clinical: Unilateral facial enlargement may be associated with enlargement of half of entire body to varied extent and degree; thickened skin over involved area with increased activity of sebaceous and sweat glands; telangiectasias and multiple nevi; polydactyly; syndactyly; macrodactyly; scoliosis.

Curtius F. Kongenitaler Partieller Riesenwuchs mit Endokrinen Storungen. *Dtsch Arch Klin Med* 1925; 147:310.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Stafne EC, Lovestedt SA. Congenital hemihypertrophy of the face (facial gigantism). *Oral Surg* 1962; 15:184.

Hemochromatosis 568

General: Iron metabolism disorder; genetically determined, but mode of inheritance unknown; male preponderance 10:1; inheritance is autosomal recessive.

Ocular: Eyelid hyperpigmentation; diabetic retinopathy.

Clinical: Hemosiderin pigment deposition in many tissues; diabetes mellitus; cutaneous hyperpigmentation; cirrhosis of the liver; hypermelanotic pigmentation of skin; heart failure.

Boreck IB, et al. Combined segregation and linkage analysis of genetic hemochromatosis using affection status, serum iron and HLA. *Am J Hum Genet* 1990; 47Z:542-550.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Davies G, et al. Deposition of melanin and iron in ocular structures in haemochromatosis. *Dr J Ophthalmol* 1872; 56: 338.

Hemolytic Anemia of Newborns (Icterus Gravis Neonatorum; Erythroblastosis Fetalis) 569

General: Rh-positive/negative infant carried by an Rh-positive/negative mother; isoimmunization of the mother by her fetus of different blood group.

Ocular: Retinal hemorrhages; ophthalmoplegia; optic atrophy; yellow conjunctiva and lids.

Clinical: Jaundice; edema; liver and spleen palpable; cutaneous purpura; bleeding from mucosa.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Dacie JV. *The hemolytic anemias.* 2nd ed. New York: Grune & Stratton. 1967.

Hennebert Syndrome (Luetic-Otitic-Nystagmus Syndrome) 570

General: Caused by congenital syphilis; manifestations in childhood; when a fistula in the labyrinth exists, compression of the external auditory meatus will produce nystagmus of a wide amplitude (diagnostic of fistula).

Ocular: Spontaneous nystagmus when the column of air in the auditory canal is compressed; interstitial keratitis; disseminated syphilitic chorioretinitis may be present.

Clinical: Vertigo; fistula in the labyrinth; deafness; other clinical manifestations of congenital syphilis may be present, such as "saddle" nose and Hutchinson teeth.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hennebert C. Reactions Vestibulaires dans les Labyrinthites Heredo-Syphilitiques. *Arch Int Laryngol Otol* 1909; 28:93.

Rucker CW. Nystagmus. *Am J Ophthalmol* 1953; 36:250.

Henoch-Schönlein Purpura (Purpura; Anaphylactoid Purpura) 571

General: Occurs chiefly in children, although it can affect persons of any age; frequently follows an upper respiratory tract infection within 3 weeks.

Ocular: Retinal hemorrhages; iritis; optic neuritis.

Clinical: Purpuric skin rash; concentrated on lower extremities; joint pain; abdominal pain; hematuria; central nervous system involvement.

Harley RD, ed. *Pediatric Ophthalmology*. 4th ed. Philadelphia: WB Saunders, 1998.

Lorentz WB Jr, Weaver RG. Eye involvement in anaphylactoid purpura. *Am J Dis Child* 1980; 134:524-525.

Ryder HG, Marcus O. Henoch-Schonlein purpura: a case report. S *Afr Med J* 1976; 50:2005-2006.

Hepatic Failure 572

General: Liver failure from infections or from toxic or inflammatory causes.

Ocular: Visual field defects; scleral icterus; night blindness; abnormal color vision; eyelid retraction; lid lag; Kayser-Fleischer ring; yellow discoloration of the conjunctiva.

Clinical: Bilirubin accumulation; reduced vitamin A levels.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Lipman RM, Deutsch TA. A yellow-green posterior limbal ring in a patient who does not have Wilson's disease. Arch Ophthalmol 1990; 108:1385.

Rustgi AK, Marcus DM. Gastrointestinal and nutritional disorders. In: Albert DM, Jakobiec FA, eds. Principles *and Practice* of *Ophthalmology*, vol. V. Philadelphia: WB Saunders, 1994.

Summerskill W, Molnar G. Eye signs in hepatic cirrhosis. N Engl J Med 1962; 266:1244.

Hereditary Ectodermal Dysplasia Syndrome (Siemens Syndrome; Keratosis Follicularis Spinulosa Syndrome; Hypohidrotic Ectodermal Dysplasia; Christ-Siemens-Touraine Syndrome; Weech Syndrome; Anhidrotic Ectodermal Dysplasia; Ichthyosis Follicularis) 573

General: Autosomal recessive inheritance; strong male preponderance (about 95%); linked to X-chromosome.

Ocular: Complete loss of eyebrows (madarosis); follicular keratosis; blepharitis; entropion or ectropion; reduced tear formation or epiphora; myopia; keratoconjunctivitis; corneal erosions and ulcers (recurrent); corneal dystrophy; cataract; increased periorbital pigmentation; mongoloid lid slant; photophobia; absence of iris; luxation of lens; papillary abnormalities; cataracts.

Clinical: Mental retardation; dry skin and anhidrosis (reduced number of sweat glands); hypotrichosis; follicular hyperkeratosis (neck, palms, soles); hypohidrosis.

Ekins MB, Waring GO III. Absent meibomian glands and reduced corneal sensation in hypohidrotic ectodermal dysplasia. *J Pediatr Ophthalmol Strabismus* 1981; 18:44-47.

Freire MN, et al. A syndrome of hypohidrotic ectodermal dysplasia with normal teeth, peculiar facies, pigmentary disturbances, psychomotor and growth retardation, bilateral nuclear cataract, and other signs. *J Med Genet* 1975; 12:308-310.

Hazen PG, et al. Premature cataracts in a family with anhidrotic ectodermal dysplasia. *Arch Dermatol* 1980; 116: 1385-1387.

Liakos GM. Anhidrotic ectodermal dysplasia with lacrimal anomalies. *Br J Ophthalmol* 1979; 63:520-522.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Oosterwijk JC, et al. Linkage analysis and regional assignment of keratosis follicularis spinulosa decalvans to Xp22.2-p21.2. *Cytogenet Cell Genet* 1991; 58:2078(abstr).

Hereditary Microcornea, Glaucoma, and Absent Frontal Sinuses 574

General: Autosomal dominant.

Ocular: Microcornea; glaucoma; epicanthal folds; optic cupping.

Clinical: Thickened palmar skin; torus palatinus; frontal sinus hypoplasia.

Holmes LB, Walton DS. Hereditary microcornea, glaucoma, and absent frontal sinuses; a family study. *J Pediatr* 1969; 74:968-971.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Hermansky-Pudlak Syndrome (Oculocutaneous Albinism and Hemorrhagic Diathesis) 575

General: Autosomal recessive; mostly affects children or young adults; tyrosinase-positive oculocutaneous albinism; abnormal platelets; increased ceroid-like material in the reticuloendothelial system.

Ocular: Translucent irides; minimally pigmented ocular fundi; nystagmus; foveal hypoplasia; large refractive errors; strabismus; fundus hypopigmentation.

Clinical: Gingival bleeding; epistaxis; easy bruisability; pale skin; interstitial lung disease; defect in platelet function; "imperfect" oculocutaneous albinism; restrictive lung disease; ulcerative colitis.

Hermansky F, Pudlak P. Albinism associated with hemorrhagic diathesis and unusual pigmented reticular cells in the bone marrow. Report of two cases with histochemical studies. *Blood* 1959; 14:162.

Kotzot D, et al. Oculocutaneous albinism, immunodeficiency, hematological disorders, and minor abnormalities: a new autosomal recessive syndrome? *Am J Med Genet* 1994; 50:224-227.

Simon JW, et al. Ophthalmic manifestations of the Hermansky-Pudlak syndrome (oculocutaneous albinism and hemorrhagic diathesis). *Am J Ophthalmol* 1982; 93:71-77.

Summers CG, et al. Hermansky-Pudlak syndrome. *Ophthalmology* 1988; 95:545-554.

Suzuki T, et al. A girl with Hermansky-Pudlak syndrome. *Acta Ophthalmol* 1991; 69:256-260.

Hermit Syndrome 576

General: Seen in alcoholic men in their 60s and 70s living in relative social isolation; secondary to history of sun exposure or chronic irritation; high prevalence in the tropics or developing countries; masquerades as conjunctivitis or orbital cellulitis; develops from carcinoma *in situ* (see Bowen Disease).

Ocular: Squamous cell carcinoma of conjunctiva; proptosis; erythema of eyelids; edema of eyelids; conjunctival hyperemia; vitreous hemorrhage; ciliary body hemorrhage; choroidal hemorrhage; paresis of extraocular muscles; ocular pain; orbital cellulitis.

Clinical: Vitamin A deficiency; nutritional disorders; recurrent infections; actinic keratoses.

Gamel JW, et al. Mucoepidermoid carcinoma of the conjunctiva. *Arch Ophthalmol* 1984; 102:730-734.

Li WW, et al. Intraocular invasion by papillary squamous cell carcinoma of the conjunctiva. *Am J Ophthalmol* 1980; 90:697-701.

Rootman J, et al. Extensive squamous cell carcinoma of the conjunctiva presenting as orbital cellulitis: the hermit syndrome. *Can J Ophthalmol* 1987; 22:40-44.

Herpes Simplex 577

General: Large, complex deoxyribonucleic acid (DNA) virus.

Ocular: Conjunctivitis; keratitis; iridocyclitis; corneal ulcer; uveitis; hyphema; hypopyon; iris atrophy; cataract; scleritis; dacryoadenitis; blepharitis; acute retinal necrosis.

Clinical: Recurrent skin vesicles on lids, perioral area, nose, and genitalia; meningitis, encephalitis.

Bodaghi B, Rozenberg F, Cassoux N,et al. Nonnecrotizing herpetic retinopathies masquerading as severe posterior uveitis. *Ophthalmology* 2003; 110: 1737-1743.

Duke-Elder S, ed. *System of Ophthalmology, vol. XV.* St. Louis: CV Mosby, 1976:68-69.

Falcon MG, et al. Management of herpetic eye disease. *Trans Ophthalmol Soc U K* 1977; 97:345-349.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Whitley RJ, Schlitt M. Encephalitis caused by herpes viruses, including B virus. In: Scheld WM, Whitley RJ, Durack DT, eds. *Infections of the central nervous system.* New York: Raven Press, 1991:41-86.

Herpes Simplex Masquerade Syndrome 578

General: Acanthamoeba keratitis occurs in those who wear soft contact lenses daily; confused with herpes simplex; *Acanthamoeba culbertsoni. Acanthamoeba castellanii,* and *Acanthamoeba polyphaga* are causative agents; agents found in distilled water, hot tubs, and swimming pools (see Acanthamoeba).

Ocular: Keratitis; corneal ulcer; corneal cysts; stromal infiltrates and necrosis; scleritis; uveitis; epiphora; pseudodendrites.

Clinical: None.

Hoft RH, Mondino BJ. The diagnosis and *Clinical* management of acanthamoeba keratitis. *Semin Ophthalmol* 6:106, 1991.

John KJ, et al. Herpes simplex masquerade syndrome: acanthamoeba keratitis. *Curr Eye Res* 1987; 6:207-212.

Moore MB, et al. Acanthamoeba keratitis associated with soft contact lenses. *Am J Ophthalmol* 1985; 100:396-403.

Samples JR, et al. Acanthamoeba keratitis possibly acquired from a hot tub. *Arch Ophthalmol* 1984; 102:707-710.

Wilhelmus KR. Parasitic keratitis and conjunctivitis. In: Smolin G, Thoft RA, eds. *The Cornea,* 3rd ed. Boston: Little, Brown and Company, 1994:262-266.

Herpes Zoster 579

General: Caused by varicella zoster virus; about 75% of cases occur in persons over age 45 years; condition is more frequent with advancing age and in patients who are immunocompromised by drugs or disease; in particular, an increasing number of patients with herpes zoster ophthalmicus are immunosuppressed.

Ocular: Conjunctivitis; keratitis; recurrent corneal ulcer; neuralgia; zoster rash of eyelids; uveitis; iris atrophy; scleritis; cataract; optic neuritis; paralysis of third nerve; proptosis; paralysis of lids; orbital apex syndrome; retinitis; neurotrophic keratitis; acute retinal necrosis; progressive outer retinal necrosis; ocular motor nerve pareses; tonic pupil; encephalitis; vasculitis.

Clinical: Local lesions involving the posterior or root ganglia; nerve damage; tissue scaring.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kattah JC, Kennerdell JS. orbital apex syndrome secondary to herpes zoster ophthalmicus. *Am J Ophthalmol* 1978; 85:378-382.

Marsh RJ. Idoxuridine (IDU) in dimethyl sulfoxide (DMSO) in the treatment of ophthalmic zoster. *Ophthalmol Digest* 1977; 39:17-19.

Partamian LG, et al. Herpes simplex type 1 retinitis in an adult with systemic herpes zoster. *Am J Ophthalmol* 1981; 92:215-220.

Herrick Syndrome (Dresbach Syndrome; Sickle Cell Disease; Drepanocytic Anemia) 580

General: Usually occurs in members of the black race; poor prognosis.

Ocular: Secondary glaucoma; telangiectasis of conjunctival vessels; scleral icterus; vitreous hemorrhages; cataract; retinal hemorrhages, exudates, and neovascularization; retinitis proliferans; microaneurysms; thrombosis of retinal venules; retinal vascular sheathing; central vein occlusion; angioid streaks; retinopathy with "black sunburst sign" in patients with SS hemoglobin; "sea fan sign" in patients with SC hemoglobin; comma signs of conjunctiva; fan-shaped neovascularization of iris; sector ischemic atrophy of iris; optic atrophy; white cotton mass of vitreous; retinal holes; color vision defects; central retinal artery obstruction; branch retinal artery obstruction; white without pressure; venous tortuosity; sickling maculopathy.

Clinical: Severe anemia with hemolytic crises; bone and joint aches; hemarthrosis; jaundice; hepatosplenomegaly.

Asdourian GK. Sickle cell retinopathy. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Goldberg MF. The diagnosis and treatment of secondary glaucoma after hyphema in sickle cell patients. *Am J Ophthalmol* 1979; 87:43-49.

Majekodunmi SI, Akinyanju OO. Ocular findings in homozygous sickle cell disease in Nigeria. *Can J Ophthalmol* 1978; 13:160-162.

Nagpal KC, et al. Ocular manifestations of sickle hemoglobinopathies. *Surv Ophthalmol* 1977; 21:391-411.

Roy MS, et al. Color vision defects in sickle cell anemia. *Arch Ophthalmol* 1987; 105:1676-1678.

Vogel M. Sickle-cell disease: a clinicopathologic case report. *Ophthalmologica* 1977; 175:171-174, 1977.

Westrich DJ, Feman SS. Macular arteriolar occlusions in sickle cell beta-thalassemia. *Am J Ophthalmol* 1986; 101: 739-740.

Heterochromia Iridis 581

General: Autosomal dominant; can be associated with Horner, Waardenburg, and Marfan syndromes; may occur as an isolated phenomenon.

Ocular: Different pigmentation in the two irides or in the sectors of one iris (heterochromia iridium).

Clinical: None.

Gladstone RM. Development and significance of heterochromia of iris. *Arch Neurol* 1969; 21:184-192.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

HIE Syndrome (Hyperimmunoglobulinemia E Syndrome) 582

General: Mononuclear cells inhibit normal neutrophil and monocyte chemotaxis; caused by *Staphylococcus aureus* and *Candida trachomatis;* autosomal dominant phenotype; onset at age 1 to 8 weeks; marked elevation of immunoglobulin E.

Ocular: Atopic keratitis; keratoconjunctivitis; photophobia; corneal ulcer; chorioretinal scars; lid edema.

Clinical: Pruritic dermatitis; dry skin; skin abscesses; pneumonia; upper respiratory infections; reduced resistance; coarse facial features.

Aswad MI, et al. Plasmapheresis treatment in patients with severe atopic keratoconjunctivitis. *Ophthalmology* 1988; 95:444-447.

Butrus SI, et al. Vernal conjunctivitis in hyperimmunoglobulinemia E syndrome. *Ophthalmology* 1984; 91: 1213-1216.

Leung DYM, Geha RS. Clinical and immunological aspects of the HIE syndrome. *Hematol Oncol Clin North Am* 1988; 2:81-100.

Hilding Syndrome (Destructive Iridocyclitis and Multiple Joint Dislocations) 583

General: Atrophy of body cartilages and joint dislocations without destruction of bone or joint surfaces.

Ocular: Severe ocular hypotony; severe plastic iridocyclitis; iris atrophy; corneal endothelial precipitates; mature cataracts; retrolental inflammatory membrane.

Clinical: Multiple joint dislocations; hyperlaxity of joint capsules; generalized cartilage destruction with nose and ear deformities.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hilding AC. Syndrome of cartilage pathology, destructive iridocyclitis, multiple joint dislocation: comparison with concurrent eye and joint diseases described in literature. *Arch Ophthalmol* 1952; 48:420.

Histidinemia (Hyperhistidinemia; Histidase Deficiency) 584

General: Autosomal recessive; abnormality of amino acid metabolism due to lack of enzyme histidine ammonia lyase; the histidine gene has been assigned to 12q22-q23 by situ hybridization techniques.

Ocular: Nystagmus; hypopigmentation of the macula.

Clinical: Speech defects; mental retardation; head nodding movements; dysarthria; defective hand grip.

Dhir SP, et al. Ocular involvement in histidinaemia. *Ophthalmic Pediatr Genet* 1987; 8:175-176.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Taylor RG, et al. Localization of histidase to human chromosome region 12q22-q24.1 and mouse chromosome region 10C2-D1. *Cytogenet Cell Genet* 1991; 56:178-181.

Histiocytosis X (Hand-Schuller-Christian Syndrome; Lipoid Granuloma; Xanthomatous Granuloma Syndrome; Schuller-Christian-Hand Syndrome; Letterer-Siwe Syndrome; Acute Histiocytosis X; Eosinophilic Granuloma; Reticuloendotheliosis Syndrome) 585

General: The term histiocytosis X has been proposed to include Letterer-Siwe disease, Hand-Schuller-Christian disease, and eosinophilic granuloma of bone; there are sufficient grounds to treat Hand-Schuller-Christian and Letterer-Siwe together as different phases of the same disease process; eosinophilic granuloma most likely represents a reaction pattern, sharing some histologic features with the first two but nonetheless carrying a more benign prognosis; Letterer-Siwe disease is referred to as acute differentiated histiocytosis; Hand-Schuller-Christian disease is referred to as subacute differentiated or chronic differentiated histiocytosis; Letterer-Siwe etiology is unknown, onset is in infancy and early childhood, and prognosis is generally poor; Hand-Schuller-Christian etiology is unknown, onset is in childhood, male preponderance is 2:1, and prognosis is chronic with remissions; eosinophil may playa contributory pathophysiologic role.

Ocular: Exophthalmos; ocular pulsations; orbital roof defects; xanthelasma; blepharitis; internal ophthalmoplegia; nystagmus; retinal hemorrhages; papilledema; optic atrophy; uveitis; hypopyon; pannus; bullous keratopathy; corneal ulcer; hypochromic heterochromia; retinal detachment; cataract; scleritis.

Clinical: Hepatosplenomegaly; lymphadenopathy; skin lesions with papular eruptions; ecchymosis; purpura; bone lesions; anemia; fatigue; anorexia; fever; xanthoma of the skin; diabetes insipidus; skull defects; lung fibrosis; cardiac insufficiency.

Christian HA. Defects in membranous bones, exophthalmos and diabetes insipidus, vol. I. New York: Paul B. Hoeber, 1919:390.

Duane TD. Clinical *Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hand A. Polyuria and tuberculosis. Arch Pediatr 1893; 10:673.

Mittelman D, et al. Ocular involvement in Letterer-Siwe disease. Am J Ophthalmol 1973; 75:261.

Pearlstone AD, Flom L. Letterer-Siwe's disease. J Pediatr Ophthalmol 1970; 77: 103-105.

Petersen RA, Kuwabara T. Ocular manifestations of familial lymphohistiocytosis. Arch Ophthalmol 1968; 79:413.

Trocme SD, Aldave AJ. The eye and the eosinophil. *Surv Ophthalmol* 1994; 39:241-252.

Histoplasmosis (Histoplasmosis Choroiditis; Histoplasmosis Maculopathy; Presumed Ocular Histoplasmosis Syndrome) 586

General: Fungal infection caused by *Histoplasma capsulatum.*

Ocular: Circumpapillary atrophy; maculopathy; scattered yellow "histo" spots; optic disk edema; disseminated choroiditis (immunocompromised patients); vitreous hemorrhage; punched-out chorioretinal lesions; choroidal neovascular membrane; exogenous endophthalmitis (isolated report).

Clinical: Pulmonary infection; fever; malaise.

Aaberg TM, Meredith TA. Hemorrhagic peripapillary lesions in presumed ocular histoplasmosis. *Am J Ophthalmol* 1977; 84:160.

Beck RW, et al. Optic disc edema in the presumed ocular histoplasmosis syndrome. *Ophthalmology* 1984; 91: 183-185.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gass JDM. *Stereoscopic atlas of macular disease.* 3rd ed. St. Louis: CV Mosby, 1987.

Gutman FA. The natural course of active choroidal lesions in the presumed ocular histoplasmosis syndrome. *Trans Am Ophthalmol Soc* 1979; 77:515-541.

Halperin LS, Oik RJ. Ocular histoplasmosis syndrome. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:860.

Pulido JS, et al. Histoplasma capsulatum endophthalmitis after cataract extraction. *Ophthalmology* 1990; 97:217.

Schlaegel TF Jr. *Ocular histoplasmosis.* New York: Grune & Stratton, 1977.

Yassur Y, et al. Treatment of macular subretinal neovascularization with red-light krypton laser in presumed ocular histoplasmosis syndrome. *Am J Ophthalmol* 1981; 91:172-176.

HLA-B27 Syndromes 587

General: HLA system is the major histocompatibility complex (MHC) found on chromosome 6; associated with inflammatory disease

Ocular: Uveitis; Reiters syndrome; keratitis; band keratopathy; iris bombe'; pigment dispersion; papillary miosis; iris nodules; CME; disk edema; pars plana exudates; choroiditis

Clinical: Ankylosing spondylitis; arthritis; inflammatory bowel disease; psoriatic arthritis

Huhtinen M, Kama A: HLA-B27 typing in the categorization of uveitis in a HLA-B27 rich population. Br J Ophthalmol 2000; 84: 413-416.

Reveille JD: HLA-B27 and the seronegative spondyloarthropathies. Am J Med Sci 1998; 316: 239-249.

HMC Syndrome (Hypertelorism, Microtia, Facial Clefting Syndrome) 588

General: Developmental defect marked by an abnormally wide space between two organs or parts; autosomal recessive.

Ocular: Hypertelorism.

Clinical: Microtia; clefting of the lip, palate, and nose; psychomotor retardation; conductive hearing loss; mild micrognathia; microcephaly; thenar hypoplasia; ectopic kidneys; atretic auditory canals; congenital heart malformations.

Baraitser M. The hypertelorism microtia clefting syndrome. *J Med Genet* 1982:19:387-388.

Bixler D, et al. Hypertelorism, microtia, facial clefting: a new inherited syndrome. *Birth Defects* 1969; 2:77-81.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Hodgkin Disease 589

General: Hodgkin disease begins in the lymph nodes and usually spreads in a predictable fashion along contiguous chains of nodes; etiology may be viral; prevalent in males.

Ocular: Keratitis; uveitis; cataract; retinal hemorrhages; vasculitis; Horner syndrome; cortical blindness; papilledema; paralysis of oculomotor nerve; episcleritis; visual field defects; infiltration of choroid, conjunctiva, lacrimal gland, and orbit; papillitis; retrobulbar neuritis; opsoclonus-myoclonus; keratitis sicca; infiltrative optic neuropathy; association with Vogt-Koyanagi-Harada syndrome; bilateral serous detachments of the macula.

Clinical: Painless cervical, axillary, or inguinal lymph node swelling; fever; weight loss; anemia; generalized pruritus.

Chuah SY, et al. Vogt-Koyanagi-Harada syndrome, a rare association of Hodgkin's disease. *Postgrad Med J* 1991; 67: 476-478.

Diddie KR, et al. Necrotizing retinitis caused by opportunistic virus infection in a patient with Hodgkin's disease. *Am J Ophthalmol* 1979; 88:668-673.

Fratkin JD, et al. Disseminated Hodgkin's disease with bilateral orbital involvement. *Arch Ophthalmol* 1978; 96: 102-104.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Jackson R, et al. Ophthalmopathy after neck irradiation therapy for Hodgkin's disease. *Cancer Treat Rep* 1979; 63: 1393-1395.

Kumar H. et al. Serous macular detachment in lymphoproliferative disorders. *Can J Ophthalmol* 1991; 26:280-282.

Lissner GS, et al. Endogenous intraocular nocardia asteroides in Hodgkin's disease. *Am J Ophthalmol* 1978; 86: 388-394.

Miller NR, Iliff WJ. Visual loss as the initial symptom in Hodgkin's disease. *Arch Ophthalmol* 1975; 93: 1158-1161.

Nagai M, et al. Hodgkin's disease and Sjogren's syndrome. *Eur J Haematol* 1993; 50:180-182.

Siatkowski RM, et al. Optic neuropathy in Hodgkin's disease. *Am J Ophthalmol* 1992; 114:625-629.

Hollenhorst Syndrome (Chorioretinal Infarction Syndrome) 590

General: Caused by inadvertent pressure on the eye by the headrest from faulty positioning of the patient's head during surgery; ocular findings were immediately discovered when patient regains consciousness after surgery.

Ocular: Slight proptosis; ecchymosis of lid; marked lid edema; dilated and fixed pupil; hazy cornea; retinal edema; serous retinal detachment; cherry-red spot of the macula; attenuations of retinal arteries; pigmentary retinopathy; optic nerve atrophy; ophthalmoplegia.

Clinical: None.

Hollenhorst RW; et al. Unilateral blindness occurring during anesthesia for neurosurgical operations. *Arch Ophthalmol* 1954; 52:819.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology*. 4th ed. Baltimore: Williams & Wilkins, 1985.

Homocystinuria Syndrome 591

General: Rare disorder of amino acid metabolism; autosomal recessive inheritance; approximately one third of patients with this disorder have normal intelligence.

Ocular: Dislocated or subluxated lenses; spherophakia; cataract; retinal detachment; optic atrophy; keratitis; ocular hypotony; iris atrophy; uveitis; situs inversus optic disk; central retinal artery occlusion; high myopia; strabismus; pupillary block glaucoma; lens zonular fibers have abnormal glycoprotein with elevated concentration of cystine; ectopia lentis (nearly constant feature).

Clinical: Mental retardation; sparseness of hair; thromboembolism; arachnodactyly; generalized osteoporosis; thrombotic lesions of arteries and veins; abnormal cystathionine synthetase.

Cogan D. Dislocated lenses and homocystinuria. *Arch Ophthalmol* 1965; 74:446.

Cross HE, Jensen AD. Ocular manifestations in the Marfan syndrome and homocystinuria. *Am J Ophthalmol* 1973; 75:405.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Streeten BW The nature of the ocular zonule. *Trans Am Ophthalmol Soc* 1982; 80:823-854.

Ramsey MS, et al. The ocular histopathology of homocystinuria. A light and electron microscopic study. *Am J Ophthalmol* 1972; 74:377.

Hoof and Mouth Disease 592

General: Viral etiology.

Ocular: Conjunctival painful red blisters.

Clinical: Mucous membranes develop painful red blisters; lymph glands swollen; preauricular lymphadenopathy.

Pau H. *Differential diagnosis of eye disease*. New York: Thieme, 1987.

Horner Syndrome (Bernard-Horner Syndrome; Cervical Sympathetic Paralysis Syndrome; Claude-Bernard-Horner Syndrome; Horner Oculopupillary Syndrome) 593

General: Paralysis of cervical sympathetic; hypothalamic lesion with first neuron involved or lesion in the pons or cervical portion of cord; syndrome present in Babinski-Nageotte, Cestan-Chenais, Dejerine-Klumpke, Pancoast, Raeder, and Wallenberg syndromes (see Babinski-Nageotte Syndrome; Cestan-Chenais Syndrome; Dejerine-Klumpke Syndrome; Pancoast Syndrome; Raeder Syndrome; Wallenberg Syndrome).

Ocular: Enophthalmos; ptosis or narrowing of palpebral fissure; ocular hypotony; miosis (degree of miosis depends on site of lesion; most pronounced when roots of cranial nerves VII and VIII and first thoracic nerve are involved); hypochromic heterochromia (children more than adults); pupil does not dilate with cocaine.

Clinical: Anhidrosis on ipsilateral side of face and neck; transitory rise in facial temperature; hemifacial atrophy; may result from a variety of conditions, including cluster headache, parasellar neoplasms or aneurysms, internal carotid dissection or occlusion, and Tolosa-Hunt syndrome.

Horner F. Ueber eine Form von Ptosis. *Klin Monatsbl Augenheilkd* 1869; 7:193.

Sears ML, et at. Homer's syndrome caused by occlusion of the vascular supply to sympathetic ganglia. *Am J Ophthalmol* 1974; 77:717.

Shaari CM, Scherl MO. Nasal obstruction and Homer's syndrome. *Otolaryngol Head Neck Surg* 1994; 111:838-840.

Thompson HS, Mensher JH. Adrenergic mydriasis in Homer's syndrome: hydroxyamphetamine test for diagnosis of postganglionic defects. *Am J Ophthalmol* 1971; 72:472.

Tomsak RL. Ophthalmologic aspects of headache. *Med Clin North Am* 1991; 75:693-706.

Trachimowicz RA, Conto JE. Evaluation of pupillary disorders. In: Roberts DK, Terry JE, eds. *Ocular diseases*, 2nd ed. Boston: Butterworth-Heinemann, 1996:204-207.

Hunt Syndrome (Ramsay-Hunt Syndrome; Geniculate Neuralgia; Herpes Zoster Auricularis) 594

General: Herpes of the geniculate ganglion; course is prolonged; characterized by severe pain that frequently precedes skin and mucosal lesions and may persist for some time after lesions have disappeared; sulfuiduronate enzyme deficiency.

Ocular: Diminished lacrimation; absence of motor corneal reflex on affected side, whereas consensual reflex of noninvolved eye remains normal.

Clinical: Herpes zoster lesions of external ear and oral mucosa; severe pain in area of external auditory meatus and pinna; diminished hearing; tinnitus; vertigo, facial palsy; diminution or total loss of superficial and deep facial reflexes; zoster lesions may involve the scalp, face, and neck; hoarseness; absence of auricular lesions has been reported; progressive dementia; extensive frontal white matter change; myoclonus; ataxia; facial paralysis; tinnitus; hearing loss; hyperacusis; vertigo; dysgeusia; seizures; cerebellar ataxia; schizophrenia-like symptoms.

Adour KK. Otological complications of herpes zoster. *Ann Neurol* 1994; 35[Suppl]:S62-S64.

Blackley B, et al. Herpes zoster auris associated with facial nerve palsy and auditory nerve symptoms: a case report with histopathological findings. *Acta Otolaryngol* 1967; 63:533.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Hori T, et al. Ramsay Hunt syndrome with mental disorder. Jpn J Psychiatry Neurol 199l; 45:873-877.

Hunt JR. On herpetic inflammations of the geniculate ganglion: a new syndrome and its complications. *J Nerv Ment Dis N Y* 1907; 34:73.

Kobayashi K, et al. Ramsay Hunt syndrome: progressive mental deterioration in association with unusual cerebral white matter change. *Clin Neuropathol* 1994; 13:88-96.

Shapiro BE, et al. Absence of auricular lesions in Ramsay Hunt syndrome. *Neurology* 1994; 44:773-774.

Hunter Syndrome (MPS II Syndrome; Mucopolysaccharidosis II; Systemic Mucopolysaccharidosis Type II) 595

General: Sex-linked recessive inheritance; clinically less severe than Hurler syndrome (MPS I) with a longer life span (into adulthood); similar to MPS I (Hurler syndrome), with chondroitin sulfate B and heparitin sulfate excreted in excess in the urine (see Sanfilippo-Good Syndrome; Morquio-Brailsford Syndrome; Scheie Syndrome; Maroteaux-Lamy Syndrome); X-linked recessive inheritance; decreased iduronate sulfatase.

Ocular: Visual fields may be constricted; splitting or absence of Bowman membrane in the periphery; stromal haze may be present; pigmentary degeneration of the retina; night blindness; narrowed retinal vessels and central choroidal sclerosis; bushy eyebrows; coarse eyelashes; ptosis; optic atrophy; papilledema; proptosis; angle-closure glaucoma; corneal clouding; scleral thickening; uveal effusion.

Clinical: Dwarfism; stiff joints; hepatosplenomegaly; gargoyle-like facies.

Abraham FA, et al. Electrophysiological and psychological findings in hunter syndrome. *Arch Ophthalmol* 1974; 91:181.

Brockhurst RJ. Uveal effusion. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. 1*. Philadelphia: WB Saunders, 1994:548.

Frangiegh GT, Traboulsi EI, Kenyon KR. Mucopolysaccharidoses. In: Gold DH, Weingest TA, eds. *The Eye in Systemic Disease*. Philadelphia: JB Lippincott, 1990:372 376.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Karpati G, et al. Multiple peripheral nerve entrapments: an unusual phenotypical variant of the Hunter syndrome (mucopolysaccharidosis II) in a family. *Arch Neurol* 1974; 31:418-422.

Kenyon KR. The systemic mucopolysaccharidoses. Ultrastructural and histochemical studies of conjunctiva and skin. *Am J Ophthalmol* 1972; 73:811.

Klintworth GK, et al. Acridine orange particles in cultured fibroblasts, a comparative study of macular corneal dystrophy, systemic mucopolysaccharidoses types I-H and II, and normal controls. *Arch Pathol Lab Med* 1979; 103: 297-299.

Hurler Syndrome (Pfaundler-Hurler Syndrome; Gargoylism; Dysostosis Multiplex; MPS IH Syndrome; Systemic Mucopolysaccharidosis Type IH; Mucopolysaccharidosis IH) 596

General: Autosomal recessive inheritance; in addition to corneal opacities and enlargement of the head at birth, other symptoms become apparent at the end of the first year; death occurs usually before age 20 years; gross excess of chondroitin sulfate Band heparitin sulfate in the urine (see Hunter Syndrome; Sanfilippo-Good Syndrome; Morquio-Brailsford Syndrome; Scheie Syndrome; Maroteaux-Lamy Syndrome). Jensen suggested that the pathogenesis of the various mucopolysaccharidoses is the same but that the variations in the defective enzymes cause the different types; most common mucopolysaccharidosis, decreased α-iduronidase.

Ocular: Proptosis; hypertelorism; thick, enlarged lids; esotropia; diffuse haziness of the cornea at birth progressive to milky opacity; retinal pigmentary changes may exist; macular edema and absence of foveal reflex; optic atrophy; megalocornea; bushy eyebrows; coarse eyelashes; mucopolysaccharide deposits of iris, lens, and sclera; enlarged optic foramen; retinal detachment; anisocoria; buphthalmos; nystagmus; secondary open-angle glaucoma; progressive retinopathy with vascular narrowing; hyperpigmentation of the fundus; bone spicule; papilledema.

Clinical: Dorsolumbar kyphosis; head deformities with depressed nose bridge; short cervical spine; short limbs; macroglossia; enlarged liver and spleen; short stature; facial dysmorphism; progressive psychomotor retardation.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:249.

Collius MLX, et al. Optic nerve swelling and optic atrophy in the systemic mucopolysaccharidoses. *Ophthalmology* 1990; 97:1445.

Frangiegh GT, Traboulsi EI, Kenyon KR. Mucopolysaccharidoses. In: Gold DH, Weingest TA, eds. *The Eye in Systemic Disease*. Philadelphia: JB Lippincott, 1990:372-376.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gills JP, et al. Electroretinography and fundus oculi findings in Hurler's disease and allied mucopolysaccharidoses. *Arch Ophthalmol* 1965; 74:596.

Mailer C. Gargoylism associated with optic atrophy. *Can J Ophthalmol* 1969; 4:266-271.

Hurler-Scheie Syndrome (MPS I H/S) 597

General: Clinical disorder with severity midway between the Hurler and Scheie syndromes; genetic compound of the two alleles Hand S; autosomal recessive.

Ocular: Corneal clouding; chronically elevated optic disk; diminished or extinguished electroretinogram; pigmentary retinopathy; glaucoma; optic atrophy; retinal pigmentary degeneration.

Clinical: Shares some clinical features of both MPS IH (Hurler) and MPS IS (Scheie). Severe bone involvement; minor intellectual impairment (see Hurler Syndrome; Scheie Syndrome); cardiac failure; cardiomyopathy; conduction defects; valvular heart disease.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Frangiegh GT, Traboulsi EI, Kenyon KR. Mucopolysaccharidoses. In: Gold DH, Weingest TA, eds. *The Eye in Systemic Disease*. Philadelphia: JB Lippincott 1990:372-376.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Hutchinson-Gilford Syndrome (progeria) 598

General: Inheritance unknown; belongs to group of ectodermal dysplasias (see Werner Syndrome-Progeria of Adults); elevated hyaluronic acid of unknown etiology, likely sporadic dominant mutation.

Ocular: Microphthalmia; hypotrichosis; microcornea; cataract.

Clinical: Appearance of "old age" in children; short stature to dwarfism; dyscephaly; atrophy of skin and subcutaneous adipose tissue; aplasia of maxilla; oligodontia; arteriosclerosis (premature); progeria.

Brown WT. Progeria: a human-disease model of accelerated aging. *Am J Clin Nutr* 1992; 55[6 Suppl):1222S-1224S.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gilford H. On a condition of mixed premature and immature development. *Trans Med Chir Soc Edinb* 1897; 80:17.

Hutchinson J. Congenital absence of hair and mammary glands with atrophic condition of the skin and its appendages in a boy whose mother had been almost wholly bald from alopecia areata from the age of six. *Trans Med Chir Soc Edinb* 1886; 69:473.

Sweeney KJ, Weiss AS. Hyaluronic acid in progeria and the aged phenotype? *Gerontology* 1992; 38:139-152.

Hutchinson Syndrome (Adrenal Cortex Neuroblastoma with Orbital Metastasis; Pepper Syndrome) 599

General: Metastatic infraorbital neuroblastoma after hematogenous dissemination of primary tumor; occurs in infants and children up to age 6 years; poor prognosis; in children neuroblastoma commonly involves the orbit; 15% of patients with neuroblastoma had proptosis and ecchymosis.

Ocular: Exophthalmos; lid hematoma; extraocular muscle palsy; subconjunctival hemorrhages; choroidal metastatic tumor; papilledema; optic atrophy.

Clinical: Severe anemia; increased sedimentation rate; urinary excretion of 3-methoxy-4-hydroxy mandelic acid.

Cox RA. Proptosis due to neuroblastoma of the adrenal cortex (Hutchinson's syndrome). *Arch Ophthalmol* 1948; 39: 713.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hutchinson R. Suprarenal sarcoma in children with metastasis in the skull. *Q J Med* 1907-1908; 1:33.

Musarella MA, et al. Ocular involvement in neuroblastoma: prognostic implications. *Ophthalmology* 1984; 91:936.

Volpe NJ, Albert DM. Metastases to the uvea. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology. vol. V.* Philadelphia: WB Saunders, 1994:3260.

Hydatid Cyst (Echinococcosis) 600

General: Caused by *Echinococcus granulosus* acquired by contact with a dog host.

Ocular: Conjunctivitis; keratitis; exophthalmos; phthisis bulbi; optic atrophy; optic neuritis; papilledema; abscesses of orbit and cornea; retinal detachment; retinal hemor-rhages; cataract; hypopyon; secondary glaucoma; hydatid cysts of the conjunctiva, eyelid, orbit, and lacrimal system; acute visual loss; vitreous mass.

Clinical: Pruritus; urticaria; pulmonary cysts; brain cysts; anaphylactic shock; death.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Ozek MM, et al. Spontaneous rupture of an intraorbital hydatid cyst. A rare cause of acute loss. *J Clin Neuro-Ophthalmol* 1993; 13:135-137.

Sinav S, et al. A primary intraocular hydatid cyst. *Acta Ophthalmologica* 1991; 69:802-804.

Talib H. Orbital hydatid disease in Iraq. *Br J Surg* 1972; 59:391-394.

Hydranencephaly 601

General: Rare; development disorder in which cerebral hemispheres are replaced by a cystic space filled with cerebrospinal fluid, covered by intact meninges; short life expectancy of weeks to months, but adulthood is possible; positive association with cocaine.

Ocular: Pupillary abnormalities; strabismus; nystagmus; ptosis; optic nerve hypoplasia; chorioretinitis; retinal vessel attenuation; incomplete anterior chamber cleavage; microphthalmia; blepharospasm.

Clinical: Irritability; convulsions; muscular rigidity; decerebrate posturing; increasing head circumference; quadriplegia; psychomotor retardation.

Dominguez R, et al. Brain and ocular abnormalities in infants with in utero exposure to cocaine and other street drugs. *Am J Dis Child* 1991; 145:688-695.

Herman DC, et al. Ophthalmic findings of hydranencephaly. *J Pediatr Ophthalmol Strabismus* 1988; 25: 106-111.

Hill K, et al. Ocular signs associated with hydranencephaly. *Am J Ophthalmol* 1961; 51:267-275.

Manschot WA. Eye findings in hydranencephaly. *Ophthalmologica* 1971; 162: 151-159.

Hydroa Vacciniforme 602

General: Sensitivity to sunlight.

Ocular: Conjunctivitis; corneal vesiculae; keratitis; cicatricial ectropion.

Clinical: Vesicular skin eruptions in areas exposed to sunlight.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Hydrophobia (Lyssa; Rabies) 603

General: Acute viral zoonosis of the central nervous system.

Ocular: Lid retraction; widening of palpebral fissure; retinal hemorrhages; mydriasis; paralysis of third, fourth, fifth, or seventh nerve; bilateral optic neuritis; branch retinal artery occlusion; vaccine-induced autoimmune demyelinative optic neuritis.

Clinical: Fever; headache; nausea; numbness; tingling; acute sensitiveness to sound and light; laryngeal and pharyngeal spasms; increased muscle tonus; convulsions; delirium; coma; death.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Corey L, Hattwick MAW Treatment of persons exposed to rabies. *JAMA* 1975; 232:272-276.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WE Saunders, 2000.

van de Geijn EJ, et al. Bilateral optic neuritis with branch retinal artery occlusion associated with vaccination. *Doc Ophthalmol* 1994; 86:403-408.

Hyperammonemia I (Carbamyl Phosphate Synthetase Deficiency; Hyperammonemia II; Ornithine Transcarbamylase Deficiency; Hyperammonemia-Hyperornithinemia-Homocitrullinuria Syndrome) 604

General: Hyperammonemias I and II are due to errors at or near the "start" of the urea cycle; in hyperammonemia I, a decrease in the activity of the enzyme carbamyl phosphate synthetase, responsible for the first step of the cycle, results in the accumulation of excess ammonia; in hyperammonemia II, the defect is in ornithine transcarbamylase; type II occurs only in infants.

Ocular: Ptosis and visual loss; retinal depigmentation and chorioretinal thinning.

Clinical: Vomiting; screaming; confusion; lethargy; ataxia; mental retardation; atrophy of cerebral cortex; decreased vibration sense; bucco-faciolingual dyspraxia; learning difficulties; widespread manifestations in the central and peripheral nervous systems.

Evans OB. Hyperammonemia. In: Bradley WG, et al. *Neurology in Clinical practice, vol. II*, 2nd ed. Boston: Butterworth- Heinemann, 1995:1514-1517.

Fenichel GM. Urea cycle disturbances. In: Fenichel GM, ed. *Clinical pediatric neurology;* 2nd ed. Philadelphia: WB Saunders, 1993:10-12.

Harley RD, ed. *Pediatric Ophthalmology*. 4th ed. Philadelphia: WB Saunders, 1998.

Levin B, et al. Hyperammonaemia: a deficiency of liver ornithine transcarbamylase. *Arch Dis Child* 1969; 44:152.

Hyperkalemic Familial Periodic Paralysis (Adynamia Episodica Hereditaria) 605

General: Recurrent paralysis of skeletal muscles; occurs by age 10 years; usually occurs during the day when patient is sitting in a chair without exercise; attacks last 30 minutes to 2 hours.

Ocular: Transient attacks of staring with lid elevation in younger children; sclera above cornea is visible in adults when they look down; lid lag present during attacks but repeated up-and-down movements of eyes help.

Clinical: Muscle weakness; difficulty swallowing and coughing; tremor; episodes of quadriparesis lasting 2 to 3 weeks; salt craving; intense thirst; stomach pain.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology*, 4th ed. Baltimore: Williams & Wilkins, 1987.

Hyperlipoproteinemia 606

General: Metabolic disorder characterized by abnormally elevated concentrations of specific lipoprotein particles in the plasma.

Ocular: Arcus; lipid keratopathy; xanthelasma; lipemia retinalis; lipemia of limbal vessels; xanthomata of choroid, conjunctiva, eyelids, iris, and retina; central retinal vein occlusion; Schnyder crystalline corneal dystrophy (association).

Clinical: Deposition of lipids at various sites throughout the body, such as skin, tendons, and vascular system.

Brownstein S, et al. Schnyder's crystalline corneal dystrophy in association with hyperlipoproteinemia: histopathological and ultrastructural findings. *Can J Ophthalmol* 1991; 26:273-279.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Levy R. Drug therapy of hyperlipoproteinemia. *JAMA* 1976; 235:2234-2236.

Margolis S. Treatment of hyperlipemia. *JAMA* 1978; 239:2696-2698.

Hyperopia, High 607

General: Defect in eyesight in which the focal point falls behind the retina, resulting in farsightedness; autosomal recessive; eye shorter than normal.

Ocular: Farsightedness.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Hyperparathyroidism 608

General: Increased secretion of parathyroid hormone. Glasslike crystals of conjunctiva; band keratopathy; optic atrophy; papilledema; vascular engorgement of retina; ptosis; scleral thinning; ectopic calcifications in the choroid and sclera; unilateral visual loss; ischemic optic neuropathy.

Clinical: Hypercalcemia; hypophosphatemia; brown tumor.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Chappel D, Farrington K. Primary hyperparathyroidism presenting as unilateral visual loss. *Postgrad Med J* 1991; 67: 469-470.

Collins JF. Handbook on *Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Golan A, et al. Band keratopathy due to hyperparathyroidism. *Ophthalmologica* 1975; 171:119-122.

Levine MR, et al. Brown tumor and secondary hyperparathyroidism. *Arch Ophthalmol* 1991; 109:847-849.

Suzuki J, et al. Bilateral metastatic sclerochoroidal calcification in a patient with hyperparathyroidism. *Ophthalmologica* 1992; 205:10-14.

Hyperpigmentation of Eyelids 609

General: Autosomal dominant; causes may include drugs, sun, or too little adrenaline.

Ocular: Unusual darkening of eyelids.

Clinical: None.

Goodman RM, Belcher RW Periorbital hyperpigmentation: an overlooked genetic disorder of pigmentation. *Arch Dermatol* 1969; 100:169-174.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Hypertension 610

General: Elevated blood pressure.

Ocular: Retinal arterial narrowing; arteriosclerosis; hemorrhages; retinal edema; cotton-wool spots; fatty exudates; optic disk edema; exudative retinal detachment; optic neuropathy; swollen optic nerve; central retinal vein occlusion; branch retinal vein occlusion; choroidal ischemia.

Clinical: Systemic hypertension; patchy loss of muscle tone in vessel walls; vascular decompensation.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. Clinical *Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Hypertrichosis (Hirsutism) 611

General: Excessive hair growth due to endocrinologic or physiologic states.

Ocular: Keratitis; excessive cilia; ectropic cilia; monilethrix; pili torti; polytrichosis; reduplication of ciliary follicles; trichomegaly; association between congenital hypertrichosis lanuginosa and congenital glaucoma.

Clinical: Excessive hair growth on any part of the body except palms and soles; eczema; psoriasis; lupus erythematosus; association with acquired immunodeficiency syndrome.

Baccard M, Morel P. Excessive growth of eyelashes in patients with acquired immunodeficiency syndrome. *Cutis* 1994; 53:83-34.

Cumming DC, et al. Treatment of hirsutism with spironolactone. *JAMA* 1982; 247:1295-1298.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Judge MR, et al. Congenital hypertrichosis lanuginosa and congenital glaucoma. *Br J Dermatol* 1991; 124:495-497.

Maguire HC. Diseases of the hair. In: Moschella SI, Pillsbury DM, Hurley HJ Jr, eds. *Dermatology*. Philadelphia: WB Saunders, 1975:1215-1221.

Hypertrichosis Cubiti (Hairy Elbow Syndrome) 612

General: Autosomal dominant; occurs in childhood and regresses spontaneously in puberty; long vellus hair on extensor surface of the distal third of the upper arms and proximal third of the forearms

Ocular : facial dysmorphias

Clinical: Mental retardation; short stature

Polizzi A, Pavone P, Ciancio E, et al. Hypertrichosis cubiti (hairy elbow syndrome): aclue to a malformation syndrome. *J Pediatr Endocrinol Metab* 2005; 10: 19-25

Visser R, Beemer Fa, Veenhoven RH, et al. Hypertrichosis cubiti: two new cases and a review of the literature. *Genet Couns* 2002; 13: 397-403.

Hypertrophic Cardiomyopathy Syndrome 613

General: Autosomal recessive; four major findings are congenital cataracts, hypertrophic cardiomyopathy, mitochondrial myopathy of voluntary muscles, and exercise-related lactic acidosis; premature death.

Ocular: Congenital cataracts; nystagmus; myopia; strabismus; myopic fundus findings.

Clinical: Hypertrophic cardiomyopathy; mitochondrial myopathy of voluntary muscles; exercise-related lactic acidosis.

Cruysberg JRM, et al. Features of a syndrome with congenital cataract and hypertrophic cardiomyopathy. *Am J Ophthalmol* 1986; 102:740-749.

Willems JMF, et al. Congenital cataract and mitochondrial myopathy of skeletal and heart muscle associated with lactic acidosis after exercise. *J Pediatr* 1975; 86:873.

Hypertrophic Neuropathy 614

General: Autosomal recessive.

Ocular: Cataract.

Clinical: Elevated spinal fluid protein; severe distal sensory and motor loss.

Gold GN, Hogenhuis LAH. Hypertrophic interstitial neuropathy and cataracts. *Neurology* 1968; 18:526-533.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Hypervitaminosis A 615

General: Excessive vitamin A ingestion.

Ocular: Papilledema; congenital cataract; congenital anophthalmos; night blindness; diplopia; exophthalmos; "hourglass" cornea and iris with reduplicated lens.

Clinical: Elevation of cerebrospinal fluid pressure; migratory polyarthritis; hepatosplenomegaly; skin changes.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Evans K, Hickey-Dwyer MU. Cleft anterior segment with maternal hypervitaminosis A. *Br J Ophthalmol* 1991; 75: 691-692.

Mikkelsen B, et al. Vitamin-A intoxication causing papilledema and Simulating acute encephalitis. *Acta Neurol Scand*. 1974; 50:642-650.

Hypervitaminosis D 616

General: Excessive vitamin D ingestion.

Ocular: Band keratopathy; epicanthal folds; osteosclerosis of orbital bones; nystagmus; papilledema; iritis; cataract; sluggish pupillary reaction.

Clinical: Hypercalcemia; calcium deposition in body tissues.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duke-Elder S, ed. *System of Ophthalmology, vol. xv.* St. Louis: CV Mosby, 1976:75.

Jacobus CH, et al. Hypervitaminosis D associated with drinking milk. *N Engl J Med* 1992; 326:1173-1177.

Hypocalcemia 617

General: Serum calcium level depressed; secondary hypocalcemia can result following foscarnet treatment for cytomegalovirus retinitis in patients with acquired immunodeficiency syndrome.

Ocular: Conjunctivitis; blepharitis; blepharospasm; madarosis; ptosis; cataract; papilledema; strabismus.

Clinical: Chronic renal failure; hypoparathyroidism; hypoproteinemia; hypomagnesemia; malabsorption; acute pancreatitis; osteoblastic metastases; rickets; osteomalacia; medullary carcinoma of the thyroid; neuromuscular abnormalities.

Fraunfelder FT, Roy FH, *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Palestine AG, et al. A randomized, controlled trial of foscarnet in the treatment of cytomegalovirus retinitis in patients with AIDS. *Ann Intern Med* 1991; 115:665-673.

Porter RH, et al. Treatment of hypoparathyroid patients with chlorthalidone. *N Engl J Med* 1978; 298:577-581.

Schneider AB, Sherwood LM. Pathogenesis and management of hypoparathyroidism and other hypocalcemic disorders. *Metabolism* 1975; 24:871-898.

Hypogonadism-Cataract Syndrome 618

General: Autosomal recessive.

Ocular: Cataracts.

Clinical: Elevated follicle-stimulating hormone levels; myotonic dystrophy; infertility.

Lubinsky MS. Cataracts and testicular failure in three brothers. *Am J Med Genet* 1983; 16:149-152.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Hypolipidemia Syndrome (Hooft Syndrome) 619

General: Autosomal recessive inheritance; disorder of tryptophan metabolism; normal glycolysis disturbed, which in turn interferes with normal fat synthesis; low levels of serum phospholipids, present from birth.

Ocular: Tapetoretinal degeneration of posterior pole with irregular, small areas of grayish-yellow discoloration scattered over posterior aspect of the retina; increased shiny reflection from the macular region, with "wet" and shiny appearance.

Clinical: Mental retardation; disturbance in normal growth; anomalies of hair, nails, and teeth; erythematous squamous skin rash (mainly face, arms, and legs).

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Francois J. Ocular manifestations in certain congenital errors of metabolism. In: *Symposium on surgical and medical management of congenital anomalies of the eye*. St. Louis: CV Mosby, 1968:171.

Hooft C, et al. Familial hypolipidaemia and retarded development without steatorrhea: another inborn error of metabolism? *Helv Pediatr Acta* 1962; 17:1.

Hypomelanosis of Ito Syndrome (Incontinentia Pigmenti Achromians; Systematized Achromic Nevus) 620

General: Probable autosomal dominant transmission; cutaneous abnormality consisting of bizarre, patterned, macular hypopigmentation over variable portions of the body with multiple associated defects in other body systems; abnormal chromosome constitutions.

Ocular: Iridal heterochromia; myopia; esotropia; microphthalmia; hypertelorism; nystagmus; strabismus; corneal opacity; choroidal atrophy; exotropia; small optic nerve; hypopigmentation of the fundus; corneal asymmetry; pannus; atrophic irides with irregular pupillary margins; cataract; retinal detachment.

Clinical: Cutaneous manifestations consisting of macular hypopigmented whorls, streaks, and patches in a bilateral or unilateral distribution affecting almost any portion of the body surface; 50% have associated noncutaneous abnormalities, including central nervous system dysfunction (seizure, delayed development) and musculoskeletal anomalies.

Ito M. Studies of melanin XI. Incontinentia pigmenti achromians: a singular case of nevus depigmentosus systematicus bilateralis. *Tohoku J Esp Med* 1952; 55[Supp1]:57.

Schwartz MF, et al. Hypomelanosis of Ito (incontinentia pigmenti achromians): a neurocutaneous syndrome. *J Pediatr* 1977; 90:23-240.

Sybert VP. Hypomelanosis of Ito: a description, not a diagnosis. *J Invest Dermatol* 1994; 103[5 Supp1]:141S-143S.

Weaver RG Jr, et al. The ocular changes of incontinentia pigmenti achromians (hypomelanosis of Ito). *J Pediatr Ophthalmol Strabismus* 1991; 28: 160-163.

Hypoparathyroidism 621

General: Deficient secretion of parathyroid hormone.

Ocular: Keratitis; blepharospasm; ptosis; cataract; madarosis; optic neuritis; papilledema; conjunctivitis; myopia; ocular colobomata.

Clinical: Decreased blood calcium; increased serum phosphate; tetany; muscle cramps; stridor; carpopedal spasms; convulsions; lethargy; personality changes; mental retardation; intracranial calcification; choreoathetosis; hemiballismus; renal agenesis.

Bajandas FJ, Smith JL. Optic neuritis in hypoparathyroidism. *Neurology* 1976; 26:451-454.

Baldellou A, et al. Congenital hypoparathyroidism, ocular colobomata, unilateral renal agenesis and dysmorphic features. *Genet Couns* 1991; 2:245-247.

Blake J. Eye signs in idiopathic hypoparathyroidism. *Trans Ophthalmol Soc UK* 1976; 46:448-451.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Goel A, et al. Hypoparathyroidism with extensive intracranial calcification: a case report. *Postgrad Med J* 1994; 70: 913-915.

Parfitt AM. Adult hypoparathyroidism: treatment with calcifediol. *Arch Intern Med* 1978; 138:874-881.

Hypophosphatasia (Phosphoethanolaminuria) 622

General: Inborn error of metabolism that entails increased urinary excretion of phosphoethanolamine and associated low alkaline phosphatase and hypercalcemia; prevalent in females; may result from absence or abnormal circulating factor regulating expression of alkaline phosphatase.

Ocular: Papilledema; optic atrophy; exophthalmos; blue sclera; conjunctival calcification; lid retraction; cataract; corneal subepithelial calcifications.

Clinical: Defect in true bone formation associated with widespread skeletal abnormalities; low serum alkaline phosphatase activity; hypercalcemia; nausea; vomiting; bowing of legs; convulsions; premature loss of teeth.

Brenner RL, et al. Eye signs of hypophosphatasia. *Arch Ophthalmol* 1969; 81:614.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*. 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Whyte MP, et al. Infantile hypophosphatasia: normalization of circulating bone alkaline phosphatase activity followed by skeletal remineralization: evidence for an intact structural gene for tissue non-specific alkaline phosphatase. *J Pediatr* 1986; 108:82-88.

Hypoproteinemia Syndrome (pluridefficiency Syndrome; Kwashiorkor Syndrome; Malnutrition Syndrome) 623

General: Manifested in children aged 4 to 5 years; widespread in underdeveloped countries; lack of adequate intake of protein; good prognosis with adequate diet therapy, otherwise fatal outcome; pseudomotor cerebri.

Ocular: Dullness of cornea and conjunctiva; thick, sticky, and foamy conjunctival excretion; corneal infiltrations and cloudiness; corneal ulcers (minimally inflammatory); keratomalacia possible in prolonged cases without therapy, eventually leading to panophthalmitis.

Clinical: Muscle atrophy; generalized edema; anorexia; vomiting; diarrhea; hepatosplenomegaly; dermatitis with desquamation; pigmentation and dyspigmentation; irritability; apathy; changes in hair color; hair becomes straight; hepatomegaly; failure to grow; weak cry.

Brown KH, et al. Xerophthalmia, protein-calorie Malnutrition, and infections in children. *J Pediatr* 1979; 95: 651-656.

Lessell S. Pediatric pseudotumor cerebri (idiopathic intracranial hypertension). *Surv Ophthalmol* 1992; 37:155-166.

Scrimshaw NS, Behar M. Malnutrition in underdeveloped countries. *N Engl J Med* 1965; 272: 137.

Williams CD. Kwashiorkor, *J Am Med Assoc* 1953; 153:1280.

Hypothalamique Carrefour Syndrome (Carrefour Hypothalamique Syndrome) 624

General: Onset is sudden with hemiplegia; etiology unknown.

Ocular: Visual loss.

Clinical: Hypertension; hemianesthesia; apraxia; astereognosia; asynergias.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Ramirez F, Arana Iniguez R. Sindrome de la Encrucijada Hipotalamica (The "Carrefour Hypothalamique" Syndrome). *Ann Fac Med (Montevideo)* 1952; 37:109.

Hypothermal Injury (Cryoinjury; Frostbite) 625

General: Loss of body heat to the point of local cold injury or freezing of tissue.

Ocular: Localized cryoinjury that can cause choroidal atrophy, retinal hemorrhages, hyperpigmentation of retina, uveitis, corneal edema, neovascularization of cornea, ectropion, lid edema, madarosis, pseudoepitheliomatous hyperplasia, iris atrophy, and paresis of extraocular muscles.

Clinical: Vesicles and blebs of affected tissue, especially ears, fingers, toes, and nose; contractures; dry gangrene of affected tissues.

Bracker MD. Environmental and thermal injury. *Clin Sports Med* 1992; 11:419-436.

Britt LD, et al. New horizons in management of hypothermia and frostbite injury. *Surg Clin North Am* 1991; 71: 345-370.

Jarrett F. Frostbite: current concepts of pathogenesis and treatment. *Rev Surg* 1974; 31:71-74.

Wingfield DL, Fraunfelder FT. Possible complications secondary to cryotherapy. *Ophthalmic Surg* 1979; 10:47-55.

Hypotrichosis with juvenile macular dystrophy syndrome (HJMD) 626

General: Rare; autosomal recessive.

Ocular: Macular dystrophy; hypotrichosis; vitreous hemorrhage in newborns

Clinical: Early hair loss

Dana MR, Werner MS, Viana MA et al. Spontaneous and traumatic vitreous hemorrhage. *Ophthalmology* 1993; 100: 1377-1383.

Leibu R, Jermans A, Hatim G, et al. Hypotrichosis with Juvenile Macular Dystrophy. *Ophthalmology* 2006; 113: 841-847

Hypovitaminosis A (Xerophthalmia) 627

General: Deficient serum levels of vitamin A; principal cause of infantile blindness in the world; due to insufficient intake of vitamin A or interference with its absorption from the intestinal tract; transport or storage in the liver; obstruction of biliary tract or pancreatic ducts.

Ocular: Bitot spot; xerosis; keratomalacia; keratitis; corneal perforation and ulcer; corneal opacity; hyperkeratosis; retinal degeneration; scotoma.

Clinical: Inadequate dietary intake or interference with absorptive storage or transport capacities, as occurs in liver disease, sprue, regional enteritis, and chronic gastroenteritis; respiratory infection; diarrhea; reduced childhood mortality.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Lee WB, Hamilton SM, Harris JP, et al. Ocular complications of hypovitaminosis A after bariatric surgery. *Ophthalmology* 2005; 112: 1031-1034.

Levy NS, Toskes PP. Fundus albipunctatus and vitamin A deficiency. *Am J Ophthalmol* 1974; 78:926.

Sommer A, Emran N. Topical retinoic acid in the treatment of corneal xerophthalmia. *Am J Ophthalmol* 1978; 86: 615-617.

Sommer A, et al. Vitamin-A-responsive panocular xerophthalmia in a healthy adult. *Arch Ophthalmol* 1978; 96: 1630-1634.

Sommer A. Vitamin A, infectious disease, and childhood mortality: a 2 solution? *J Infect Dis* 1993; 167:1003-1007.

Hysteria (Malingering; Ophthalmic Flake Syndrome) 628

General: Willful or unwillful exaggeration or simulation of symptoms of an illness without physiologic cause; frequently secondary to a state of anxiety; may be seen more in children; physical or sexual abuse may be a predisposing factor.

Ocular: Anxiety-induced angiospastic or central serous retinopathy; self-induced conjunctivitis; traumatic epithelial erosions; herpetic keratitis; angioneurotic edema; contact dermatitis; ptosis; recurrent herpetic vesicles; anisocoria; peculiar pupillary reflexes; accommodative spasm; amaurosis fugax; anxiety-induced optic neuritis; disturbance of conjugate movement; dyschromatopsia; facial tic; hypersecretion glaucoma; increased or decreased tear secretion; night blindness; nystagmus; photophobia; strabismus; visual loss; psychogenic amaurosis with headaches.

Clinical: Aphonia; deafness; paralysis of limb; hemiplegia; dissociative state; anxiety; insomnia; tachycardia; shortness of breath; fatigue; vertigo, chest pains.

Barris MC, et al. Visual impairment in hysteria. *Doc Ophthalmol* 1992; 82:369-382.

Catalano RA, et al. Functional visual loss in children. *Ophthalmology* 1986; 93:385-390.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kramer KK, et al. Ocular malingering and hysteria: diagnosis and management. *Surv Ophthalmol* 1979; 24:89-96.

Miller BW A review of practical tests for ocular malingering and hysteria. *Surv Ophthalmol* 1973; 17:241-246.

Ziegler DK, Schlemmer RB. Familial psychogenic blindness and headache: a case study. *J Clin Psychiatry* 1994; 55: 114-117.

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Impetigo 629

General: Superficial primary pyoderma caused by streptococci and *Staphylococcus aureus.*

Ocular: Pustular, crusting lesions of lids and brows; conjunctivitis; corneal ulcer; cicatricial ankyloblepharon.

Clinical: Thin-roofed vesicles that develop a thin amber crust occur on face and exposed areas of the extremities; extremely common skin infections caused by S. *aureus* in patients infected with human immunodeficiency virus (HIV).

Berger TG. Treatment of bacterial, fungal, and parasitic infections in the HIV-infected host. *Semin Dermatol* 1993; 12:296-300.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Incipient Prechiasmal Optic Nerve Compression Syndrome 630

General: Caused by an expansive prechiasmal tumor or other lesion slowly compressing the optic nerve; frequently unilateral.

Ocular: Gradually progressive dimming of vision with near-normal acuity; reduced color perception (dyschromatopsia); subtle monocular field defects (progressive); positive Marcus Gunn pupillary sign (afferent pupillary light defect); optic nerve atrophy, depending on duration of compression before removal of the lesion; central or hemicentral scotoma.

Gittinger JW Jr. Chiasmal disorders. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV*. Philadelphia: WB Saunders, 1994:2615-2629.

Hollenhrost RW, Younge BR. Ocular manifestations produced by adenomas of the pituitary gland: analysis of 1000 cases. In: *Tumors*. Amsterdam: Excerpta Medica, 1973:53.

Jane JA, McKissock W Importance of failing vision in the early diagnosis of suprasellar meningioma. *Br Med J* 1962; 2:5.

Knight CL, et al. Syndrome of incipient prechiasmal optic nerve compression. *Arch Ophthalmol* 1972; 87:1.

Potter GD, Trokel SL. Anatomic and radiographic analysis of the optic canal and orbital apex. *Arch Ophthalmol* 1970; 83:584.

Infantile Neuroaxonal Dystrophy (Seitelberger Disease [2]; Spastic Amaurotic Axonal Idiocy) 631

General: Axonal disease; occurs chiefly in female infants, less frequently in older children; etiology possibly vitamin E deficiency; autosomal recessive; selective axonal degeneration in the retina.

Ocular: Nystagmus; blindness; degeneration of optic pathways and long tracts.

Clinical: Muscular hypotonia; decreased pain sense; arrest of development in late infancy; areflexia; atonic bladder; dementia; spasticity; ataxia; spread of axonal lesions in the posterior gray horns of spinal cord, restiform bodies, and tegmentum of the lower brainstem; atrophy of the cerebral cortex; degeneration of the caudate nucleus and putamen with accumulation of lipids in these nuclei and in the globus pallidus may be associated; accumulation of iron-containing pigment in the globus pallidus and putamen occurs.

Goebel HH, Lehmann J. An ultrastructural study of the retina in human late infantile neuroaxonal dystrophy. *Retina* 1993; 13:50-55.

Martin JJ, et al. Skin and conjunctival biopsies in infantile neuroaxonal dystrophy. *Acta Neuropathol* 1979; 45: 247-25l.

Seitelberger F. Eine Unbekannte Form von Infantiler Lipoid speicher Krankheit des Gehirns. *Proc 1st Int Cong Neuropathol (Turin)* 1952; 3:323-333.

Infantile Type of Neuronal Ceroid Lipofuscinosis (Ceroid Lipofuscinosis; Hagberg-Santavuori Syndrome; Haltia-Santavuori Syndrome; Santavuori-Haltia Syndrome) 632

General: Age of onset 8 to 18 months; autosomal recessive; widespread loss of photoreceptor function; cerebral deterioration; death occurs at age 5 to 9 years (see Ceroid Lipofuscinosis).

Ocular: Visual failure simultaneously with or before neurologic signs; blindness; brownish macula and other signs of macular degeneration with narrow retinal vessels; atrophic optic disk; hypopigmented dystrophic peripheral retina without pigments; nystagmus; exotropia; formed and unformed visual hallucinations.

Clinical: Psychomotor deterioration; generalized muscular hypotonia; ataxia; myoclonic jerks; "knitting" hyperkinesia; and microcephaly; convulsions rare.

Bateman JB, Philippart M. Ocular features of the Hagberg-Santavuori syndrome. *Am J Ophthalmol* 1986; 102: 262-271.

Berson EL. Retinitis pigmentosa and allied diseases. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:548.

Brod RD, et al. Diagnosis of neuronal ceroid lipofuscinosis by ultrastructural examination of peripheral blood lymphocytes. *Arch Ophthalmol* 1987; 105:1388-1393.

Francois T. Metabolic tapetoretinal degenerations. *Surv Ophthalmol* 1982; 26:293-333.

Lanska DJ, Lanska MJ. Visual "release" hallucinations in Juvenile neuronal ceroid-lipofuscinosis. *Pediatr Neurol* 1993; 9:316-317.

Infectious Mononucleosis (Mononucleosis; Epstein-Barr Virus, Acute; Acute Epstein-Barr Virus) 633

General: Asymptomatic in childhood; manifested in late adolescence of early adulthood; associated with Burkitt lymphoma and nasopharyngeal carcinoma.

Ocular: Conjunctivitis; ptosis; hippus; dacryocystitis; episcleritis; hemianopsia; nystagmus; retinal and subconjunctival hemorrhages; optic neuritis; orbital edema; scotoma; paralysis of extraocular muscles; uveitis; peripheral choroiditis; keratitis; papilledema; scleritis; retrobulbar neuritis, Sjögren syndrome; retinitis, choroiditis.

Clinical: Fever; widespread lymphadenopathy; pharyngitis; hepatic involvement; presence of atypical lymphocytes and heterophile antibodies in the blood; fatigue.

Anderson MD, et al. Retrobulbar neuritis complicating acute Epstein-Barr virus infection. *Clin Infect Dis* 1994; 18:799-80l.

Cheeseman SH. Infectious mononucleosis. In: Tierney LM, ed. *Current medical diagnosis and treatment, 33rd annual revision.* Stamford: Appleton & Lange, 1994:1103-1104.

Duke-Elder S, ed. *System of Ophthalmology. vol. VIII.* St. Louis: CV Mosby, 1976:382-385.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Pflugfelder SC, et al. Epstein-Barr virus and the lacrimal gland pathology of Sjogren's syndrome. *Adv Exp Med Biol* 1994; 350:641-646.

Wong KW; et al. Ocular involvement associated with chronic Epstein-Barr virus disease. *Arch Ophthalmol* 1987; 105:788-792.

Yoser SL, et al. Systemic viral infections and their retinal and choroidal manifestations. *Surv Ophthalmol* 1993; 37:313-352.

Influenza 634

General: Acute respiratory infection of specific viral etiology.

Ocular: Conjunctivitis; subconjunctival hemorrhages; keratitis; tenonitis; ptosis; cellulitis of orbit and lid; dacryocystitis; retinal hemorrhage; cataract; episcleritis; hypopyon; optic neuritis; uveitis; panophthalmitis; vitreal hemorrhage; paralysis of third or fourth nerve; uveitis following vaccination for influenza.

Clinical: Headache; fever; malaise; muscular aching; substernal soreness; nasal stuffiness; nausea.

Blanche P, et al. Development of uveitis following vaccination for influenza. *Clin Infect Dis* 1994; 19:979.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Cohen KL, McCarthy LR. Haemophilus influenzae ophthalmia neonatorum. *Arch Ophthalmol* 1980; 98:1214-1215.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH, eds. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Inverted Y Syndrome 635

General: Etiology unknown.

Ocular: Partial A syndrome with additional deviation occurring only in downgaze; occurs primarily with exodeviation.

Clinical: None.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Iridal Adhesion Syndrome (Iris Adhesion Syndrome, Iridocorneal Endothelial Syndrome) 636

General: Surgically related phenomenon following intraocular surgery in which iris pigment epithelium proliferates and adheres to cut edge of anterior capsule, drawing iris posteriorly to posterior and anterior capsules.

Ocular: Posterior synechiae; irregular pupil.

Clinical: None.

Dickerson D. Surgery-related iridal adhesion syndrome. *Ophthalmol Times* 1984; 9:6.

Kim DK, et al. Long-term outcome of aqueous shunt surgery in ten patients with iridocorneal endothelial syndrome. *Ophthalmology* 1999; 106:1030-1034.

Lanzl IM, et al. Outcome of trabeculectomy with mitomycin-c in the iridocorneal endothelial syndrome. *Ophthalmology* 2000; 107:295-297.

Teekhasaenee C, Ritch R. Iridocorneal endothelial syndrome in Thai patients. *Arch Ophthalmol* 2000; 118:187-192.

Iridogonodysgenesis (Iris Hypoplasia With Glaucoma) 637

General: Autosomal dominant; similar to Rieger syndrome or hereditary juvenile glaucoma.

Ocular: Stroma of iris hypoplastic; light irides; congenital glaucoma; optic atrophy; microphthalmia; opacities in Descemet membrane.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Pearce WG, et al. Autosomal dominant iridogonodysgenesis: a genetic and clinical study. *Birth Defects* 1982; 18: 561-569.

Iris Dysplasia Hypertelorism-Psychomotor Retardation Syndrome 638

General: Autosomal dominant inheritance; some features in common with Rieger syndrome.

Ocular: Hypertelorism; telecanthus; hypoplasia of the iris stroma; abnormally prominent Schwalbe line; synechiae between iris and cornea; pear-shaped pupils.

Clinical: Bilateral or unilateral hip dislocation; facial anomalies; psychomotor retardation; hypotonia and hyperlaxity of joints.

DeHauwere RC, et al. Iris dysplasia, orbital hypertelorism, and psychomotor retardation: a dominantly inherited developmental syndrome. *J Pediatr* 1973; 82:679.

Rieger H. Beitrage zur Kenntnis seltener Missbildungen der Iris. *Graefes Arch Ophthalmol* 133:602, 1935.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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von Noorden GK, Baller SR. The chamber angle in split-pupil. *Arch Ophthalmol* 1963; 70:598.

Iris Neovascularization with Pseudoexfoliation Syndrome 639

General: Anoxia secondary to iris vessel obstruction; electron microscopic studies reveal endothelial thickening with decreased lumen size and fenestration of vessel walls.

Ocular: Material found on posterior and anterior iris surface, anterior lens surface, ciliary processes, zonules, and anterior hyaloid membranes; neovascularization of iris stroma; increased permeability of iris vessels.

Henkind P. Ocular neovascularization. *Am J Ophthalmol* 1978; 85:287-301.

Ringvold A, Davanger M. Iris neovascularization in eyes with pseudoexfoliation syndrome. *Br J Ophthalmol* 1981; 65:138-141.

Vannas A. Fluorescein angiography of the vessels of the iris in pseudoexfoliation of the lens capsule, capsular glaucoma, and some other forms of glaucoma. *Acta Ophthalmol* 1969; 105[Suppl]:9.

Wand M. Neovascular glaucoma. In: Albert DM. Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. III* Philadelphia: WB Saunders, 1994:488.

Iris Pigment Layer Cleavage 640

General: Autosomal dominant; cleavage of pigment of iris and ciliary body.

Ocular: Cataracts; reduced sagittal and spherical lens diameters; glaucoma; retinal detachment; microphakia; spherophakia.

Clinical: None.

Kafer O. Dominant Vererbte Spaltung des Pigmentblattes van Iris und Ciliarkoeper mit Consekutiver Microphakie, Ectopia Lentis and Cataract. *Graefes Arch Klin Exp Ophthalmol* 1977;202:133-141.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Iris Retraction Syndrome (Posterior Synechiae and Iris Retraction Syndrome) 641

General: Rhegmatogenous retinal detachment, hypotony, and retrodisplacement of the iris with seclusion of the pupil, often associated with ciliochoroidal detachment, inflammation, and posterior vitreous retraction; caused by lowering of pressure behind iris partially due to posterior removal of fluid from subretinal space.

Ocular: Retinal detachment; hypotony; iris retraction; angle closure glaucoma; iris bombs; cataract; vitreous retraction; seclusion of the pupil; following intraocular surgery.

Clinical: None.

Campbell DG. Iris retraction associated with rhegmatogenous retinal detachment syndrome and hypotony. *Arch Ophthalmol* 1984; 102:1457-1463.

Greenfield DS, et al. Iris retraction syndrome after intraocular surgery. *Ophthalmology* 1995; 102:98-100.

Jarrett WH. Rhegmatogenous retinal detachment complicated by severe intraocular inflammation, hypotony and choroidal detachment. *Trans Am Ophthalmol Soc* 1981; 79:644-683.

Iris Nevus Syndrome (Cogan-Reese Syndrome; Chandler Syndrome; Iridocorneal Endothelial Syndrome; ICE Syndrome) 642

General: Usually unilateral but may be bilateral; usually in young adult women; nonfamilial; cause unknown; Chandler, Cogan-Reese, and iridocorneal endothelial syndromes have been considered three separate syndromes but are now recognized as a single spectrum of diseases.

Ocular: Unilateral glaucoma in eyes with peripheral anterior synechiae; multiple iris nodules; ectopic Descemet membrane; corneal edema; stromal iris atrophy; iris pigment epithelial atrophy; ectropion uveae; ectopic pupil; keratoconus; herpes simplex virus deoxyribonucleic acid (DNA) has been detected in patients with iridocorneal endothelial syndrome from corneal specimens.

Clinical: Glasslike membrane covering the anterior iris surface; corneal endothelial degeneration and accompanying ectopic endothelial membranes are responsible for occlusion of the filtration meshwork and subsequent pressure increase.

Alvarado JA, et al. Detection of herpes simplex viral DNA in the iridocorneal endothelial syndrome. *Arch Ophthalmol* 1994; 112:1601-1609.

Buckley RJ. Pathogenesis of the ICE syndrome. *Br J Ophthalmol* 1994; 78:595-596.

Chandler PA. Atrophy of the stroma of the iris, endothelial dystrophy, corneal edema, and glaucoma. *Am J Ophthalmol* 1956; 41:607.

Cogan DG, Reese AB. A syndrome of iris nodules, ectopic Descemet's membrane and unilateral glaucoma. *Doc Ophthalmol* 1969; 26:424.

Radius RL, Herschler J. Histopathology in the iris-nevus (Cogan-Reese) syndrome. *Am J Ophthalmol* 1980; 89: 780-786.

Rodrigues MM, et al. Clinical, electron microscopic, and immunohistochemical study of the corneal endothelium and Descemet's membrane in the iridocorneal endothelial syndrome. *Am J Ophthalmol* 1986; 101:16-27.

Iron Deficiency Anemia (Microcytic Hypochromic Anemia) 643

General: Conjunctival pallor; small retinal hemorrhages; diplopia; visual field defects; fixed and dilated pupil; swelling or pallor of optic disk; retrobulbar neuritis; optic atrophy; it represents a state of inadequate body stores of iron; diagnosis of iron deficiency anemia is the *sine qua non* of chronic iron loss; most usual source is the gastrointestinal tract.

Clinical: Lethargy; increased susceptibility to infections; lymphadenopathy; hepatomegaly; splenomegaly.

Bentler E. The common anemias. *JAMA* 1988; 259:2433-2437.

Cogan DG. *Neurology of the visual system*, 3rd ed. Springfield, IL: Charles C. Thomas, 1968.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Huber A. *Eye symptoms in brain tumors*, 2nd ed. St. Louis: CV Mosby, 1971.

Irvine Syndrome (Irvine-Gass Syndrome; Hruby-Irvine-Gass Syndrome) 644

General: Failing vision after uneventful cataract extraction; caused by spontaneous rupture of vitreous face with vitreous adhesions to the wound followed by macular edema; no gender or race preference; more common in older adults; positive association with other systemic vascular disease; may follow Nd:YAG laser posterior capsulotomy.

Ocular: Failing improvement or decrease in visual acuity after cataract surgery, due to macular changes; permanent impairment if the condition is not restored before secondary degenerative changes occur; vitreous opacities; rupture of the hyaloid face (spontaneous); vitreous adhesions to the wound; cystoid macular edema; papilledema; optic atrophy.

Clinical: None.

Blair NP, Kim SH. Cystoid macular edema after ocular surgery. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. III.* Philadelphia: WB Saunders, 1994:488.

Gass JDM, Norton EWD. Cystoid macular edema and papilledema following cataract extraction. *Arch Ophthalmol* 1966; 76:646.

Irvine SR. A newly defined vitreous syndrome following cataract surgery. *Am J Ophthalmol* 1953; 36:599.

Jampol LM. Cystoid macular edema following cataract surgery. *Arch Ophthalmol* 1988; 106:894.

Naumann GOH, Seibel W. Surgical revision of vitreous and ins-incarceration in persisting cystoid maculopathy (Hruby-Irvine-Gass syndrome). *Dev Ophthalmol* 1985; 11:181-187.

Ischemic Orbital Compartment Syndrome 645

General: Associated with spine surgery in the prone position

Ocular: Proptosis; elevated intraocular pressure; decreased vision; periocular pain; ischemic optic atrophy.

Clinical: Facial swelling; spinal stenosis or other diagnosis that would require long surgical procedures on the prone position

Leibovitch I, Casson R, Laforest C, et al. Ischemic orbital compartment syndrome as a complication of spinal surgery in the prone position. *Ophthalmology* 2006; 113: 105-108.

Isotretinoin Teratogen Syndrome 646

General: Maternal ingestion of isotretinoin (Accutane) during early pregnancy.

Ocular: Small palpebral fissures; deep orbits; systemic treatment with isotretinoin has been associated with blepharitis and conjunctivitis.

Clinical: Prominent forehead; low-set, small, undifferentiated ears; depressed nasal bridge; small chin.

Benke PJ. Isotretmoin teratogen syndrome. *JAMA* 1984; 24:3267-3269.

IVIC Syndrome (Radial Ray Defects, Hearing Impairment, Internal Ophthalmoplegia, Thrombocytopenia) 647

General: Autosomal dominant; Institute Venezolano de Investigacionas Cientificas (IVIC); observed in 1800s from Canary Islands in Venezuela; has been observed in descendants of a family that migrated to Venezuela from the Canary Islands.

Ocular: Strabismus; internal ophthalmoplegia.

Clinical: Malformed upper limb; short distal phalanx; hearing loss; thrombocytopenia; leukocytosis; imperforate anus; radial ray defect.

Areas S., et al. The IVIC syndrome: a new autosomal dominant complex pleiotropic syndrome with radial ray hypoplasia, hearing impairment, internal ophthalmoplegia, and thrombocytopenia. *Am J Med Genet* 1980; 6:25-29.

Czeizel A, et al. IVIC syndrome: report of a third family [Letter]. *Am J Med Genet* 1989; 33:282-283.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Jabs Syndrome (Synovitis, Granulomatous Uveitis, and Cranial Neuropathies) 648

General: Autosomal dominant.

Ocular: Granulomatous uveitis; iritis; sixth nerve palsy.

Clinical: Granulomatous synovitis; corticosteroid responsive hearing loss; boggy polysynovitis; boutonneuse deformities; granulomatous arthritis; skin involvement; fever; hypertension; large-vessel vasculitis.

Blau EB. Familial granulomatous arthritis, iritis, and rash. *J Pediatr* 1985; 107:689-693.

Jabs DA, et al. Familial granulomatous, synovitis, uveitis, and cranial neuropathies. *Am J Med* 1985; 78:801-804.

Jacobs Syndrome (Triple X Syndrome; XXX Syndrome; Super Female Syndrome) 649

General: Caused by sex chromosomal anomaly with 44 autosomal and 3X sex chromosomes; due to nondisjunction; often associated with autosomal trisomies; majority of cases are asymptomatic.

Ocular: Hypertelorism; epicanthus; mongoloid, slanted lid fissure; strabismus.

Clinical: Microcephaly; oligophrenia, occasionally with secondary amenorrhea; abnormal dentition; high-arched palate; hypogenitalism; occasionally mental retardation; early menopause.

Jacobs PA, et al. Evidence for the existence of the human "superfemale." *Lancet* 1959; 1:423.

Kohn G, et al. Trisomy X in three children. *J Pediatr* 1968; 72:248.

Jacobsen-Brodwall Syndrome 650

General: Syndrome consists of inborn defect in erythropoiesis; dysplastic kidneys; eye lesions; malformation of the teeth; impaired hearing; etiology unknown; features of Alport, Fanconi, and Lowe syndromes do not conform with the findings in this syndrome.

Ocular: Loss of vision progressing to blindness; secondary glaucoma; vitreous opacities; cataract; macular changes; retinal hemorrhages and exudates.

Clinical: Anemia; atypical development of teeth, with caries and resorption of permanent teeth; kidney dysplasia; hearing defect.

Jacobsen CD, Brodwall EK. A *Clinical* syndrome with inborn defect in erythropoiesis, dysplastic kidneys, eye lesions, malformation of the teeth and impaired hearing. *Acta Med Scand* 1974; 195:231.

Whalen RE, et al. Hereditary nephropathy, deafness and renal foam cells. *Am J Med* 1961; 31:171.

Jacod Syndrome (Negri-Jacod Syndrome; Petrosphenoidal Space Syndrome) 651

General: Lesion involving cranial nerves II to VI; most frequently a malignant nasopharyngeal tumor originating in lateropharyngeal area.

Ocular: Ophthalmoplegia; unilateral blindness; trigeminal neuralgia (ophthalmic branch); descending optic atrophy if cranial nerve II is involved; amaurosis fugax.

Clinical: Trigeminal neuralgia (at the beginning the first and second divisions of nerve V are involved; later the third division is affected as well); unilateral or bilateral enlargement of cervical lymph nodes (30%); deafness; palatal muscle paralysis.

Fischer A, et al. Familial nasopharyngeal carcinoma. *Pathology* 1984; 16:23-24.

Jacod M. Sur la Propagation Intracranienne des Sarcomes de la Trompe d'Eustache Syndrome du Carrefour Petro-Sphenoidale Paralysie des 2, 3, 4, 5, et 6 Paires Craniennes. *Rev Neurol* l921; 38:33.

Riggs HE. Cranial nerve syndromes associated with nasopharyngeal malignancy. *Arch Neurol Psychiatr* 1957; 77: 473.

Jadassohn-Lewandowsky Syndrome (Pachyonychia Congenita) 652

General: Autosomal dominant inheritance; three variants: type I has symmetric keratoses of hands and feet and follicular keratosis of body; type II same as type I, plus leukokeratosis; type III same as type I with corneal changes; gene for this disorder has been found to be closely linked to the keratin gene cluster on 17q12-q21; disorder usually develops in early infancy.

Ocular: Dyskeratosis of the cornea; bilateral cataract.

Clinical: Keratosis and hyperhidrosis of palms and soles, whereas the remaining skin is usually rather dry; bullous lesions may occur with secondary infections, mainly during warm seasons; leukokeratosis of oral mucosa (mainly tongue); follicular keratosis; congenital pachyonychia (nails not only may be thickened but also may be frequently inflamed and lost with aggravation at sites of regrowth); hoarse voice; epidermoid cysts; oral leukokeratosis.

Banscher S. The Jadassohn-Lewandowsky Syndrome. *Hautarzt* 1973; 24:439.

Neisser A, Jacobi E, eds. *Ikonographia dermatologica*. Berlin: Urban & Schwarzenberg, 1906:29.

Munro CS, et al. A gene for pachyonychia congenita is closely linked to the keratin gene cluster on 17q12-q21. *J Med Genet* 1994; 31:675-678.

Oriba HA, et al. Callused feet, thick nails and white tongue. Pachyonychia congenita. *Arch Dermatol* 1991; 127:113-114, 116-117.

Paller AS, et al. Pachyonychia congenita tarda. A late-onset form of pachyonychia congenita. *Arch Dermatol* 1991; 127:701-703.

Jansen Disease (Metaphyseal Dysostosis) 653

General: Affects both sexes; etiology unknown; autosomal dominant.

Ocular: Exophthalmos.

Clinical: Mental retardation; muscular atrophy; flat nose; large mouth; dwarfism; deafness; metaphyses of all bones affected with marked widening.

Jansen M. Ubex Atypische Condrodystrophie (Achondroplasic) und Uber Eine Noch Nicht Beschriebene Angeborene Washstumsstoring des Knochensystems Metaphysare Dysostosis. *Orthop Chir* 1934; 61:253-286.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981:434.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Japanese River Fever (Mite-Borne Typhus; Rural Typhus; Scrub Typhus; Tropical Typhus; Tsutsugamushi Disease; Typhus) 654

General: Acute febrile illness by *Rickettsia tsutsugamushi* transmitted by the larval form of a mite.

Ocular: Keratitis; uveitis; paracentral scotoma; vitreous opacity; nystagmus; retinal hemorrhages; exudates; edema.

Clinical: Chills; fever; malaise; headache; lymphadenopathy; generalized aching.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kazar J, Brezina R. Control of rickettsial diseases. *Eur J Epidemiol* 1991; 7:282-286.

Manor E, et al. Papilledema in endemic typhus. *Am J Ophthalmol* 1977; 84:559.

Sheehy TW; et al. Scrub typhus: a comparison of chloramphenicol and tetracycline in its treatment. *Arch Intern Med* 1973; 132:77-80.

Taniguchi Y, et al. Tsutsugamushi disease (scrub typhus). *Int J Dermatol* 1992; 31:693-695.

Jensen Disease (Juxtapapillary Retinopathy) 655

General: Etiology unknown.

Ocular: Circumscribed inflammatory changes of the choroid; field defect.

Clinical: None.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Jeune Disease (Asphyxiating Thoracic Dystrophy; Thoracic-Pelvic-Phalangeal Dystrophy) 656

General: Autosomal recessive; similar to Ellis-van Creveld syndrome; positive associations of this disorder with cystinuria has been reported in two sisters.

Ocular: Retinal dysfunction; granular pigmentation of the choroid; nystagmus; small white patches in peripheral fundus; retinal degeneration; coloboma of iris; eyes symmetrically involved; retinal aplasia; photophobia; strabismus; pigmentary retinopathy.

Clinical: Long, narrow thorax; short anteriorly clubbed ribs forming a continuous tube with the abdominal cavity; dwarfing skeletal dysplasia; progressive renal failure; liver abnormalities; severe respiratory insufficiency; long, narrow trunk; dystrophic rib cage with respiratory distress; short limbs; polydactyly.

Airede AK. Asphyxiating thoracic dystrophy (Jeune's disease): a case report. *East Afr Med J* 1994; 71:67-69.

Bard LA, et al. Retinal involvement in thoracic-pelvic-phalangeal dystrophy. *Arch Ophthalmol* 1978; 96:278-281.

Phillips CI, et al. Asphyxiating thoracic dystrophy (Jeune's disease) with retinal aplasia: a sibship of two. *J Pediatr Ophthalmol Strabismus* 1979; 16:279-283.

Wilson DJ, et al. Retinal dystrophy in Jeune's syndrome. *Arch Ophthalmol* 1987; 105:651-654.

Johnson Syndrome (Adherence Syndrome; Adherent Lateral Rectus Syndrome) 657

General: Congenital delayed development; most frequent in children below age 3 years; spontaneous disappearance possible; two principal types of disturbances: (i) adhesions between sheaths of external rectus and inferior oblique with resulting limits in abduction; (ii) adhesions between sheaths of superior rectus and superior oblique with resulting limits in elevation.

Ocular: Forced muscle duction test may prove presence or absence of adherence versus paralysis.

Clinical: None.

Adler FM. Some confusion factors in the diagnosis of paralysis of the vertically acting muscles. *Trans Am Acad Ophthalmol Otolaryngol* 1948; 52:447.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger. 1976.

Johnson LV. Adherence syndrome (pseudoparalysis of lateral or superior rectus muscles). *Arch Ophthalmol* 1950; 44: 870.

Jones Syndrome (Cherubism; Mandibular Cystic Dysplasia) 658

General: Etiology unknown; both sexes affected; present at birth.

Ocular: White line beneath the iris on sclera; hypertelorism; optic atrophy.

Clinical: Rounded cheeks; jaw fullness; submandibular region swelling; narrow, V-shaped palate.

Al-Gazali LI, et al. Cherubism, optic atrophy and short stature. *Clin Dysmorphol* 1993; 2:140-141.

Font RL, Blanco G, Soparkar CN, et al. Giant cell reparative granuloma of the orbit associated with cherubism. *Ophthalmology* 2003; 110: 1846-1849.

Jones WA. Familial multilocular cystic disease of the jaw. *Am J Cancer* 1933; 17:946-950.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Joubert Syndrome (Familial Cerebellar Vermis Agenesis) 659

General: Autosomal recessive; both sexes affected; onset in early infancy.

Ocular: Choroidal coloboma; nystagmus; ocular fibrosis, telecanthus.

Clinical: Episodic hyperpnea; apnea; ataxia; psychomotor retardation; rhythmic protrusion of tongue; mental retardation; micrognathia; complex cardiac malformation; cutaneous dimples over wrists and elbows.

Jacobson DM, et al. Joubert's syndrome, ocular fibrosis, and normal histidine levels. *Am J Ophthalmol* 1992; 113: 714-716.

Joubert M, et al. Familial agenesis of the cerebellar vermis. *Neurology* 1969; 19:813-825.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott. 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Squires LA, et al. Dysmorphic features of Joubert syndrome. *Dysmorphol Clin Genet* 1991; 5:72-77.

Jugular Foramen Syndrome (Vernet Syndrome) 660

General: Injuries, aneurysms, and tumors (more commonly due to metastatic lesion than primary neoplasms) affecting the foramen jugulare are the primary causes for the syndrome to develop; if sympathetic fibers surrounding the carotid artery are involved, this will produce Homer triad; note similarity of clinical findings of Villaret syndrome or "retroparotid space syndrome," which may include epiphora and lagophthalmos and in which cranial nerves IX to XII and the cervical sympathetics are involved.

Ocular: Enophthalmos; ptosis; miosis.

Clinical: Paralysis of the ninth, tenth, and eleventh cranial nerves with resulting impairment of related function, that is, dysphagia, loss of taste on the posterior third of the tongue, and nasal regurgitation; anhidrosis; paralysis of the sternocleidomastoid muscle and part of the trapezium (upper portion); hoarseness; tachycardia; dysarthria; weight loss.

Call WH, Pulec JL. Neurilemoma of the jugular foramen. Transmastoid-removal. *Ann Otol Rhinol Laryngol* 1978; 87[3 Part 1]:313-317.

George B, Lot G, Tran Ba Huy P. The juxtacondylar approach to the jugular foramen (without petrous bone drilling). *Surg Neurol* 1995; 44:279-284.

Hanson MR, Sweeney PJ. Disturbances of lower cranial nerves. In: Bradley WG, et al., eds. *Neurology in Clinical Practice,* 2nd ed. Boston: Butterworth-Heinemann, 1995:261-262.

Kamitani H, Masuzawa H, Kanazawa I, et al. A combined extradural-posterior petrous and suboccipital approach to the jugular foramen tumors. *Acta Neurochir (Wien)* 1994; 126: 179-184.

Robbins KT, Fenton RS. Jugular foramen syndrome. *J Otolaryngol* 1980; 9:505-516.

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Sawada H, et al. Accessory nerve neuroma presenting as recurrent Jugular foramen syndrome. *Neuroradiology* 1992; 34:417-419.

Schweinfurth JM, et al. Jugular foramen syndrome as a complication of metastatic melanoma. *Am J Otolaryngol* 1993; 14:168-174.

Seyfried DM, Rock JP. The transcondylar approach to the jugular foramen: a comparative anatomic study. *Surg Neurol* 1994; 42:265-271.

Vernet M. *Les Paraliesies Laryngees Associees*. Thesis, Lyon, 1916:233.

Junius-Kuhnt Syndrome (Kuhnt-Junius Disease; Macular Senile Disciform Degeneration [I]; Macula Lutea Juvenile Degeneration [2]) 661

General: Onset in advanced age or in juvenile period; etiology unknown; possible autosomal dominant or recessive inheritance.

Ocular: Impairment of central vision; central scotoma; atrophic macular degeneration surrounded by retinal hemorrhages, resulting in mountlike lesion; exudative and atrophic reaction with deposit in and about macula.

Clinical: None.

Deutman AF. Hereditary dystrophies of the central retina and choroid. In: Winkelman JE, Crone RA, eds. *Perspectives in Ophthalmology, vol.* 2. Amsterdam: Excerpta Medica, 1970.

Deutman AF. Macular dystrophies. In: Ryan SJ, ed. *Retina, vol. II.* 2nd ed. St. Louis, Mosby, 1994.

Kimura SJ, Caygill WM, eds. *Retinal diseases.* Philadelphia: Lea & Febiger, 1966.

Magalini SJ, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981:440.

Juvenile Diabetes-Dwarfism-Obesity Syndrome (Mauriac Syndrome; Dwarfism-Hepatomegaly-Obesity-Juvenile Diabetes Syndrome) 662

General: Etiology is obscure, although nutritional deficiencies, metabolic disorders, and deficiency in insulin have been considered; develops slowly, with slow growth and difficulties in management of diabetic condition.

Ocular: Cataract; diabetic retinopathy with retinal hemorrhages, exudates, microaneurysms, neovascularization, vaso and glial proliferation (grades I to IV diabetic retinopathy; hypertensive retinopathy); occasional optic neuritis.

Clinical: Hepatomegaly; diminished growth; osteoporosis; hypertension; arteriosclerosis; obesity (with moon face); juvenile diabetes; abdominal colic.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Guest GM. The Mauriac syndrome. *Diabetes* 1953; 2:415.

Mauriac P. Gros Ventre. Hepatomegalie. Troubles de la Croissance Chez les Enfants Diabetiques, Traites Depuis Plusieurs Annees par l'Insuline. *Gaz Hebd Sci Med (Bordeaux)* 1930; 51:402.

Juvenile Rheumatoid Arthritis (JRA; Still Disease) 663

General: Onset before age 16 years; greater occurrence of systemic manifestations, monarticular and oligoarticular joint involvement, and iridocyclitis.

Ocular: Hypopyon; band keratopathy; uveitis; cataract; papillitis; glaucoma; macular edema; ocular pain; vitreous cells; synechiae; scleritis; presumed to have an autoimmune etiology; antiocular antibodies, including iris protein antibodies, have been found in the sera of patients.

Clinical: Salmon pink macular rash; arthritis; hepatosplenomegaly; leukocytosis; chronic pain; joint swelling; low-grade fever; anemia; rheumatoid nodules.

Bloom JN, et al. Serum antiocular antibodies in patients with juvenile rheumatoid arthritis. *J Pediatr Ophthalmol Strabismus* 1993; 30:243-248.

Chylack LT Jr, et al. Ocular manifestations of juvenile rheumatoid arthritis. *Am J Ophthalmol* 1975; 79:1026.

Fraunfelder FT. Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Hunt DW, et al. Iris protein antibodies in serum of patients with juvenile rheumatoid arthritis and uveitis. *Int Arch Allergy Immunol* 1993; 100:314-318.

Merriam JC, et al. Early-onset pauciarticular juvenile rheumatoid arthritis. *Arch Ophthalmol* 1983; 101:1094.

Smiley WK. The eye in juvenile rheumatoid arthritis. *Trans Ophthalmol Soc UK* 1974; 94:817-829.

Juvenile Xanthogranuloma (JXG; Nevoxanthoendothelioma) 664

General: Childhood disease; unknown etiology.

Ocular: Uveal tract tumor presenting as spontaneous hyphema; secondary glaucoma; uveitis; corneal, lid, and epibulbar tumors; proptosis; retinal and choroidal lesions (rare).

Clinical: Multiple benign tumors, primarily of the skin; usually appear in the first 3 years of life; lesions appear as yellow-to-brown papules or nodules.

DeBarge LR, et al. Chorioretinal, iris, and ciliary body infiltration by juvenile xanthogranuloma masquerading as uveitis. *Surv Ophthalmol* 1994; 39:65-71.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kaufmann JG, et al. Juvenile xanthogranuloma of the corneoscleral limbus. *Ophthalmic Surg* 1993; 24:428-430.

Zimmerman LE. Ocular lesions of juvenile xanthogranuloma. Nevoxanthoendothelioma. *Am J Ophthalmol* 1965; 60: 1011-1035.

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Kabuki Makeup Syndrome (Niikawa-Kuroki Syndrome) 665

General: Etiology unknown; originally termed Kabuki makeup syndrome because dysmorphic facies resembled the stylized makeup worn by Kabuki actors; also seen in ethnic groups other than Japanese.

Ocular: Ectropion of lower eyelid; long palpebral fissures; sparse lateral half of eyebrows; highly arched eyebrows.

Clinical: Prominent ears; cleft or highly arched palates; brachydactyly; dermatoglyphics; padlike swelling of fingertips; short stature; mental retardation; susceptibility to infection; characteristic facies; developmental delay; mental and growth retardation with specific craniofacial malformations including a depressed nasal tip; musculoskeletal abnormalities, evolving phenotype over time suggesting an underlying defect of the connective tissue.

Kaiser-Kupfer MI, et al. The Niikawa-Kuroki (Kabuki make-up) syndrome in an American black. *Am J Ophthalmol* 1986; 102:667-668.

Kuroki Y, et al. A new malformation syndrome of long palpebral fissure, large ears, depressed nasal tip and skeletal anomalies associated with postnatal dwarfism and mental retardation. *J Pediatr* 1981; 99:570.

Niikawa N, et al. Kabuki make-up syndrome: a syndrome of mental retardation, unusual facies, large and protruding ears and postnatal growth deficiency. *J Pediatr* 1981; 99:565.

Shiekh TM, et al. Niikawa-Kuroki syndrome. *Pediatr Res* 1986; 20:340a.

Kahler Disease (Myelomatosis; Multiple Myeloma) 666

General: Disseminated malignancy of plasma cells located predominantly in the bone marrow.

Ocular: Tumor of orbit common, with proptosis or displacement of globe; conjunctival sledging and segmentation; crystalline deposits of cornea and conjunctiva; cotton-wool spots; retrobulbar neuritis; occlusion of central retinal artery and vein; palsy of sixth nerve; vitreous hemorrhage; dilated veins and hemorrhages; retinal microaneurysms; choroidal detachment; amaurosis fugax; myeloma infiltrates in orbit, iris, choroid, retina, sclera, and optic nerve; corneal opacities; ciliary body cysts; iritis; glaucoma; subluxation of lens; papilledema; corneal edema; cavernous sinus syndrome; bilateral superficial punctate keratitis; central retinal vein occlusion; crystalline keratopathy in a vortex distribution; spontaneous endocapsular hematoma.

Clinical: Bone pain; fractures; dehydration; hypercalcemia; hyperuricemia; proteinuria; inclination to infection; hyperviscosity.

Auran JD, et al. Multiple myeloma presenting as vortex crystalline keratopathy and complicated by endocapsular hematoma. *Cornea* 1992; 11:584-585.

Hayasaka S, et al. Central retinal vein occlusion in two patients with immunoglobulin G multiple myeloma associated with blood hyperviscosity. *Ann Ophthalmol* 1993; 25: 191-194.

Klintworth GK, et al. Analysis of corneal crystalline deposits in multiple myeloma. *Am J Ophthalmol* 1978; 86:303-313.

Lam S, et al. Cavernous sinus syndrome as the initial manifestation of multiple myeloma. *J Clin Neuro-Ophthalmol* 1987; 7:135.

Lee H, et al. Periorbital ecchymoses as the initial sign in multiple myeloma. *Ann Ophthalmol* 1982; 14:1066-1068.

Maisel JM, et al. Multiple myeloma presenting with ocular inflammation. *Ann Ophthalmol* 1987; 19:170-174.

Mewis-Levin L, et al. Plasma cell myeloma of the orbit. *Ann Ophthalmol* 1981; 13:477-481.

Perry HD, et al. Intraepithelial corneal immunoglobulin crystals in IgG-kappa multiple myeloma. *Cornea* 1993; 12:448-450.

Kallmann Syndrome (Hypogonadotropic Hypogonadism-Anosmia Syndrome) 667

General: Disorder of hypothalamic function involving the control of releasing factors, with hypogonadism and anosmia as the clinical signs; agenesis of olfactory bulbs; midline cranial anomalies (cleft lip, cleft palate, imperfect fusion); autosomal recessive phenotype.

Ocular: Color blindness (variable occurrence).

Clinical: Failure of sexual maturation; decrease in primary and secondary sex characteristics; loss of smell; hypertension; mental retardation; schizophrenia.

Cowen MA, Green M. The Kallmann's syndrome variant (KSV) model of the schizophrenias. *Schizophr Res* 1993; 9:1-10.

Kallmann RJ, et al. The genetic aspects of primary eunuchoidism. *Am J Ment Defic* 1944; 48:203.

Males JL, et al. Hypogonadotropic hypogonadism with anosmia-Kallmann's syndrome. *Arch Intern Med* 1973; 131:501.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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White BK, et al. The syndrome of anosmia with hypogonadotropic hypogonadism: a genetic study of 18 new families and a review. *Am J Med Genet* 1983; 15:417-435.

Kaposi Disease (Kaposi Sarcoma; Kaposi Hemorrhagic Sarcoma; Multiple Idiopathic Hemorrhagic Sarcoma; Kaposi Varicelliform Eruption) 668

General: Vascular tumor of unknown cause; seen most often in males, Jews, and those from eastern Europe, the southern Mediterranean, and Africa; human immunodeficiency virus-related Kaposi syndrome is the most common type of cancer seen in acquired immunodeficiency syndrome patients.

Ocular: Ocular adnexa, varicelliform eruption, including lids, conjunctivae, lacrimal glands, and orbit, may be involved; hemorrhage; extensive injection and thickening of conjunctival tissues; conjunctival involvement more evident in bulbar conjunctiva.

Clinical: Vascular sarcomas usually occur on the legs, although widespread cutaneous and visceral tumors may develop; secondary malignancies are very common; lymphedema.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson. 1982.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WE Saunders. 2000.

Kaposi M, Kohn M. Idiopathisches Multiples Pigrnentsarkonder Haul. *Arch Dermatol Syphil* 1872; 4:265-273.

Ron IG, et al. Conjunctival involvement in classic (indolent) HIV negative Kaposi's sarcoma. *Br J Ophthalmol* 1994: 78:488-489.

Volberding P. Therapy of Kaposi's sarcoma in AIDS. *Semin Oncol* 1984; 11:60.

Karsch-Neugebauer Syndrome (Nystagmus-Split Hand Syndrome) 669

General: Autosomal dominant.

Ocular: Horizontal nystagmus; strabismus; cataract; fundus changes.

Clinical: Split hand and split foot deformities; monodactylous hands.

Karsch J. Erbliche Augenmissbildung in Verbendung mit Spalthand Und-Fuss. *Z Augenhulk* 1936; 89:274-279.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Neugebauer H. Splathand Und-Fuss mit Familiaerer Besonderheit. Z *Orthop* 1962; 95:500-506.

Kartagener Syndrome (Sinusitis-Bronchiectasis-Situs Inversus Syndrome; Bronchiectasis-Dextrocardia-Sinusitis; Kartagener Triad) 670

General: Autosomal recessive; onset in early infancy; occasionally dominant; finding of various structural defects in patients with this condition suggests that there are several genetic determinants.

Ocular: Myopia; glaucoma; conjunctival melanosis; iris coloboma; tortuous and dilated retinal vessels; retinal pigmentary degeneration; pseudopapillitis.

Clinical: Immotile cilia; situs inversus; bronchiectasis; sinusitis; various cardiovascular and renal abnormalities; dyspnea; productive cough; recurrent respiratory infections; palpitation; otitis media; nasal speech; conductive hearing loss; nasal polyps; situs inversus viscerum with hepatic dullness on left side.

Kartagener M. Zuir Pathogenese der Bronchiektasien, Bronchrektasien bei Situs Inversus. *Beitr Klin Tbk* 1933; 83:489.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Sturgess JM, et al. Genetic aspects of immotile cilia syndrome. *Am J Med Genet* 1986; 25:149-160.

Kasabach-Merritt Syndrome (Capillary Angioma-Thrombocytopenia; Hemangioma-Thrombocytopenia; Thrombocytopenia Purpura-Hemangioma) 671

General: Angioma causing sequestration of platelets and platelet deficiency.

Ocular: Capillary hemangiomas of the orbit; retinal detachments.

Clinical: Extraorbital hemangiomas found on trunk, extremities, and palate or in subglottic space; thrombocytopenia; found in infants; purpura and bleeding.

Kasabach HH, Merritt KK. Capillary hemangioma with extensive purpura: report of a case. *Am J Dis Child* 1940; 59: 1063-1 070.

Kelsch RD. Kasabach-Merritt syndrome: a case report and literature review. *Compendium* 1994; 15:590-598.

Lambert SR, et al. Serous retinal detachments in thrombotic thrombocytopenic purpura. *Arch Ophthalmol* 1985; 103: 1172-1174.

Kaufman Oculocerebrofacial Syndrome 672

General: Autosomal recessive; significant positive and negative features.

Ocular: Hypertelorism; epicanthal folds; ptosis; mongoloid obliquity; microcornea; pale optic disk; laterally broad eyebrows sparse.

Clinical: Flat philtrum; congenital hypotonia; micrognathia; respiratory distress; high narrow palate; lordosis; constipation; flat feet.

Kaufman R, et al. An oculocerebrofacial syndrome. *Birth Defects* 1971:7:135-138.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Kearns-Sayre Syndrome (Ophthalmoplegia Plus Syndrome; Kearns-Shy Syndrome; Kearns Disease) 673

General: Etiology unknown; sporadic (nonhereditary); onset before age 20 years; external ophthalmoplegia; complete heart block.

Ocular: Pigmentary degeneration of retina; progressive external ophthalmoplegia; corneal decompensation; optic neuritis.

Clinical: Abnormal mitochondria with paracrystalline inclusion in muscle cell; heart block; limb weakness; hyperglycemic acidotic coma; death; cerebellar dysfunction.

Bachynski BN, et al. Hyperglycemic acidotic coma and death in Kearns-Sayre syndrome. *Ophthalmology* 1986; 93: 391-396.

Chang TS, et al. Corneal decompensation in mitochondrial ophthalmoplegia plus (Kearns-Sayre) syndrome. A clinicopathologic case report. *Cornea* 1994; 13:269-273.

DiMauro S, Schon EA, Rowland LP. Mitochondrial encephalopathies. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Philadelphia: Williams & Wilkins, 1995:620.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Jean R, et al. Kearns' syndrome with metabolic disorders. *Arch Fr Pediatr* 1972; 29:436.

Kearns TPM, Sayre GP. Retinitis pigmentosa, external ophthalmoplegia and complete heart block. *Arch Ophthalmol* 1958; 60:280-289.

Menkes JR. Kearns-Sayre syndrome. In: Menkes JR, ed. *Textbook of Child Neurology*, 5th ed. Williams & Wilkins, 1995:851-852.

Ota y, et al. Early retinal involvement in mitochondrial myopathy with mitochondrial DNA deletion. *Retina* 1994; 14: 270-276.

Kenny Syndrome 674

General: Congenital syndrome.

Ocular: Nanophthalmos with hyperopia; papilledema; vascular tortuosity; macular crowding; bilateral optic atrophy.

Clinical: Dwarfism; thickened long bone cortex; transient hypocalcemia.

Boynton JR, et al. Ocular findings in Kenny's syndrome. *Arch Ophthalmol* 1979; 97:896-900.

Kenny FM, Linarelli L. Dwarfism and cortical thickening of tubular bones. *Am J Dis Child* 1966; 111:201-207.

Majewski F, et al. The Kenny syndrome: a rare type of growth deficiency with tubular stenosis, transient hypoparathyroidism and anomalies of refraction. *Eur J Pediatr* 1981; 136:21-30.

Rebolleda Fernandez G, et al. Bilateral optic atrophy in Kenny's syndrome. *Acta Ophthalmol* 1992; 70:135-138.

Keratitis Fugax Hereditaria 675

General: Autosomal dominant; onset from age 4 to 12 years; characterized by acute attacks of keratitis.

Ocular: Keratitis; corneal opacities.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Valle O. Keratitis fugax hereditaria. *Duodecim* 1964; 80:659-664.

Keratoconus Posticus Circumscriptus (KPC; KPC with Associated Malformations) 676

General: Autosomal recessive; rare; abnormality in corneal curvature centrally localized on its posterior surface in association with opacification of the overlying stroma; may be an anterior chamber cleavage defect with failure of normal separation of the lens and iris from the cornea.

Ocular: Corneal opacities; retinal coloboma; ptosis; hyperopia; iridocorneal adhesions; hypertelorism.

Clinical: Cleft lip; cleft palate; neck webbing; short stature; mental retardation; inguinal hernia; undescended testes; tight heel cords; vertebral anomalies; delayed bone age; double ureters; cone-shaped epiphyses; stubby limbs and digits; limitation of extension and supination of the elbows; brachydactyly; fifth finger clinodactyly; frequent urinary tract infections; prominent nose; mild maxillary hypoplasia; low posterior hairline; short, broad feet with bilateral pes cavus; bilateral ureteric reflux.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Young ID, et al. Keratoconus posticus circumscriptus, cleft lip and palate, genitourinary abnormalities, short stature, and mental retardation in sibs. *J Med Genet* 1982; 19:332-336.

Keratodermia Palmaris Et Plantaris (Palmoplantar Keratodermia; Keratosis Palmoplantaris) 677

General: Autosomal recessive; hereditary disorder; diffuse or focal thickening of the palms and soles.

Ocular: Hyperkeratosis of lid and cornea; ectropion; leukoma; corneal ulceration; pronounced photophobia; hereditary optic atrophy; epiphora; conjunctivitis.

Clinical: Localized or disseminated hyperkeratotic changes of the palms and soles with a tendency toward fissure and secondary infection.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Schopf E, et al. Syndrome of cystic eyelids, palmo-plantar keratosis, hypodontia and hypotrichosis as a possible autosomal recessive trait. *Birth Defects* 1971; 7:219-221.

Kiloh-Nevin Syndrome (Muscular Dystrophy of External Ocular Muscles; Ocular Myopathy) 678

General: Etiology unknown; autosomal dominant.

Ocular: Ptosis; orbicularis muscle weakness; ocular myopathy; diplopia progressing to bilateral myopathic ophthalmoplegia; may be associated with pigmentary retinopathy and heart block (see Kearns-Sayre syndrome).

Clinical: Progressive muscular dystrophy in which facial muscles may be involved; occasionally, hereditary ataxia; pain; myokymia.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duszowa J, et al. The Kiloh-Nevin syndrome. *Klin Oczna* 1974; 44:805.

Franzini A, et al. Pain syndrome and focal myokymia due to anterior interosseous neurovascular relationships: report of a case and neurophysiological. *J Neurosurg* 1995; 82:578-580.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kiloh LG, Nevin S. Progressive dystrophy of the external ocular muscles (ocular myopathy). *Brain* 1951; 74:115.

Olenius M, Nylen B. Ocular myopathy with palpebral ptosis. *Ann Plast Surg* 1987; 19:146-153.

Kimmelstiel-Wilson Syndrome (Diabetes Mellitus-Hypertension-Nephrosis Syndrome; Diabetes-Nephrosis Syndrome; Diabetic Glomerulosclerosis; Intercapillary Glomerulosclerosis; Renal Glomerulohyalinosis-Diabetic Syndrome) 679

General: Occurs in patients with diabetes mellitus of several years' duration.

Ocular: Retinal lesions, including hemorrhages, exudates, and neovascularization.

Clinical: Hypertension; proteinuria; edema; glomerulonephrosis; arteriosclerosis; capillary or intercapillary glomerulosclerosis; eosinophilic nodules; hyaline degeneration of the renal arterioles.

Geeraets WI. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kimmelstiel P, Wilson C. Benign and malignant hypertension and nephrosclerosis: a clinical and pathological study. *Am J Pathol* 1936; 12:45-81.

Kinsbourine Syndrome (Dancing Eyes Syndrome; Opsoclonus-Myoclonus Syndrome) 680

General: Etiology unknown; occurs in infancy and early childhood; myoclonic encephalopathy; has been reported as the only manifestation of a postinfectious syndrome without evidence of encephalitis.

Ocular: Twitching of lids and eyebrows on occasion, more pronounced with activity than at rest; irregular vertical movements, jerky in appearance and sometimes with some lateral nystagmic components.

Clinical: Sporadic, jerky movements of head, trunk, and limbs, usually more pronounced when the child is active; lack of coordination; ataxia; irritability; mental retardation; chronic neurologic deficits.

Bhatia K, et al. "Isolated" postinfectious myoclonus. *J Neurol Neurosurg Psychiatry* 1992; 55:1089-1091.

Geeraets WI *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kinsbourine M. Myoclonic encephalopathy of infants. *J Neurol Neurosurg Psychiatr* 1962; 25:271.

Koh PS, et al. Long-term outcome in children with opsoclonus-myoclonus and ataxia and coincident neuroblastoma. *J Pediatr* 1994; 125:712-716.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Kirk Syndrome 681

General: Etiology unknown; raised hereditary masses over central cornea; rare, only one family known to be affected.

Ocular: Photophobia; excessive lacrimation; amyloid corneal deposits.

Clinical: None.

Kirk HQ, et al. Primary familial amyloidosis of the cornea. *Trans Am Acad Ophthalmol Otolaryngol* 1973; 77: 411-417.

Kjellin Syndrome 682

General: Autosomal recessive disorder; degeneration progressive.

Ocular: Yellow retinal flecks that lie at the pigment epithelial level; poor visual perceptual skills; central retinal degeneration.

Clinical: Spastic paraparesis; dementia; progressive lower extremity weakness; dysarthric speech; muscle atrophy.

Farmer SG, et al. Fleck retina in Kjellin's syndrome. *Am J Ophthalmol* 1985; 99:45-50.

Kjellin K. Familial spastic paraplegia with amyotrophy, oligophrenia, and central retinal degeneration. *Arch Neurol* 1959; 1: 133.

Traboulsi EI. Fleck retina in Kjellin's syndrome. *Am J Ophthalmol* 1985; 99:738-739.

Klein Syndrome 683

General: Autosomal dominant; belongs to the group of iridodermatoauditive dysplasias.

Ocular: Hypertelorism; blepharophimosis; hypertrichosis; blue irides; heterochromia.

Clinical: Bilateral labyrinthine deafness; mandibular retrognathism; skull deformities and arched palate; partial albinism of skin and hair; syndactylism (cutaneous).

Duke-Elder S, ed. *System of Ophthalmology, vol. III, part* 2. St. Louis: CV Mosby, 1964.

Klein D. Albinisme Partiel (Leucisme), Accompagne de Surdi-mutite, d'Osteomyodysplasie, de Raideurs Articulaires Congenitales Multiples et d'Autres Malformations Congenitales. *Arch Julius Klaus Stift Vererb Forsch* 1947; 22:336, and *Helv Paediatr Acta* 1950; 5:38.

Klinefelter Syndrome (Gynecomastia-Aspermatogenesis Syndrome; XXY Syndrome; XXXY Syndrome; XXYY Syndrome; Reifenstein-Albright Syndrome) 684

General: Occurrence in 1% of retarded males; phenotypically males with positive female sex chromatin; karyotype shows 47 chromosomes, 44 autosomes, and 3 sex chromosomes with the complement XXY.

Ocular: Anophthalmos; coloboma; corneal opacities.

Clinical: Testicular hypoplasia; sterility; gynecomastia; eunuchoid physique; mental retardation; association with progressive systemic sclerosis and systemic lupus erythematosus.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Klinefelter HF Jr. Syndrome characterized by gynecomastia, aspermatogenesis without aleydigism and increased excretion of follicle-stimulating hormone. *J Clin Endocrinol* 1942; 2:615.

Kobayashi S, et al. Klinefelter's syndrome associated with progressive systemic sclerosis: report of a case and review of the literature. *Clin Rheumatol* 1991; 10:84-86.

Mandoki MW, et al. A review of Klinefelter's syndrome in children and adolescents. *J Am Acad Child Adolesc Psychiatry* 1991; 30:167-172.

Welter DA, et al. Klinefelter's syndrome with anophthalmos. *Am J Ophthalmol* 1974; 77:895.

Klippel-Feil Syndrome (Congenital Brevicollis; Synostosis of Cervical Vertebra) 685

General: Autosomal recessive inheritance; females more commonly affected; progressive paraplegia may develop late in life.

Ocular: Esotropia; hypertropia combined with torticollis; occasional horizontal nystagmus.

Clinical: Platybasia; congenital upward displacement of scapula (Sprengel deformity); brevicollis; immobility of neck (painless); low posterior hairline; peculiar facies; cleft palate; short stature; congenital brevicollis; vertebral abnormalities; autosomal dominant mode of inheritance has been reported .

Brik M, Athayde A. Bilateral Duane's syndrome, paroxysmal lacrimation and Klippel-Feil anomaly. *Ophthalmologica* 1973; 167:1.

Klippel LM, Feil A. Anomalie de la Colonne Vertebrale par l'Absence des Vertebrales Cervicales; Cage Thoracique Remontant Jusqua la Base du Crane. *Bull Soc Anat (Paris)* 1912; 14:185.

Mathieu M, et al. A familial syndrome with micrognathia, cleft palate, short neck, and stature, vertebral anomalies and mental retardation. *Genet Couns* 1993; 4:299-303.

Klippel- Trenaunay-Weber Syndrome (Parkes-Weber Syndrome; Angio-Osteo-Hypertrophy Syndrome) 686

General: Most frequently inherited as irregular dominant; however, reported to be recessive with parent consanguinity; association of Klippel-Trenaunay-Weber syndrome and Sturge-Weber syndrome has been reported.

Ocular: Enophthalmos; unilateral hydrophthalmos; conjunctival telangiectasia; atypical iris coloboma; cataract; irregular and dilated retinal vessels; choroidal angiomas; exudative outer retinal vascular masses.

Clinical: Vascular nevi; varicose vessels; capillary angiomas; lymphangioma; arteriovenous aneurysm; hypertrophy of soft tissues and bones (local); phlebitis; thrombosis; syndactyly; polydactyly; early eruption of teeth; hemifacial hypertrophy.

Brod RD, et al. Unusual retinal and renal vascular lesions in the Klippel-Trenaunay-Weber syndrome. *Retina* 1992; 12:355-358.

Furukawa T, et al. Sturge-Weber and Klippel-Trenaunay syndrome with nevus Ota and Ito. *Arch Dermatol* 1970; 102: 640.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Klippel M, Trenaunay P. Naevus Variqueux Osteo-Hypertrophique. *J Prat* 1900; 14:65.

Weber FP. Angioma-formation in connection with hypertrophy of limbs and hemi-hypertrophy. *Br J Dermatol* 1907; 19:231.

Kloepfer Syndrome 687

General: Rare autosomal recessive disease; manifestations at age 2 months; death between ages 21 and 30 years.

Ocular: Progressive loss of vision to complete blindness associated with progressive dementia.

Clinical: Severe blistering in sunlight; no increase in weight and height after erythema subsides at age 5 to 6 years; mental age does not progress beyond the level of imbeciles; progressive degenerative dementia occurs during or immediately after adolescence.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kloepfer HW. Progress report on study of a type of progressive juvenile dementia with oligophrenia and erythema. In: *Proceedings of the 10th International Congress of Genetics*, Vol. II, Montreal, August 20-27, 1958. Toronto: University of Toronto Press, 1958:146.

Klüver-Bucy Syndrome (Temporal Lobectomy Behavior Syndrome) 688

General: Occurs after temporal lobectomy, carried out therapeutically for temporal lobe epilepsy.

Ocular: "Psychic blindness" or visual agnosia.

Clinical: Changes in emotional behavior (possible rage reactions); hypersexuality; bulimia (changes in dietary habits); loss of recognition of people; strong oral tendencies (i.e., licking, biting, chewing); deficiency of memory; psychic blindness; aberrant sexual behavior; hypermetamorphosis; aphasia; visual agnosia; memory deficit; speech disturbance; syndrome in adults is commonly associated with neurodegenerative conditions, following radiation therapy, or after temporal lobectomy; syndrome in children has been recognized almost exclusively in association with acute bitemporal injury or dysfunction.

Hooschmand H, et al. Kluver-Bucy syndrome: successful treatment with carbamazepine. *JAMA* 1974; 229:1782.

Kluver H, Bucy P. "Psychic blindness" and other symptoms following bilateral temporal lobectomies in rhesus monkeys. *Am J Physiol* 1937; 119:352.

Lanska DJ, Lanska MI Kluver-Bucy syndrome in juvenile neuronal ceroid lipofuscinosis. *J Child Neurol* 1994; 9: 67-69.

Kniest Dysplasia (Metatropic Dwarfism Type II) 689

General: Autosomal dominant disease; Swiss cheese pattern on cartilage biopsy specimen; due to either alteration metabolism of proteoglycans or abnormality in collagen synthesis; both sexes affected; collagen type II collagenopathy; produced by a single amino acid substitution in the type II collagen triple helix.

Ocular: Retinal detachment; severe myopia; cataracts; dislocated lenses; blepharoptosis; vitreoretinal degeneration; vitreous traction; congenital glaucoma; hypertelorism; mild synophrys; epicanthal folds; perivascular lattice degeneration, white without pressure.

Clinical: Severe short stature; typical facies with flat nasal bridge; cleft palate; hearing loss; joint contractures; lordosis; kyphosis.

Douglas GR. The ocular findings in Kniest dysplasia. *Am J Ophthalmol* 1986; 100:860-861.

Maumenee IH, Traboulsi EI. The ocular findings m Kniest dysplasia. *Am J Ophthalmol* 1985; 100:155-160.

Regenbogen LS, Co seas GJ, eds. *Ocular-auditory syndromes.* New York: Masson, 1985; 153-156.

Wilkin DJ, et al. A single amino acid substitution (G 103D) in the type II collagen triple helix produces Kniest dysplasia. *Hum Mol Genet* 1994; 3:1999-2003.

Knobloch Syndrome 690

General: Autosomal recessive; retinal detachment with occipital encephalocele.

Ocular: High myopia; retinal detachment; vitreoretinal degeneration; persistent papillary membrane; posterior vitreous detachment; retinochoroidal staphylomas.

Clinical: Occipital encephalocele; normal intelligence; congenital midline scalp defect; unusual plantar creases.

Cohen MM, et al. Syndrome with cephalocele. *Teratology* 1982; 25:161-172.

Duh EJ, Yao YL, Dagll M, et al. Persistance of fetal vasculature in a patient with Knobloch syndrome. *Ophthalmology* 2004; 111: 1885-1888.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Passos-Bueno MR, et al. Knobloch syndrome in a large Brazilian consanguineous family: confirmation of autosomal recessive inheritance. *Am J Med Genet* 1994; 52:170-173.

Koby Syndrome (Floriform Cataract) 691

General: Autosomal dominant; both sexes affected.

Ocular: Multiple opacities of different shapes (annular, floriform, and polychromatic); found especially around embryonic nucleus.

Clinical: None.

Doggart JR. Congenital cataract. *Trans Ophthalmol Soc UK* 1957; 77:31-37.

Koby FE. Cataracte Familiale d'un Type Particulier, Se Transmettant Apparentent Suivant le Mode Dominant. *Arch Ophthalmol* 1923; 40:492-503.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: m Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Koerber-Salus-Elschnig Syndrome (Sylvian Aqueduct Syndrome; Nystagmus Retractorius Syndrome) 692

General: Caused by tumor or inflammation in region of aqueduct of Sylvius, third and fourth ventricle, or corpora quadrigemina.

Ocular: Lid retraction may be associated with midbrain lesions above the posterior commissure; paresis of vertical gaze; tonic spasm of convergence on attempted upward gaze; clonic convergence movements or convergence nystagmus; vertical nystagmus on gaze up or down; nystagmus retractorius with spasmodic retraction of the eyes when an attempt is made to move them in any direction; occasional extraocular muscle paresis.

Clinical: Headaches; dizziness; hypertension; possible hemiparesis; ataxia; hemitremor; Babinski's sign.

Elschnig A. Nystagmus Retractorius, ein Cerebrales Herdsymptom. *Med Klin* 1913; 9:8.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol. III* 4th ed. Baltimore: Williams &Wilkins, 1988.

Salus R. On acquired retraction movements of the eyes. *Arch Ophthalmol* 1913; 42:34.

Smith JL, et al. Nystagmus retractorius. *Arch Ophthalmol* 1959; 62:864.

Kohn-Romano Syndrome (BPES Syndrome) 693

General: Autosomal dominant transmittance; tetrad with telecanthus, ptosis, epicanthus inversus, and blepharophimosis; male preponderance; location of the abnormal gene responsible for this syndrome has been postulated to be a 3q2.

Ocular: Telecanthus; ptosis; epicanthus inversus; blepharophimoses; divergent strabismus; nystagmus; esotropia; anomalies of the lacrimal punctum; reduced corneal diameter.

Clinical: Highly arched palate; low-set ears with deformed pinnas.

de Die-Smulders CEM, et al. Further evidence for the location of the BPES gene at 3q2 [Letter]. *J Med Genet* 1991; 28:725.

Garden JW. Blepharophimosis, ptosis, epicanthus inversus, and lacrimal stenosis. *Am J Ophthalmol* 1969; 67:153.

Kohn R, Romano PE. Ptosis, blepharophimosis, epicanthus inversus, and telecanthus: the syndrome with no name. *Am J Ophthalmol* 1971; 72:625.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Komoto Syndrome (Congenital Eyelid Tetrad; CET) 694

General: Autosomal dominant; all races affected; most patients are of normal intelligence.

Ocular: Ptosis; epicanthus inversus; blepharophimosis; telecanthus.

Bergin DJ, La Piana FG. Natural history of the congenital eyelid tetrad (Komoto's syndrome). *Ann Ophthalmol* 1981; 13:1145-1148.

Bergin DJ, et al. Natural history of the congenital eyelid tetrad. *Ann Ophthalmol* 1981; 13:1145-1148.

Komoto J. Veber die Operation be; Hereditaren Phimosis Congenita mit Ptosis. *Nippon Ganka Gakkai* 1920; 24, and *Klin Monatsbl Augenheilkd* 1921; 66:952.

Krabbe (2) Syndrome (Sturge-Weber-Krabbe Syndrome; Galactocerebrosidase Deficiency) 695

General: Etiology unknown; some evidence of irregularly dominant transmission; variant of Sturge-Weber syndrome; both sexes affected; present from birth; appears that the GALC locus lies somewhere in the region 14q21-q31.

Ocular: Buphthalmos; conjunctival angiomas; choroidal angiomatosis; retinal aneurysm.

Clinical: Cerebral angiomas (possible calcium deposition); flat angioma of the skin in the distribution area of the trigeminal nerve (V) (nevus flammeus); mental deterioration due to progressive atrophy of the brain; contralateral hemiplegia; facial hemiatrophy.

Brownstein S, et al. Optic nerve in globoid leukodystrophy (Krabbe's disease). Ultrastructural changes. *Arch Ophthalmol* 1978; 96:864-870.

Duke-Elder S, ed. *System of Ophthalmology, vol. III, part* 2. Baltimore: Williams & Wilkins, 1976: 1122.

Krabbe K. Facial and meningeal angiomatosis associated with calcification of the brain cortex: a clinical and anatopathologic contribution. *Arch Neural Psychiatr* 1934; 32:737.

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Zlotogora J, et al. Krabbe disease locus mapped to chromosome 14 by genetic linkage. *Am J Hum Genet* 1990; 47: 37-44.

Krause Syndrome (Congenital Encephalo-Ophthalmic Dysplasia; Encephalo-Ophthalmic Syndrome) 696

General: No hereditary factors involved; no predilection for either sex; more frequent in premature infants; death frequently from intercurrent infections.

Ocular: Microphthalmos; enophthalmos; ptosis; strabismus; secondary glaucoma; iris atrophy; anterior and posterior synechiae; scleral atrophy; persistent remnants of hyaloids artery; intraocular hemorrhages and exudates; cyclitic membranes; cataracts; retinal hypoplasia and hyperplasia; choroidal and retinal malformation; retinal glial membranes; retinal detachment; choroidal atrophy; optic nerve malformation; optic atrophy.

Clinical: Congenital cerebral dysplasia; hydrocephalus or microcephaly; mental retardation; heterotopia.

Krause AC. Congenital encephala-ophthalmic dysplasia. *Arch Ophthalmol* 1946; 36:387.

Miller M. et al. A chromosomal anomaly with multiple ocular defects. *Am J Ophthalmol* 1963; 55:901.

Kufs Disease (Adult Chronic G_{M2} Gangliosidosis; Gangliosidosis G_{M2} Adult Type; Hallervorden-Kufs Syndrome) 697

General: Autosomal recessive; hexosaminase A decrease; onset in the third and fourth decades of life; slow death within 19 or 20 years of onset; very rare type of adult neuronal ceroid lipofuscinosis; autosomal recessive and dominant inheritance have been reported.

Ocular: Retinal storage; ocular lesions usually are absent; rare macular discoloration; no pigmentary degeneration of the retina.

Clinical: Dementia and behavioral changes; progressive gait and postural deterioration; mild ataxia; dysarthria; ascending muscular atrophy; pes cavus; cerebral degeneration; apathy; progressive myoclonus epilepsy; aphasia; facial dyskinesias; lipid infiltration of the brain cells is the principal pathologic feature (see Ceroid Lipofuscinosis).

Bateman JB, Philippart M. Ocular features of Hagberg-Santavuori syndrome. *Am J Ophthalmol* 1986; 102:262-271.

Brod RD, et al. Diagnosis of neuronal ceroid lipofuscinosis by ultrastructural examination of peripheral blood lymphocytes. *Arch Ophthalmol* 1987; 105:1388-1393.

Dom R, et al. Adult ceroid-lipofuscinosis (Kufs' disease) in two brothers, retinal and visceral storage in one; diagnostic muscle biopsy in the other. *Acta Neuropathol* 1979; 45:67-72.

Kufs H. Uber eine Spatform der Anaurotischen Idiotie und Ihre Heredofamilia ren Grundlagen. Z *Neurol Psychiatr* 1925; 95:169-188.

Martin JJ. Adult type of neuronal ceroid lipofuscinosis. *Dev Neurosci* 1991; 13:331-338.

Vital A, et al. Adult dementia due to intraneuronal accumulation of ceroid lipofuscinosis (Kufs' disease): ultrastructural study of two cases. *J Geriatr Psychiatry Neurol* 1991; 4:110-115.

Kugelberg-Welander Syndrome (Juvenile Muscular Atrophy) 698

General: Autosomal recessive; juvenile spinal muscular atrophy; affects both sexes; onset in late childhood or adolescence.

Ocular: Ptosis; ophthalmoplegia; exotropia; orbicularis oculi paresis.

Clinical: Slowly progressive proximal muscle atrophy; lower extremities usually are affected first, with the upper limbs being affected late; frequently, fasciculation; proximal muscle weakness, especially of the lower extremities; elevated serum creatine kinase levels.

Dorsher PT, et al. Wohlfart-Kugelberg-Welander syndrome: serum creatine kinase and functional outcome. *Arch Phys Med Rehab* 1991; 72:587-591.

Kugelberg E, Welander L. Familial neurogenic muscular atrophy simulating ordinary proximal dystrophy. *Acta Psychiatr* 1954; 29:42-43.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Menkes JH. Diseases of the motor unit. In: Menkes JH, ed. *Textbook of child neurology*, 5th Ed. Philadelphia: Williams & Wilkins, 1995:815-820.

Rowland LP. Progressive spinal muscular atrophies of childhood. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Philadelphia: Williams & Wilkins, 1995:743,

Urbanek KO, Chudackova J, Veliskova J. ACTH and steroids in Kugelberg- Welander disease, *Acta Univ Pallacki Olomuc Fac Med* 1990; 126:147-150.

Wollar PH, Reece JM. Ocular findings in a patient with Kugelberg- Welander syndrome: a case report. *J Pediatr Ophthalmol Strabismus* 1978; 15:15-18.

Kuru Syndrome (Laughing Death) 699

General: Restricted to Fore tribe of eastern New Guinea; prevalent in children and adult women; etiology unknown, possibly related to the tribe's practice of cannibalism; uncertain whether significant genetic factors also are involved.

Ocular: Strabismus; nystagmus.

Clinical: Ataxia; trembling leg muscles; incoordination; exaggeration of voluntary movements; jerks; slurred speech; fecal incontinence; aphonia; dysphagia.

Gajdusek DC, et al. Experimental transmission of kuru-like syndrome to chimpanzees. *Nature* 1966; 209:794-796.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Kussmaul Disease (Kussmaul-Maier Disease; Necrotizing Angiitis; PAN; Periarteritis Nodosa; Polyarteritis Nodosa) 700

General: Progressive process of vascular inflammation and necrosis, manifested by numerous nodules along the course of small- and medium-sized arteries; lesions are segmental in distribution, have a predilection for bifurcation and involve all but the pulmonary arteries; arteries in gastrointestinal tract, kidneys, and muscles are particularly affected; affects primarily males between ages 20 and 50 years.

Ocular: Retinal detachment; cotton-wool patches; polyarteritis nodosa lesion of arteries; pseudoretinitis pigmentosa; conjunctivitis; corneal ulcer; tenonitis; ptosis; exophthalmos; proptosis; uveitis; optic atrophy; cataract; scleritis; paralysis of extraocular muscles; neuroretinitis; anterior uveitis; macular star; peripheral ulcerative keratitis; retinal vasculitis; pseudotumor of the orbit; central retinal artery occlusion.

Clinical: Fever; myalgia; hypertension; gastrointestinal disorders; neuropathy; respiratory infection; weight loss; anginal pain; hemiplegia; convulsion; acute brain syndrome; skin lesions; diffuse erythema; purpura; urticaria; gangrene; tachycardia; pericarditis; aortitis; painful facial swelling; diplopia.

Akova YA, et al. Ocular presentation of polyarteritis nodosa. Clinical course and management with steroid and cytotoxic therapy. *Ophthalmology* 1993; 100: 1775-1781.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kussmaul A, Maier R. Ueber Eine Bisher Nicht Beschriebene Eigenthumliche Artenener Krankung (Periarteritis Nodosa), die Mit Morbus Brightii und Rapid Fortschreitender Allgemeiner Muskellahumung Einhergeht. *Dtsch Arch Klin Med* 1866; 1:484-518.

Matsuda A, et al. A case of neuroretinitis associated with long-standing polyarteritis nodosa. *Ophthalmologica* 1994; 208:168-171.

Solomon SM. Solomon JH. Bilateral central retinal artery occlusions in polyarteritis nodosa. *Ann Ophthalmol* 1978; 10:567-569.

Kyrle Disease (Hyperkeratosis Follicularis and Parafollicularis in Cutem Penetrans; Hyperkeratosis Penetrans) 701

General: Etiology unknown; associated in siblings, suggesting heredofamilial condition; syndrome may belong in the diabetic syndromes; rare skin disorder; onset usually in the third to sixth decades of life.

Ocular: Subcapsular cataracts; corneal changes characterized as minute, yellow-brown subepithelial opacities, noted to be more dense and more deeply penetrating in the peripheral cornea than in the central cornea.

Clinical: Multiple flesh-colored, horny papules associated with hair follicles.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981,467.

Sehgal VN, et al. Perforating dermatoses: a review and report of four cases. *J Dermatol* 1993; 20:329-340.

Lacrimal Duct Defect 702

General: Autosomal dominant.

Ocular: Imperforate nasolacrimal ducts with or without absence of puncta and canaliculi.

Clinical: None.

Lumbroso BD. On a case of congenital atresia of the lacrimal ducts with familial characteristics. *Acta Genet Med Gemellol* 1960; 9:290-295.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Lacrimo-Auriculo-Dento-Digital Syndrome (Ladd-Levy-Hollister Syndrome) 703

General: Autosomal dominant; all features have been reported as isolated traits.

Ocular: Aplasia or hypoplasia of puncta; obstruction of nasolacrimal ducts; bilateral lacrimal duct fistula; poor tear production; absent lacrimal puncta; chronic dacryocystitis; dry eyes; epiphora.

Clinical: Cup-shaped pinnas; mixed hearing deficit; small and peg-shaped lateral maxillary incisors; mild enamel dysplasia; fifth finger clinodactyly; duplication of the distal phalanx of the thumb; triphalangeal thumb; syndactyly; unilateral radial aplasia; poor saliva production; salivary gland hyposecretion; dental hypoplasia; dysplasia; cup-shaped ears with hearing loss; digital anomalies.

Bamfprtj JS, Kaurah P. Lacrimo-auriculo-dento-digital syndrome: evidence for lower limb involvement and severe congenital renal anomalies. *Am J Med Genet* 1992; 43:932-937.

Heinz GW, et al. Ocular manifestations of the lacrimo-auriculodento-digital syndrome. *Am J Ophthalmol* 1993; 115: 243-248.

Hoyme HE, Kreutz JM. The Levy Hollister syndrome. *Proc Greenwood Genet Center* 1985; 4:122-123.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Langer-Giedion Syndrome (Trichorhinophalangeal Syndrome, Type II) 704

General: Rare congenital condition.

Ocular: Iris colobomata.

Clinical: Mental retardation, bulbous nose, sparse hair, cone-shaped epiphyses, microcephaly, multiple exostoses, redundant skin; less consistently, "floppy infants," hyperextensible joints, recurrent upper respiratory tract infections, delayed speech development, and characteristic facies.

Buhler EM. Langer-Giedion syndrome and 8q- deletion. *Am J Med Genet* 1982; 11:359.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Lanzieri Syndrome 705

General: Developmental anomaly that belongs to group of craniofacial malformations; present from birth.

Ocular: Microphthalmia; anophthalmos; iris coloboma; cataracts; retinal and choroidal coloboma; optic nerve coloboma.

Clinical: Dwarfism; dyscephalia; dental anomalies; hypertrichosis; skin atrophy; absence of fibula, some tarsal and metatarsal bones.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Lanzieri M. On a rare association of craniofacial malformative syndrome and congenital absence of the fibula. *Ann Ottal Clin Ocul* 1961; 87:667.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Laron Syndrome 706

General: Autosomal recessivel; insulinelike growth factor I hormone resistance

Ocular: Microphthalmia; reduced retinal vascularization; optic nerve hypoplasia; pseudopapilledema

Clinical: Short stature; abnormally small extremities; subnormal head circumference, increased body fat and delayed sexual development

Bourla OH, Laron Z, Snir M, et al. Insulinlike growth factor 1 affects ocular development. *Ophthalmology* 2006; 113: 1197-1200

Larsen Syndrome 707

General: Etiology unknown; autosomal recessive; possibly dominant in some cases.

Ocular: Hypertelorism; bilateral chronic keratitis; corneal neovascularization; lower lid entropion.

Clinical: Frontal bossing; depressed nasal bridge; flat face; flat and broad thumbs; skeletal dysplasia with multiple joint dislocations; unusual faces; long, cylindrical fingers; spatulate thumbs; dental abnormalities; cardiac defects; hydrocephalus; laryngotracheomalacia; dislocation of the cervical spine; tracheomalacias; heart disease; severe respiratory infection; clubfeet; multiple joint deformities; hydrocephalus; tracheal stenosis.

Bixler D, et al. Hypertelorism, microtia, and facial clefting. *Am J Dis Child* 1969; 118:495-500.

Klenn PJ, Iozzo RV. Larsen's syndrome with novel congenital anomalies. *Hum Pathol* 1991; 22:1055-1057.

Larsen LJ, et al. Multiple congenital dislocations associated with characteristic facial abnormality. *J Pediatr* 1950; 37:574-581.

Laville JM, et al. Larsen's syndrome: review of the literature and analysis of thirty-eight cases. *J Pediatr Orthop* 1994; 14:63-73.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Lateral Sinus Thrombosis (Sigmoid Sinus Thrombosis) 708

General: Predominant in children; acute onset secondary to chronic otitic infections; high mortality rate without treatment; any microorganism may be responsible for infection and secondary thrombosis; associated with oral contraceptive usage.

Ocular: Pain behind the eye; muscle palsies; papilledema (50%).

Clinical: Fever; headaches; nausea; vomiting; swelling over mastoid region; intracranial hypertension; seizures; hemiplegia.

Dickins JR, Graham SS. Neurotologic presentation of sagittal sinus thromboses associated with oral usage. *Am J Otol* 1993; 14:544-547.

Kelly KE, et al. Diagnosis of septic sigmoid sinus thrombosis with magnetic resonance imaging. *Otolaryngol Head Neck Surg* 1991; 105:617-6j24.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Vick NA. *Grinker's neurology*, 7th ed. Springfield, IL: Charles C. Thomas, 1976.

Lattice Degeneration and Retinal Detachment 709

General: Autosomal dominant; progressive; lattice degeneration precedes retinal detachment by about 20 years; familial occurrence of lattice degeneration in nonmyopes has been reported.

Ocular: Myopia; retinoschisis; peripheral retinal degeneration; lattice degeneration.

Clinical: None.

Everet TWG. Study of a family with lattice degeneration and retinal detachment. *Am J Ophthalmol* 1968; 65: 229-232.

Lemeke HH, Pischel DK. The time interval in the development of lattice degeneration of the retina. *Am J Ophthalmol* 1966; 61:1216.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Laurence-Moon-Bardet-Biedl Syndrome (Bardet-Biedl Syndrome; Retinitis Pigmentosa-Polydactyly-Adiposogenital Syndrome) 710

General: Recessive, dominant autosomal, and recessive sex-linked gene; male preponderance; onset in childhood; cases of Laurence-Moon belong to the group of heredoataxias.

Ocular: Ptosis; epicanthus; nystagmus; strabismus; night blindness; myopia; hypermetropia; iris coloboma; retinitis pigmentosa "bone corpuscles"; macular degeneration; attenuation of retinal vessels; choroidal atrophy; optic nerve atrophy; cataract; microphthalmia; keratoconus.

Clinical: Obesity (Fröhlich type); hypogenitalism; reduced intelligence and mental retardation; turricephaly; shortness of stature; atresia ani; genu valgum; congenital heart disease; polydactyly; body hair scant or absent; pseudogynecomastia.

Berson EL, et al. Progressive cone-rod degeneration. *Arch Ophthalmol* 1968; 80:68.

Fannemel M, et al. High-resolution chromosome analysis in autosomal recessive disorders: Laurence-Moon-Bardet-Biedl syndrome. *Clin Genet* 1993; 43:111-112.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

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McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Rizzo JF, et al. Retinal and neurologic findings in the Laurence-Moon-Bardet-Biedl phenotype. *Ophthalmology* 1986; 93: 1452-1456.

Lead Poisoning 711

General: Now rare and mostly of industrial origin; cumulative poisoning; excreted slowly; absorption slow by any route; prolonged exposure required for development of symptoms; acute poisoning virtually nonexistent.

Ocular: Sclerosis and obliteration of choroidal vessels; retinal arterial spasms; retrobulbar neuritis; papilledema; optic atrophy; cortical blindness; divergence palsy; papillary paralysis; bilateral abducens paralysis; accommodative palsy; mechanism of ocular pathology with this condition is not well defined, although there is evidence pointing to the level of cyclic adenosine monophosphate.

Clinical: Loss of appetite; weight loss; colic; constipation; insomnia; headache; dizziness; irritability; moderate hypertension; albuminuria; anemia; blue line edge of gum; encephalopathy; peripheral neuropathy leading to paralysis; convulsions; mania; coma.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Chisolm JJ. Treatment of lead poisoning. *Mod Treatment* 1967; 4:710.

Citirik M, Acaroglu G, Mutlway AH, et al. Lead poisoning : report of a case. Ann Ophthalmol 2004; 36: 32-36

Fox DA. Lead-induced alterations in rod-mediated visual functions and cGMP metabolism: new insights. *Neurotoxicology* 1994; 15:503-512.

Leber Hereditary Optic Neuropathy (Optic Atrophy Amaurosis; Pituitary Syndrome; Leber Syndrome) 712

General: Male preponderance; in acute phase of neuropathy there are three characteristic fundus changes: circumpapillary micro angiopathy, pseudoedema around the disk, and absence of staining on fluorescein angiography; possibly a toxic metabolic disorder, an abnormality of cyanide metabolism, or an effect of smoking; maternally inherited disease affecting young males presenting with unilateral or bilateral visual loss; second eye becomes involved within weeks to months later; positive association with an inherited mutation in mitochondrial deoxyribonucleic acid (DNA).

Ocular: Sudden severe loss of vision, which usually reaches its maximum after I or 2 months; complete blindness rare; central vision remains seriously impaired; occasional considerable visual improvement; sheathing of retinal vessels; circumpapillary telangiectatic microangiopathy; initial low-grade optic neuritis, then bilateral optic atrophy (partial or complete); possible swelling of the disk with hemorrhages and exudates, but usually transitory; nystagmus; macular colobomas; optic disk edema; cataracts; keratoconus; hyperemia of the disk; swelling of peripapillary nerve fiber layer.

Clinical: Headaches and vertigo; Uhthoff sign.

Godel V, et al. Congenital Leber amaurosis, keratoconus, and mental retardation in familial juvenile nephronophthisis. *J Pediatr Ophthalmol Strabismus* 1978; 15:89-91.

Leber T. Beitrage zur Kenntniss dee Atrophischen Veranderungen des Schnerven nebst Bemerkungen uber die normale Structur des Nerven. *Arch Ophthalmol* 1868; 14:164-176.

Newman NJ. Leber's hereditary optic neuropathy. *Ophthalmol Clin N Am* 1991; 4:431.

Hurko O. Mitochondrial DNA mutations in Leber's optic neuropathy. *Ann Neurol* 1994; 35:636.

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Nikoskelainen EK, et al. Leber's hereditary optic neuroretinopathy: a maternally inherited disease. *Arch Ophthalmol* 1987; 105:665-671.

Smith JL, et al. Ocular fundus in acute Leber optic neuropathy. *Arch Ophthalmol* 1973; 90:349.

Wagner RS, et al. High hyperopia in Leber's congenital amaurosis. *Arch Ophthalmol* 1985; 103:1507-1509.

Leber Tapetoretinal Dystrophy Syndrome (Amaurosis Congenita; Retinal Aplasia; Retinal Abiotrophy; Pigmentary Retinitis with Congenital Amaurosis; Dysgenesis Neuroepithelialis Retinae; Alstrom-Olsen Syndrome) 713

General: Autosomal recessive inheritance; consanguinity; occurs from teens to 30 years of age.

Ocular: Nystagmus; keratoconus; narrow retinal arteries; yellowish-brown or gray macular lesions; grayish atrophic retinal lesions; salt-and-pepper-like retinal pigmentation or typical "bone corpuscle" pigmentary changes; keratoglobus.

Clinical: Mental retardation; microcephaly; mongoloid appearance; oculodigital sign; association with Down syndrome has been reported; hypoplasia of the cerebellar vermis; mild-to-moderate ventriculomegaly.

Elder MJ. Leber congenital amaurosis and its association with keratoconus and keratoglobus. *J Pediatr Ophthalmol Strabismus* 1994; 31:38-40.

Firat T. Clinical and genetic investigations in Leber's tapeto-retinal dystrophy. *Ann Ophthalmol* 1970; 2:664.

Hayasaka S, et al. Leber congenital amaurosis in an infant with Down syndrome. *Ann Ophthalmol* 1992; 24:250-252.

Lambert SR, et al. Concordance and recessive inheritance of Leber congenital amaurosis. *Am J Med Genet 1993; 4:* 275-277.

Leber T. Uber Retinitis Pigmentosa und Angeborene Amaurose. *Arch F Ophthalmol (Berlin)* 1869; 15:1.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Steinberg A, et al. Central nervous involvement in Leber congenital amaurosis. *J Pediatr Ophthalmol Strabismus* 1992; 29:224-227.

Lecithin-Cholesterol Acyltransferase (LCAT) Deficiency 714

General: LCAT enzyme involved in metabolism of cholesterol deficiency; autosomal recessive; rare.

Ocular: Cloudy cornea; diplopia; photophobia; corneal opacities.

Clinical: Autoimmune hyperlipoproteinemic anemia; renal failure; hypertension.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Klein HG, et al. Fish eye syndrome: a molecular defect in the lecithin-cholesterol acyltransferase (LCAT) gene associated with normal alpha-LCAT-specific activity. Implications for classification and prognosis. *J Clin Invest* 1993; 92:479-485.

Vrabec MP, et al. Ophthalmic observation in lecithin cholesterol acyltransferase deficiency. *Arch Ophthalmol* 1988; 106:225-229.

Legg-Perthes Disease (Legg-Calve-Perthes Disease; Legg-Calve-Waldenstrom Syndrome; Coxa Plana; Legg Disease; Legg-Calve Disease) 715

General: Etiology not established; occurs in children between ages 3 and 12 years; more common in boys; unilateral involvement more common than bilateral; retardation of bone age; occasionally familial; rare in blacks; possible role for protein C and S deficiency, thrombophilia, venous hypertension, and hypofibrinolysis in the pathophysiologic mechanism of this disease.

Ocular: Iris processes (pectinate ligaments in the anterior chamber angle).

Clinical: Osteochondrosis of the capitular epiphysis of the femur; limpness associated with muscular spasm.

Glueck CJ, et al. Protein C deficiency, thrombophilia, and hypofibrinolysis: pathophysiologic causes of Legg-Perthes disease. *Pediatr Res* 1994; 35[4 Part 1]:383-389.

Legg AT. The cause of atrophy in joint disease. *Am J Orthop Surg* 1908-1909; 6:84-90.

Lichter PR. Iris processes in 340 eyes. *Am J Ophthalmol* 1969; 68:872-878.

Liu SI, Ho TC. The role of venous hypertension in the pathogenesis of Legg-Perthes disease. A *Clinical* and experimental study. *J Bone Joint Surg Am* 1991; 73:194-200.

Perthes G. Uber Arthritis Deformans Juvenilis. *Dtsch Z Chir* 1910; 107:111-159.

Leigh Syndrome (Subacute Necrotizing Encephalomyelopathy; Infantile Subacute Necrotizing Encephalomyelopathy; Hyperpyruvicemia with Hyper-Alpha-Alaninemia; Gangliosidosis G_{M2} Type 3) 716

General: Autosomal recessive; metabolic disease occurring in infancy and childhood with increased levels of serum lactate, serum pyruvates, blood α-ketoglutarate, and aminoaciduria; course is remittent with early neuro-ophthalmologic manifestations and psychomotor retardation; the later the onset of clinical manifestations, the longer the survival time; acute form in young infants, subacute form in older infants, and chronic course in juveniles; Mutation at nt8993 of mitochondrial DNA has been reported as a common cause of Leigh syndrome; biochemical analysis revealed cytochrome c oxidase deficiency with this condition.

Ocular: Nystagmoid movements or nystagmus; disconjugate ocular movements due to tegmental involvement of brainstem; degrees of visual impairment, depending on pathologic changes involving optic nerves and tracts; optic nerve atrophy; oculomotor palsy; supranuclear gaze palsy; blindness.

Clinical: Spasticity of extremities; ataxia; muscular weakness; hemiparesis; progressive mental deterioration; hearing defects; dysphagia; dyspnea; mild hypotonia; slow development; intermittent abnormal respiratory rhythm; cranial nerve palsies.

Borit A. Leigh's necrotizing encephalomyelopathy: neuro-ophthalmological abnormalities. *Arch Ophthalmol* 1971; 85:438.

Fenichel GM. Subacute necrotizing encephalomyelopathy (Leigh disease). In: Fenichel GM, ed. *Clinical pediatric neurology,* 2nd ed. Philadelphia: WB Saunders, 1993:130-131.

Howard RO, Albert DM. Ocular manifestations of subacute necrotizing encephalomyelopathy (Leigh's disease). *Am J Ophthalmol* 1972; 74:386.

Kalimo H, et al. Familial subacute necrotizing encephalopathy of the adult form (adult Leigh syndrome). *Ann Neurol* 1979; 6:200-206.

Leigh D. Subacute necrotizing encephalomyelopathy in an infant. *J Neurol Neurosurg Psychiatr* 1951; 14:216.

Lombes A, et al. Biochemical and molecular analysis of cytochrome c oxidase deficiency in Leigh's syndrome. *Neurology* 1991; 41:491-498.

Santorelly FM, et al. The mutation at nt8993 of mitochondrial DNA is a common cause of Leigh's syndrome. *Ann Neurol* 1993; 34:827-834.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV* Philadelphia: WB Saunders, 1994:2777.

Wijburg FA, et al. Leigh syndrome associated with a deficiency of the pyruvate dehydrogenase complex results of treatment with a ketogenic diet. *Neuropediatrics* 1992; 23: 147-152.

Leiomyoma 717

General: Rare, benign tumor that arises from smooth muscle; usually well encapsulated.

Ocular: Pigmented tumor of ciliary body; proptosis; distorted pupil; ectropion; iris tumor; glaucoma; cataract; preferential location: ciliary body, peripheral choroid, supraciliary or suprachoroidal space; has a predilection for younger patients and females.

Clinical: Metastases have not been described.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Sevel D, Tobias B. The value of fluorescein iridography with leiomyoma of the iris. *Am J Ophthalmol* 1972; 74: 475-478.

Shields JA, et al. Observations on seven cases of intraocular leiomyoma. The 1993 Byron Demorest Lecture. *Arch Ophthalmol* 1994; 112:521-528.

Sunba MSN, et al. Tumours of the anterior uvea. III. Oxytalan fibers in the differential diagnosis of leiomyoma and malignant melanoma of the iris. *Br J Ophthalmol* 1980; 64:867-874.

Leishmaniasis 718

General: Caused by protozoa *Leishmania.*

Ocular: Conjunctivitis; ulcerative keratitis; ulcerating granulomatous lid lesions; lid edema; interstitial keratitis; subacute focal retinitis; retinal hemorrhage; iridocyclitis; unilateral chronic granulomatous blepharitis.

Clinical: Lesions in spleen, liver, and large intestine; fever; leukopenia; cutaneous lesions on the face.

Bialasiewicz AA, et al. Einseitige chronisch-granulomatose Blepharitis als Leitbefund der orientalischen kutanen Leishmaniase in Deutschland. Oiemsa-Farbung als Schnelldiagnostik und Erstbeschreibungeiner systemischen Therapie mit gamma-Interferon. *Klin Monatsbl Augenheilkd* 1992; 200:219-223.

Duke-Elder S, MacFaul PA. *System of Ophthalmology. vol. XIII.* St. Louis: CV Mosby, 1974.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Lainson R, Shaw JJ. Leishmaniasis of the New World: taxonomic problems. *Br Med Bull* 1972; 28:44.

Lenoble-Aubineau Syndrome (Nystagmus-Myoclonia Syndrome) 719

General: Familial; pathogenesis not known; prevalent in males; manifest during first years of life; X-linked dominant inheritance has been reported in one family.

Ocular: Congenital nystagmus associated with fasciculations of muscles spontaneously elicited by mechanical stimulation or cold.

Clinical: Tremors of head and limbs; myoclonic movements of extremities and trunk; hypospadias; abnormalities of teeth; facial asymmetry; localized edema.

Lenoble E, Aubineau E. Nystagmus-Myoclonia Syndrome: Une Variete Nouvelle de Myoclonie Congenitale Pouvant Etre Hereditaire et Familiale A Nystagmus Constant. *Rev Med (Paris)* 1906; 26:471.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Lens-Iris Diaphram Retropulsion Syndrome 720

General: Associated with small incision phacoemulsification

Ocular: Infusion of fluid into the anterior chamber; posterior displacement of the lens-iris diaphragm; posterior iris bowing; pupil dilatation; ocular discomfort

Clinical: Deep anterior chamber with small incision phacoemulsification

Cionni RJ, Barros MG, Osher RH, et al. Management of lens-iris diaphragm retropulsion syndrome during phacoemulsification. *J Cataract Refract Surg* 2004; 30: 953-956.

Lenz Microphthalmia Syndrome 721

General: X-linked recessive; female carriers.

Ocular: Microphthalmia; microcornea; ocular coloboma; colobomatous microphthalmia.

Clinical: Skeletal abnormalities of vertebral column, clavicles, and limbs; severe renal dysgenesis and hydroureters; dental anomalies; hypospadias and bilateral cryptorchidism; severe speech impairment; shortness of stature; long, cylindrical, and thin thorax; sloping shoulders; flat feet.

Antoniades K, et al. A sporadic case of Lenz microphthalmia syndrome. *Ann Ophthalmol* 1993; 25:342-345.

Herrmann J, Opitz JM. The Lenz microphthalmia syndrome. *Birth Defects* 1969; 5:138-148.

Pagon RA. Ocular coloboma. *Surv Ophthalmol* 1981; 25:223-236.

Leptomeningeal Adhesive Thickening (Chronic Adhesive Arachnoiditis) 722

General: Follows a chronic leptomeningeal infection, trauma, or subarachnoid hemorrhage; insidious onset.

Ocular: Diplopia.

Clinical: Headache; nausea; vomiting; vertigo; epileptic seizures.

Adams RD, Victor M. Diseases of the spinal cord. Chronic adhesive arachnoiditis. In: Adams RD, Victor M, eds. *Principles of Neurology*, 5th ed. New York: McGraw-Hill, 1993: 1099-1100.

Kasper DL, et al., eds. Harrison's *Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005

Cartlidge NE, Al-Hakim M, Bradley WG. Chronic spinal arachnoiditis. In: Bradley WG, ed. *Neurology* in *Clinical Practice*, 2nd ed. Boston: Butterworth-Heinemann, 1995:1819-1820.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Leri Syndrome (pleonosteosis Syndrome; Carpal Tunnel Syndrome) 723

General: Autosomal dominant type of congenital osseous dystrophy; early epiphyseal bone formation of extremities; Morton metatarsalgia syndrome may result; onset in early infancy.

Ocular: Microphthalmia; anophthalmia; oculomotor paralysis; corneal clouding; cataract.

Clinical: Dwarfism (disproportionate); articular deformities; cutaneous deformities; carpal tunnel syndrome (median nerve compression); deformities of thumbs and great toes; laryngeal stenosis.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Leri A. Une Dystrophic Osseuse Generalisee et Hereditaire: la Pleonosteose Familiale. *Presse Med* 1922; 30: 13, and *Bull Soc Med Hop (Paris)* 1921; 45:1228, 1924; 48:25.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Shaw DG. Leri's pleonosteosis [Letter]. *Br J Radiol* l981; 54:819.

Spinner M, Spencer PS. Nerve compression lesions of the upper extremity: a *Clinical* and experimental review. *Clin Orthop* 1970; 104:46.

Lermoyez Syndrome 724

General: Form of Ménière disease; however, hearing acuity improves during the climax of the vestibular attacks; onset in third or fourth decade of life.

Ocular: Nystagmus (spontaneous) directed toward opposite side of involved vestibular system or to the side of the increased tonic state.

Clinical: Dizziness; vertigo; tinnitus; improvement of hearing during vestibular attacks; sweating; nausea; tremor; low tone hearing loss.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Lermoyez M. Le Vertige qui Gait Entendre (Angiospasme Labyrinthique). *Presse Med XXVIIe Annee* 1919.

Schuknecht HF. Further observations on the pathology of Meniere's disease. *Ann Otol* 1962; 71:1039.

Young YH, Wu CH. Electronystagmographic findings in a case of Lermoyez's syndrome. *Auris Nasus Larynx* 1994; 21:118-121.

Leroy Syndrome 725

General: Possible mild increase in mucopolysaccharide excretion.

Ocular: Nasal epicanthic folds; corneal opacities.

Clinical: High, narrow forehead; narrow nasal bridge.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Johnson CC. Epicanthus. *Am J Ophthalmol* 1968; 66:939.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Leukemia 726

General: Acute or chronic blood disorder.

Ocular: Engorgement of conjunctival vessels; papillary hypertrophy; aggregations of tumor cells in conjunctiva, choroid, and orbit; secondary glaucoma; retinal venous engorgement and tortuosity with pronounced constrictions; retinal hemorrhages; retinal detachment; cotton-wool spots; macular edema; papilledema; optic atrophy; optic neuritis; paralysis of extraocular muscles; hypopyon; vitreous opacities; retinal sea fans; perilimbal subconjunctival infiltrates; corneal leukemic infiltration (rare); shallow serous retinal detachments; hyphema; iris neovascularization; central retinal vein occlusion; vitreous infiltrates.

Clinical: Frequent involvement of central nervous system; intracranial hemorrhage; thrombocytopenia; rising white cell count.

Brown GC, et al. Leukemic optic neuropathy. *Int Ophthalmol* 1981; 3:11-16.

Currie JN, et al. Optic neuropathy in chronic lymphocytic leukemia. *Arch Ophthalmol* 1988; 106:654-660.

Font RL, et al. Acute monocytic leukemia recurring as bilateral perilimbal infiltrates. *Ophthalmology* 1985; 92:1681-1685.

Leveille AS, Morse PH. Platelet-induced retinal neovasculanzation in leukemia. *Am J Ophthalmol* 1981; 91:640-644.

Murtha TJ. Hematologic disorders: leukemia, dysproteinemia, and anemia. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994:2986.

Robb RM, et al. A pathological study of eye involvement in acute leukemia of childhood. *Trans Am Ophthalmol Soc* 1978; 76:90-101.

Rodgers R, et al. Ocular involvement in congenital leukemia. *Am J Ophthalmol* 1986; 101:730-732.

Swartz M, Schumann GB. Acute leukemic infiltration of the vitreous diagnosed by pars plana aspiration. *Am J Ophthalmol* 1980; 90:326-330.

Lewis Syndrome (Tuberoserpiginous Syphilid of Lewis) 727

General: Lesions more common on nose and ears but may involve eyelids primarily; clinical manifestations are similar to lupus vulgaris.

Ocular: Lesions most frequently involve the lower eyelids; tear ducts may become involved, with lesions of mouth and nose as a direct extension or via the lymphatic route; granulomatous conjunctival lesions, usually an extension of involvement of buccal and nasal mucosa; iridocyclitis may occur; corneal ulcers as seen in tuberculous granulomatosis.

Clinical: Skin lesions similar to those of the face may be seen on other parts of extremities or trunk.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Lhermitte-Levy Syndrome (Hallucinosis-Red Nucleus; Lhermitte-Delthil-Gamier Syndrome) 728

General: Lesion of unknown nature in the upper portion of peduncle and subthalamic region; occurs in the elderly.

Ocular: Visual hallucinations.

Clinical: Auditory hallucinations; paralysis; rhythmic trembling; stroke.

Lhermitte MJ, Delthil G. Syndrome Contrelateral du Noyau Rouge avec Hallucinations Visuelles et Auditives. *Rev Neurol* 1938; 70:623-628.

Lhermitte MJ, Levy G. Phenomenes d'Allucinose Chez une Malade Presentant une Torsion et une Contracture Athetoides Intentionnelles du Bras. *Soc Neurol* 1931; 7:644.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Lichen Planus 729

General: Conjunctival disorder associated with dermatologic disorder; disappears spontaneously.

Ocular: Conjunctivitis; cicatrizing conjunctivitis; keratin plaque on bulbar conjunctiva.

Clinical: Grayish-white papules; oral lesions may precede skin lesions.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Neumann R, et al. Immunohistopathologic features and therapy of conjunctival lichen planus. *Am J Ophthalmol* 1993; 115:494-500.

Roy FH. *Ocular Differential Diagnosis*, 7th ed. Philadelphia: Lippincott Williams & Wilkins, 2002.

Lignac-Fanconi Syndrome (Fanconi-Lignac Syndrome; Cystinosis Syndrome; Cystine Storage-Aminoaciduria-Dwarfism Syndrome; Renal Rickets; Nephropathic Cystinosis) 730

General: Autosomal recessively inherited storage disorder in which nonprotein cystine accumulates within cellular lysosomes; occurs primarily in children; prognosis in children with renal tubular insufficiency and dwarfism poor, with survival past age 10 years rare without renal transplant.

Ocular: Cystine crystals located in conjunctiva, cornea, sclera, iris, ciliary body, lens, and perhaps choroid; general clouding of cornea caused by dense deposition of cystine crystals; pupillary block glaucoma; photophobia; band keratopathy; posterior synechiae with thickened stroma of iris; decreased visual function; patchy retinopathy; visual field constriction

Clinical: Fanconi syndrome with rickets; dwarfism; glomerular dystrophy; renal failure; oral motor dysfunction.

Cogan DG, et al. Cystinosis in an adult. *J Am Med Assoc* 1957; 164:394.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Jonas AJ, et al. Nephropathic cystinosis with central nervous system involvement. *Am J Med* 1987; 83:966-970.

Kaiser-Kupfer MI, et al. Long-term ocular manifestation in nephropathic cystinosis. *Arch Ophthalmol* 1986; 104: 706-711.

Kenyon KR, Sensenbrenner JA. Electron microscopy of cornea and conjunctiva in childhood cystinosis. *Am J Ophthalmol* 1974; 78:68-76.

Lignac GOE. Present status of knowledge of cystine metabolism. *Ned Tijdschr Geneeskd* 1924; 1:2998.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Tsilou ET, Rubin BI, Reed G, et al. Nephropathic cystinosis. *Ophthalmology* 2006; 113: 1002-1009

Wan WL, et al. Pupillary-block glaucoma associated with childhood cystinosis. *Am J Ophthalmol* 1986; 101: 700-705.

Linear IgA Disease 731

General: Bullous dermatosis with pruritic urticarial lesions with overlying vesicles or bullae; skin lesions heal without scarring; homogeneous deposition of immunoglobulin A (lgA) at the dermal-epidermal junction and, rarely, deposition of other immunoglobulin present; heterogeneous disease with regard to its clinical features, target antigens, and immunogenetics; association with HLA-B8, DR3, Cw7, and the linked rare tumor necrosis factor-α allele; may be induced by amiodarone.

Ocular: Chronic conjunctivitis; subconjunctival fibrosis; symblepharon; chronic progressive conjunctival cicatrization.

Clinical: Recurrent blistering skin disorder consisting of urticarial macules and plaques with vesicular eruptions on trunk and extremities; subepidermal vesiculation.

Aultbrinker EA, et al. Linear IgA disease. *Ophthalmology* 1988; 95:340-343.

Foster CS, et al. Immunosuppressive therapy for progressive ocular cicatricial pemphigoid. *Ophthalmology* 1982; 89: 341-353.

Primka EJ III, et al. Amiodarone-induced linear IgA disease. *J Am Acad Dermatol* 1994; 31:809-811.

Wojnarowska F, et al. Linear IgA disease: a heterogeneous disease. *Dermatology* 1994; 189[Suppl 1]:52-56.

Linear Nevus Sebaceus of Jadassohn (Nevus Sebaceus of Jadassohn; Jadassohn-Type Anetoderma; Organoid Nevus syndrome; Sebaceus nevus syndrome) 732

General: Skin nevus caused by failure of separation of skin appendages from adjacent epithelium during the third month of gestation.

Ocular: Proptosis; epibulbar lipodermoids; colobomata of eyelids, iris, and choroid; antimongoloid fissures; ocular motor palsies; nystagmus; teratomas of orbit and aberrant lacrimal glands; corneal vascularization; vision defects; conjunctival dermolipomas; choristomas of conjunctiva, sclera; corneal vascularization/opacification; colobomas of uvea, retina, optic disk, and lids; optic nerve hypoplasia; microphthalmia; anophthalmia; hemangioma of the sclera/conjunctiva.

Clinical: Circumscribed lesions of the face and scalp with excessively large sebaceous glands; papillomatous epidermal hyperplasia; seizures; skeletal abnormalities, particularly in skull; failure to thrive; convulsion; mental retardation.

Brenner S, et al. Jadassohn-type anetoderma m association with keratoconus and cataract. *Ophthalmologica* 1977; 174:181-184.

Garella M, Ghosh B, Thakar M, et al. Linear nevus sebaceous syndrome with bilateral complex limbal choristomas and extraocular muscle involvement. *Ann Ophthalmol* 2004; 36: 59-62

Haslam RHA, Wirtschafler JD. Unilateral external oculomotor nerve palsy and nevus sebaceus of Jadassohn. *Arch Ophthalmol* 1972; 87:293.

Jadassohn J. Bemerkungen fur Histolgre der System Atisirten Naeviund uber "Talgdrusen-Naevi." *Arch Dermatol Syphil* 1895; 33:355.

Katz B, et al. Optic nerve hypoplasia and the syndrome of nevus sebaceous of Jadassohn. *Ophthalmology* 1987; 94: 1570.

Wilkes SR, et al. Ocular malformation in association with ipsilateral facial nevus of Jadassohn. *Am J Ophthalmol* 1981; 92:344-352.

Lipodystrophy (Kobberling-Dunnigan Syndrome) 733

General: Disturbance of the fat metabolism; autosomal dominant; affects females predominantly; occurs at puberty.

Ocular: Enophthalmos; lack of lid apposition; choroidal atrophy; optic disk pallor; corneal opacity.

Clinical: Progressive symmetrical loss of subcutaneous fat in upper part of body, including face and orbits; fat accumulation of neck, shoulders, buffalo hump, and genitalia; hyperthyroidism; lipoatrophic diabetes; hepatosplenomegaly; acanthosis nigricans; hyperlipemia; lean muscular limbs; phlebectasia; insulin resistance; hyperglycemia; type IV lipoproteinemia.

Dunnigan MG, et al. Familial lipoatrophic diabetes with dominant transmission: a new syndrome. *Q J Med* 1974; 43: 33-48.

Kobberling J, et al. Lipodystrophy of the extremities: a dominantly inherited syndrome associated with lipoatrophic diabetes. *Humangenetik* 1975; 29:111-120.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Liposarcoma 734

General: Aggressive malignant neoplasms of lipogenic cells; occurs at any age, but rarely before age 30 years and most commonly in the fifth decade; occurs almost exclusively in adults and is found most often in the thigh or retroperitoneum.

Ocular: Paresis of extraocular muscle; proptosis; orbital liposarcoma; eyelid edema.

Clinical: Neoplasms of deeper soft tissues; metastasis to lungs, liver, lymph nodes, and periosteum.

Enterline HT, et al. Liposarcoma: a clinical and pathological study of 53 cases. *Cancer* 1960; 13:932-950.

Favrot SR, et al. Orbital liposarcoma. *Otolaryngol Head Neck Surg* 1994; 111:111-115.

Henderson JW. *Orbital tumors. 2nd ed.* New York: Brian C. Decker, 1980:253-258.

Springield D. Liposarcoma. *Clin Orthop Rel Res* 1993; 289:50-57.

Lissencephaly Syndrome (Miller-Dieker Syndrome) 735

General: Autosomal recessive; consanguinity; association with deletion of the LIS1 gene located at chromosome 17p13.

Ocular: Hypertelorism.

Clinical: Microcephaly; small mandible; bizarre facies; failure to thrive; retarded motor development; mental retardation; dysphagia; decorticated and decerebrate postures; polydactyly; malformations of brain, heart, kidneys, and other organs; spastic paraplegia; agyri-apachygyria; inverted gray-to-white matter ratio; absence of white-gray interdigitations; hypoplastic brainstem; characteristic facial dysmorphism.

Bobyns WB, et al. Lissencephaly. A human brain malformation associated with deletion of the LIS1 gene located at chromosome 17p13. *JAMA* 1993; 270:2838-2842.

Dobyns WB, et al. The Miller-Dieker syndrome: lissencephaly and monosomy. *J Pediatr* 1983; 102:552-558.

Kuchelmeister K, et al. Neuropathology of lissencephalies. *Childs Nerv Syst* 1993; 9:394-399.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Listerellosis (Listeriosis) 736

General: Caused by Gram-positive bacillus *Listeria monocytogenes.* High mortality among pregnant women, their fetuses, and immunocompromised persons with symptoms of abortion, neonatal death, septicemia, meningitis, brain abscesses, endocarditis.

Ocular: Conjunctivitis; keratitis; corneal abscess and ulcer; blepharitis; uveitis; endophthalmitis; cataract; secondary glaucoma.

Clinical: Vomiting; cardiorespiratory distress; diarrhea; hepatosplenomegaly; maculopapular skin lesions.

Bagnarello AG, et al. Listeria monocytogenes endophthalmitis. *Arch Ophthalmol* 1977; 95: 1004-1005.

Berenguer J, et al. Listenosis in patients infected with human immunodeficiency virus. *Rev Infect Dis* 1991; 13: 115-119.

Farber JM, Peterkin PI. Listeria monocytogenes, a food-borne pathogen. *Microbiol Rev* 1991; 55:476-511.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Manschot WA. Congenital listeriosis with ocular involvement. *Ophthalmologica* 1971; 162: 167-172.

Little Syndrome (Nail-Patella Syndrome; Hereditary Osteo-Onycho-Dysplasia; HOOD Syndrome) 737

General: Inherited as autosomal dominant; affects males and females equally.

Ocular: Hypertelorism; ptosis; epicanthus; microcornea; keratoconus; sclerocornea; cataract; microphakia; light pigmentation of iris root with dark pigmented "clover-leaf" spots, referred to as the Lester line, not seen in all cases.

Clinical: Absent or hypoplastic patella; hypoplastic or dislocated head of radius; exostosis of skull bones; bilateral horns of iliac crests; longitudinal ridging of fingernails; glomerulonephritis; renal involvement; bilateral antecubital pterygia; arthrogryposis; disorder has been mapped to the long arm of chromosome 9; sensorineural hearing loss.

Campeau E, et al. Linkage analysis of the nail-patella syndrome. *Am J Hum Genet* 1995; 56:243-247.

Chan PC, et al. Living-related renal transplantation in a patient with nail-patella syndrome. *Nephron* 1988; 50: 164-166.

Fenske HD, Spitalny LA. Hereditary osteo-onychodysplasia. *Am J Ophthalmol* 1970; 70:604.

Flickinger RR, Spivey BE. Lester's line in hereditary osteo-onychodysplasia. *Arch Ophthalmol* 1969; 82:700.

Guidera KJ, et al. Nail-patella syndrome: a review of 44 orthopaedic patients. *J Pediatr Orthop* 1991; 1:737-742.

Hussain SS, Hope GA. Sensorineural hearing loss and nail patella syndrome. *Arch Otolaryngol Head Neck Surg* 1994; 120:674-675.

Little EM. Congenital absence or delayed development of the patella. *Lancet* II 1897:781-784.

Rizzo R, et al. Familial bilateral antecubital pterygia with severe renal involvement in nail-patella syndrome. *Clin Genet* 1993; 44:1-7.

Yakish SD, Fu FH. Long-term follow-up of the treatment of a family with nail-patella syndrome. *J Pediatr Orthop* 1983; 3:360-363.

Locked-In Syndrome 738

General: Usually caused by extensive pontine hemorrhage; awake but paralyzed patient; unable to communicate following basilar artery occlusion; trauma.

Ocular: Ocular bobbing; bilateral paresis of horizontal gaze; spared vertical eye movements, and hearing.

Clinical: Paralysis of all four extremities and the lower cranial nerves without interference with consciousness.

Keane JR. Locked-in syndrome after head and neck trauma. *Neurology* 1986; 36:80.

Larmand P, et al. Abnormal vertical eye movements in the locked-in syndrome. *Ann Neurol* 1982; 11:100-102.

Tyssen CC, Terbruggen JP. Locked-in syndrome associated with ocular bobbing. *Acta Neurol* 1986; 73:444-446.

Lockjaw (Tetanus) 739

General: Acute infectious disease affecting nervous system; causative agent is *Clostridium tetani;* bacteria enters body through a puncture wound, abrasion, cut, or burn.

Ocular: Chemosis; keratitis; nystagmus; uveitis; corneal ulcer; cellulitis of orbit; hypopyon; panophthalmitis; pupil paralysis; pseudoptosis; blepharospasm; paralysis of third or seventh nerve; may occur following perforating ocular injuries.

Clinical: Severe muscle spasms; dysphagia; trismus; facial palsy; muscle stiffness; irritability.

Benson WH, et al. Tetanus prophylaxis following ocular injuries. *J Emerg Med* 1993; 11:677-683.

Bettman JW. The prophylaxis of tetanus. *Am J Ophthalmol* 1963; 56:806-808.

Eckmann L. *Tetanus: prophylaxis and therapy.* New York: Grune & Stratton, 1963.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WE Saunders, 2000.

Saltissi S, et al. Ophthalmoplegic tetanus. *Br Med J* 1976; 1:437.

Loffler Syndrome (Eosinophilic Pneumonitis) 740

General: Etiology unknown, but such considerations as drug hypersensitivity, parasites, mycoses, and periarteritis nodosa have been advanced; eosinophilia up to 80%; condition self-limited and benign; may occur after using crack cocaine, after administration of medications such as minocycline, or as an idiopathic disorder.

Ocular: Endophthalmitis; retinal infarction with hemorrhages and exudates.

Clinical: Dry cough; shortness of breath; increased body temperature; weight loss; malaise; anorexia; fever; dyspnea; pleural rales; pericardial effusion; prolonged expiration; wheezing.

Bando T, et al. Minocycline-induced pneumonitis with bilateral hilar lymphadenopathy and pleural effusion. *Int Med* 1994; 33:177-179.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Loffier W. Zur Differenatial Diagnose der Lungeninfiltrierungen: Uber fluchtige Succedaninfiltrate (mit Eosinophilie). *Beitr Klin Tuberkulose* 1032; 79:338.

Nadeem S, et al. Loffier's syndrome secondary to crack cocaine. *Chest* 1994; 105:1599-1600.

Nishio M, et al. Idiopathic acute eosinophilic pneumonia. *Int Med* 1992; 31:1139-1143.

Warnock ML, et al. Chronic eosinophilic pneumonia: a manifestation of allergic aspergillosis. *Am J Clin Pathol* 1974; 62:73.

Longfellow-Graether Syndrome 741

General: Rare; etiology unknown.

Ocular: Grossly dilated retinal veins; intermittent attacks of uniocular blindness.

Clinical: None.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Roy FH. *Ocular Differential Diagnosis*. 7th ed. Philadelphia: Lippincott Williams & Wilkins, 2002.

Lost Lens Syndrome 742

General: Occurs when the intraocular lens is completely dislocated into the vitreous cavity, caused by luxation of the implant through a zonular disinsertion or an unrecognized opening in the posterior capsule or trauma.

Ocular: Decreased visual acuity; retinal detachment; cystoid macular edema.

Brockman EB, et al. Visual disability resulting from a dislocated intraocular lens. *J Cataract Refract Surg* 1993; 19: 312-313.

Smith SG, Lindstrom RL. Malpositioned posterior chamber lenses: etiology, prevention, and management. *Am Intraocular Implant Soc J* 1985; 11:584-591.

Stark WJ, et al. Management of posteriorly dislocated intraocular lenses. *Ophthalmic Surg* 1980; 11:495-497.

Louis-Bar Syndrome (Cephalo-Oculocutaneous Telangiectasis; Ataxia-Telangiectasia Syndrome) 743

General: Autosomal recessive; thymic abnormality leading to an immunologic deficiency has been suggested as the cause; chromosomal translocations are found in 5% to 10% of peripheral T cells from most patients.

Ocular: Rapid blinking in upward gaze; "pseudo-ophthalmoplegia"; fixational nystagmus (see Roth-Bielschowsky Syndrome) halting intermittently, mainly on lateral and upward gaze; on head turning, eyes are involuntarily directed to opposite side with slow return to the primary position; ocular motor apraxic movement; loss of optokinetic responses; poor convergence ability;

telangiectasias of anterior segment and sclera; fine, bright, symmetrical red streaks of the temporal and nasal conjunctiva (usually first seen at age 4 to 6 years); prominent veins in canthal regions of conjunctiva.

Clinical: Progressive cerebellar ataxia; slow and scanning speech; mental retardation; cutaneous telangiectasis and fine spots of pigmentation; recurrent sinopulmonary infections; hypotonia; diminished growth; cutaneous telangiectasis of ears, cheeks, and antecubital space; deficiency of IgA; lymphoreticular malignancy; high cancer risk in children with progressive cerebellar ataxia most commonly lymphoma (B-cell type) or leukemias.

Adams RD, Victor M. Ataxia-telangiectasia. In: *Principles of neurology,* 5th ed. New York: McGraw-Hill, 1993: 821-822.

Fenichel GM. Ataxia-Telangiectasia. In: Fenichel GM, ed. *Clinical pediatric neurology.* 2nd ed. Philadelphia: WB Saunders, 1993:238-239.

Font RL, Ferry AP. The phacomatoses. *Int Ophthalmol Clin* 1972; 12:1.

Gatti RA. Ataxia-telangiectasia. *Dermatol Clin* 1995; 13:1-6.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Harding AE. Cerebellar and spinocerebellar disorders: ataxia-telangiectasia. In: Bradley WG, et al., eds. *Neurology in Clinical Practice. vol. II,* 2nd ed. Boston: Butterworth-Heinemann, 1995:1778-1780.

Louis-Bar MME. Sur un Syndrome Progressif Comprenant des Telangiectasies Capillaires Cutanee et Conjonctivales Symetriques, a Disposition Naevoide et des Troubles Cerebelleux. *Confinia Neurol* 1941; 4:32.

Louping III Syndrome 744

General: Tick-borne disease of sheep; occurs in Britain.

Ocular: Difficulty in blinking; retrobulbar neuritis; transient diplopia.

Clinical: Cerebellar ataxia; mild encephalitis; weak facial muscles.

Duke-Elder S, MacFaul PA. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Lowe Syndrome (Oculo-Cerebro-Renal Syndrome) 745

General: Essential enzyme or protein abnormality is unknown; sex-linked recessive trait (male incidence only); onset in early infancy.

Ocular: Nystagmus; congenital glaucoma; miotic pupils; no pupillary reaction; ectropion uveae; malformation of the anterior chamber angle and of the iris; Schlemm canal may be absent with imperfect angle cleavage; blue sclera; cloudy cornea; cataracts; megalocornea; corneal dystrophy; buphthalmos; microphthalmos; microphakia; mydriasis; strabismus; lens punctate cortical opacities.

Clinical: Mental, psychomotor, and growth retardation; aminoaciduria; albuminuria; glycosuria; renal tubular acidosis; rickets; osteomalacia; muscular hypotony; hyporeflexia; hyperactivity with bizarre choreoathetoid movements and screaming.

Bartsocas CS. Etiology of ocular manifestations in Lowe's syndrome. *Ann Ophthalmol* 1970; 2:368.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fagerholm P, et al. Lowe's oculocerebrorenal syndrome-variation in lens changes in the carrier state. *Acta Ophthalmol* 1991; 69:102-104.

Fisher NF. Oculocerebrorenal syndrome of Lowe. *Arch Ophthalmol* 1967; 77:642.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Lubarsch-Pick Syndrome (Primary Amyloidosis; Idiopathic Amyloidosis; Amyloidosis) 746

General: Rare condition of unknown etiology; inherited as a dominant trait, with male preponderance; characterized by amyloid accumulation in muscles and in gastrointestinal and genitourinary tracts.

Ocular: Internal and external ophthalmoplegia; diminished lacrimation; amyloid deposits in conjunctival, episcleral, and ciliary vessels; vitreous opacities; amyloid deposits in the corneal stroma; retinal hemorrhages and perivascular exudates; paralysis of extraocular muscles; pseudopodia lentis; strabismus fixus convergens; keratoconus.

Clinical: Peripheral neuropathy (extremities); heart failure; defective hepatic and renal functions with hepatosplenomegaly; waxy skin lesions; muscular weakness (progressive); multiple myeloma; hoarseness; chronic gastrointestinal symptoms.

Biswas J, et al. Primary nonfamilial amyloidosis of the vitreous. A light microscopic and ultrastructural study. *Retina* 1992; 12:251-253.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Goebel MH, Friedman AM. Extraocular muscle in idiopathic primary amyloidosis. *Am J Ophthalmol* 1971; 71:1121.

Lubarsch O. Zur Kenntnis Ungewohnlicher Amyloidablagerungen. *Virchows Arch Pathol Anat* 1929; 271:867.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Sharma P, et al. Strabismus fixus convergens secondary to amyloidosis. *J Pediatr Ophthalmol Strabismus* 1991; 28: 2367.

Wong VG, McFarlin DE. A case of primary familial amyloidosis. *Arch Ophthalmol* 1967; 78:208.

Lyme Disease 747

General: Caused by tick bite; symptoms resolve after treatment.

Ocular: Keratitis may occur up to 5 years after the first episode; diplopia; photophobia; ischemic optic neuropathy; iritis; panophthalmitis; conjunctivitis; exudative retinal detachment; choroiditis; vitreitis; multiple cranial nerve palsies; association with acute, posterior, multifocal, placoid, pigment epitheliopathy; branch retinal artery occlusion.

Clinical: Arthritis; increased intracranial pressure; effusion of knees; swelling of wrists.

Bialasiewicz AA. Bilateral diffuse choroiditis and exudative retinal detachments with evidence of Lyme disease. *Am J Ophthalmol* 1988; 105:419-422.

Carvounis, PE, Mehta AP, Geist CE: Orbital myositis associated with *borrelia burgdorferi* (Lyme disease) infection. *Ophthalmology* 2004; 111: 1023-1028.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000,

Lesser RL. Ocular manifestations of Lyme disease. *Am J Med* 1995; 98(4A):60S-62S.

MacDonald AB. Lyme disease: a neuro-ophthalmologic view. *J Clin Neuro-Ophthalmol* 1987; 7: 185.

Niutta A, et al. Monolateral chorioretinitis with multiple foci in one case of Lyme disease. *Ann Ophthalmol* 1993; 25: 257-261.

Schechter SL. Lyme disease associated with optic neuropathy. *Am J Med* 1986; 81:143.

Wolf MD, et al. Acute, posterior, multifocal, placoid, pigment epitheliopathy and Lyme disease. *Arch Ophthalmol* 1992; 110:750,

Lymphadenosis Benigna Orbitae 748

General: Onset from age 18 to 88 years; localized inflammatory process of undetermined origin; duration usually 2 to 12 months.

Ocular: Exophthalmos; well-circumscribed painless swelling around the eyes; glaucoma; restricted eye movements.

Clinical: Malignant tumor; insect bites; lymphadenosis benigna cutis.

Bafverstedt B, et al. Lymphadenosis benigna orbitae. *Acta Ophthalmol* 1956; 34:367-370.

Levin PS, Dutton JJ. A 20-year series of orbital exenteration. *Am J Ophthalmol* 1991; 112:496-501.

Orlowski WJ, Korobowicz J. The lymphadenoglaucoma, *Am J Ophthalmol* 1961; 52:101-106.

Lymphangioma 749

General: Poorly circumscribed infiltrating lesions consisting of lymphatics/dysplastic blood vessels; occurs predominantly in children and young adults.

Ocular: Conjunctival hemorrhages; cellulitis of lid; ptosis; exophthalmos; amblyopia; astigmatism; extraocular muscle imbalance; optic disk edema; retinal striae.

Clinical: Benign tumors of the lymph system.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Iliff WJ, Green WR, Orbital lymphangiomas. *Ophthalmology* 1979; 86:914-929.

Reese AB. Expanding lesions of the orbit. *Trans Ophthalmol Soc UK* 1971; 91:85-104.

Lymphedema 750

General: Abnormal accumulation of lymph in the extremities; occurs from multiple causes.

Ocular: Conjunctival chemosis; ectropion; ptosis; strabismus; hyperpigmentation of eyelids; chorioretinal dysplasia; distichiasis; eyelid lymphedema.

Clinical: Abnormal lymphatic drainage; painless swelling; fibrosis of skin and subcutaneous tissues; skin becomes thickened, brown, multiple papillary projections (lymphostatic verrucosis); microcephaly; lymphedema.

Angle B. et al. Microcephaly, lymphedema, and chorioretinal dysplasia: report of two additional cases. *Am J Med Genet* 1994; 53:99-101.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005,

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Creager MA, Dzau VJ. Vascular diseases of the extremities: lymphatic disorders. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:1135-1143,

Kolin T, et al. Hereditary lymphedema and distichiasis. *Arch Ophthalmol* 1991; 109:980-981,

Lymphocytic Choriomeningitis (Aseptic Meningitis) 751

General: Virus of which natural host is house mouse; more common in late fall and winter; usually benign.

Ocular: Palpebral edema; optic neuritis; ocular motor palsies; pupillary and accommodative pareses; strabismus; diplopia; conjunctival injection.

Clinical: Fever; meningeal irritation; headaches; confusion; coma; stiffness of neck; Babinski sign; may occur early in the course of human immunodeficiency virus infection; syndrome is considered to be usually viral in origin, with enteroviruses accounting for most cases, but rare bacterial organisms, such as *Mycobacterium tuberculosis*, *Leptospira* sp, *Brucella* sp, *Borrelia burgdorferi*, and others, may cause aseptic meningitis; drug-induced aseptic meningitis also should be considered.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Chaudrhry HJ, Cunha BA. Drug-induced aseptic meningitis. Diagnosis leads to quick resolution. *Postgrad Med* 1991; 90:65-70.

Duke-Elder S, MacFaul PA. *System of Ophthalmology, vol. XIII* St. Louis: CV Mosby, 1974.

Nelsen S, et al. The aseptic meningitis syndrome. *Am Fam Physician* 1993; 48:809-815.

Newton HB. Common neurologic complications of HIV-l infection and AIDS. *Am Fam Physician* 1995; 51:387-398.

Lymphogranuloma Venereum (Nicolas-Favre Disease; Tropical Bubo LGV; Lymphogranuloma Inguinale) 752

General: Venereally transmitted infection caused by chlamydia.

Ocular: Conjunctivitis; chronic lid edema; keratitis; pannus; corneal ulcer; keratoconus; episcleritis; uveitis; tortuosity of retinal vessels; retinal hemorrhages.

Clinical: Enlargement of inguinal lymph nodes; lymphadenitis; lymphogranuloma.

Buus DR, et al. Lymphogranuloma venereum conjunctivitis with a marginal corneal perforation. *Ophthalmology* 1988; 95:799-802.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duke-Elder S, ed. *System of Ophthalmology, vol. VX* St. Louis: CV Mosby, 1976:94.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Lanchard TJ, Mabey DC. Chlamydial infections. *Br J Clin Practice* 1994; 48:201-205.

Lymphoid Hyperplasia (Reactive Lymphoid Hyperplasia; Lymphoid Tumors; Malignant Lymphoma; Pseudolymphoma; Pseudotumor; Burkitt Lymphoma; Neoplastic Angioendotheliomatosis) 753

General: Occurs in tropical Africa; young children; idiopathic orbital inflammation; systemic disease is rarely associated but occasionally occurs with either vasculitis or lymphomas; etiology of Burkitt lymphoma currently includes three factors: (i) Epstein-Barr virus, (ii) malaria, and (iii) chromosomal translocations activating the *c-myc* oncogene, which induces uncontrolled B-cell proliferation.

Ocular: Proptosis; extraocular motility disturbances; lesions of orbit, lacrimal gland, conjunctiva, and uvea; cortical blindness; retinal artery occlusion; retinal vascular and pigment epithelial alterations; vitreitis.

Clinical: Maxillary tumor; Epstein-Barr virus; cranial neuropathy.

Brooks HL, et al. Orbital Burkitt's lymphoma in a homosexual man with acquired immune deficiency. *Arch Ophthalmol* 1984; 102:1533-1537.

Cheung MK, et al. Diagnosis of reactive lymphoid hyperplasia by chorioretinal biopsy. *Am J Ophthalmol* 1994; 118: 457-462.

Liesegang TJ. Ocular adnexal lymphoproliferative lesions. *Mayo Clin Proc* 1993; 68:1003-1010.

de The G. The etiology of Burkitt's lymphoma and the history of the shaken dogmas. *Blood Cells* 1993; 19:667-673.

Donoso LA. Meningeal carcinomatosis secondary to malignant lymphoma (Burkitt's pattern). *J Pediatr Ophthalmol Strabismus* 1981; 18:48-50.

Elner VM, et al. Neoplastic angioendotheliomatosis. *Ophthalmology* 1986; 93:1237-1245.

Gaidano G, Dalla-Favera R. Biologic and molecular characterization of non-Hodgkin's lymphoma. *Curr Opin Oncol* 1993; 5:776-784.

Jakobiec FA, et al. Clinicopathologic characteristics of orbital lymphoid hyperplasia. *Ophthalmology* 1979; 86: 948-966.

Macular Halo Syndrome 754

General: Probably variant of Niemann-Pick disease; major differences are the ocular lesion described as macular halo with a granular appearance instead of the classic cherry-red spot and the lack of major visual symptoms.

Ocular: Crystalloid opacities of foveolae; granular macula; macular halo.

Clinical: Hepatosplenomegaly; hyperlipidemia; histiocytes of bone marrow, spleen, and liver.

Cogan DG, Tedermann DD. Retinal involvement with reticuloendotheliosis of unclassified type. *Arch Ophthalmol* 1964; 71:489-491.

Cogan DG, et al. Macular halo syndrome. *Arch Ophthalmol* 1983; 101:1698-1700.

Macular Dystrophy 755

General: Sex-linked; macular dystrophy of fundus.

Ocular: Posterior pole changes; marked decrease in visual acuity; congenital stationary night blindness; color blindness; vitreotapetoretinal dystrophy; choroidal dystrophy; senile macular choroidal degeneration; retinoschisis; prognosis for retaining functional visual acuity is good.

Clinical: Lipidoses; neurologic disorders.

Forsius H, et al. X-linked hereditary retinoschisis. *Br J Ophthalmol* 1962; 46:678.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Macular Dystrophy, Concentric Annuli (Bull's-Eye Macular Dystrophy) 756

General: Autosomal dominant; no male-to-male transmission observed.

Ocular: Dyschromatopsia; foveal hyperpigmentation; perifoveal hyperpigmentation; perifoveal circular pigment epithelial atrophy.

Clinical: None.

Coppeto J, Ayazi S. Annular macular dystrophy. *Am J Ophthalmol* 1982; 93:279-284.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Macular Dystrophy ectodermal Dysplasia and Ectrodactyly (EEM syndrome) 757

General: Associated with abetaloproteinemia

Ocular: Macular dystrophy; hypotroichosis; vitreous hemorrhage; persistant hyloid artery

Cekic O, Totan Y, Batman C: Traumatic vitreous hemorrhage from a persistant hyloid artery. *J Pediatr Ophthalmol Strabismus* 2000; 37:117-118.

Spraul CW, Giorrniklaus HR: Vitreous hemorrhage. *Surv Ophthalmol* 1997; 42:3-39

Macular Dystrophy, Fenestrated Sheen Type 758

General: Autosomal dominant; progressive; onset in the sixth decade.

Ocular: Yellowish retractile sheen in sensory retina at the macula; red fenestrations present within sheen; hypopigmentation of retinal pigment epithelium.

Clinical: None.

Fishman GA, et al. Dominantly inherited cystoid macular edema. *Ann Ophthalmol* 1979; 11:21-27.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Sneed SR, Sieving PA. Fenestrated sheen macular dystrophy. *Am J Ophthalmol* 1991; 112:1.

Macular Edema, Cystoid 759

General: Autosomal dominant; edema due to leaking perimacular capillaries.

Ocular: Retinal capillary leakage all over posterior pole of the eye; whitish punctate deposits in vitreous; hyperopia; "beaten bronze" atrophy to macula; strabismus.

Clinical: Patients show cystoid macular edema at a young age with gradual progressive decrease in visual acuity starting between the first and the fourth decades of life.

Deutman FA, et al. Dominantly inherited cystoid macular edema. *Am J Ophthalmol* 1976; 82:540-548.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Reichel E, Sandberg MA. Hereditary macular degeneration. In: Albert DM, Jakobiec FA. eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994: 1250.

Mad Hatter Syndrome 760

General: Chronic mercury intoxication; symptoms seldom improve regardless of treatment.

Ocular: Constricted visual fields; lens opacities.

Clinical: Anorexia; peripheral neuritis; ataxia; hearing impairment; hyperreflexia; progressive mental depression; tremor; insomnia; fatigue; irritability; lethargy; hallucinations; shyness; withdrawal; gingivitis; teeth loosening.

Kaplan HI, Sadock B. *Comprehensive Textbook of Psychiatry,* 2nd ed. Baltimore: Williams & Wilkins, 1984.

Kark RAP, et al. Mercury poisoning and its treatment with N-acetyl-D-L penicillamine. *N Engl J Med* 1971; 185:10.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Streeten BW Pathology of the lens. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:2210.

Maffucci Syndrome (Multiple Enchondromatosis; Progressive Dyschondroplasia and Multiple Hemangiomas; Kast Syndrome; Osteochondromatosis) 761

General: Rare; etiology unknown; no hereditary factor; manifest in ages I to 5 years; characterized by numerous cartilage tumors involving mainly small bones of hands and feet; malignant transformation common; no simple mendelian inheritance.

Ocular: Hemangiomas of lid and retina.

Clinical: Multiple enchondromas with secondary bony deformities (dyschondroplasia of Ollier); chondrosarcomas; multiple hemangiomas and phlebolithiasis; orthostatic hypotension (depending on extent and size of hemangiomas); frequent fractures following minimal trauma; precocious pseudopuberty.

Elmore SM, Cantrell WC. Maffucci's syndrome. *J Bone Joint Surg* 1966; 48:1607-1613.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Maffucci A. Di un Caso di Encondroma ad Angioma Multiple. *Movimento Medchir* 1881; 3:399.

Vaz RM, Turner C. Ollier disease associated with ovarian juvenile granuloma cell tumor and precocious pseudopuberty. *J Pediatr* 1986; 108:945-947.

Majewski Syndrome 762

General: Autosomal recessive; normal chromosomes; perinatal mortality; has been suggested to be related to Mohr syndrome.

Ocular: Cataract; optic disk edema; optic atrophy; hypertelorism; absent lashes and brows; persistent pupillary membrane.

Clinical: Short rib polydactyly; cleft lip; cleft palate; narrow thorax; short tibia; hypoplastic epiglottis; lung and visceral abnormalities.

Cherstroy ED, et al. Difficulties in classification of the short rib polydactyly syndromes. *Eur J Pediatr* 1980; 133: 57-61.

Chess J, et al. Ocular pathology of the Majewski syndrome. *Br J Ophthalmol* 1982; 66:736-741.

Majewski F, et al. Polysyndaktylie, Verkuerzte Gliedmassen, und Genitalfehlbildungen. Z *Kinderheilkd* 1971; 111: 118-138.

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Silengo MC, et al. Orofacial-digital syndrome II. Transitional type between the Mohr and the Majewski syndromes: report of 2 new cases. *Clin Genet* 1987; 31:331-336.

Malaria 763

General: Caused by *Plasmodium*, which is transmitted by mosquito bite, blood transfusion, or contaminated needles and syringes.

Ocular: Proliferative retinitis; vascular embolism; keratitis; ocular herpes simplex; blepharitis; optic atrophy; papilledema; papillitis; optic neuritis; anisocoria; Argyll Robertson pupil; vitreal hemorrhages and opacity; cataract; myopia; strabismus; uveitis; scleral icterus; scotoma; lagophthalmos; ptosis; subconjunctival hemorrhages; paralysis of third, fourth, or sixth nerve; epibulbar hemorrhage involving the conjunctiva, episclera, tendinous insertion of the medial rectus.

Clinical: Fever; anemia; splenomegaly; death.

Bell RW. Ophthalmologic findings in malaria. *Ann Ophthalmol* 1975; 7: 1429.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hidayat AA, et al. The diagnostic histopathologic features of ocular malaria. *Ophthalmology* 1993; 100: 1183-1186.

Lewallen S, et al. Ocular fundus findings in Malawian children with cerebral malaria. *Ophthalmology* 1993; 100: 857-861.

Runyan TE, Ostberg RC. An unusual macular lesion associated with malaria. *Ann Ophthalmol* 1977; 9:1521-1525.

Malignant Hyperpyrexia Syndrome (Postcataract Hyperpyrexia Syndrome; Postinduction Hyperpyrexia Syndrome) 764

General: Etiology uncertain, but believed to be a secondary response to suxamethonium and halothane used with general anesthesia; high mortality rate of about 70%.

Ocular: Malignant hyperpyrexia following congenital cataract or strabismus surgery under general anesthesia.

Clinical: Rapid elevation of body temperature and vastly enhanced metabolic activity; hyperapnea; tachycardia.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Snow JC, Kerman TJ. Malignant hyperpyrexia. *EENT Dig* 1970; 49:427.

Petersdorf RG. Malignant hyperthermia. In: Isselbacher KJ, ed. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:2476.

Rosenberg H, Fletcher JE. An update on the malignant hyperthermia syndrome. *Ann Acad Med Singapore* 1994; 23[6 Suppl]:84-97.

Malignant Hyperthermia Syndrome 765

General: Pharmacogenetic disease with uninhibited flow of calcium ion into muscle substance; leads to combined metabolic and respiratory acidosis and liberation of heat; cellular death may result; autosomal dominant; more common in children and young adults; unusual disorder of skeletal and cardiac muscle triggered by anesthetic agents.

Ocular: Blepharoptosis; squint; pupils fixed and dilated.

Clinical: Hernias; kyphosis; clubfoot; excessive muscular bulk; muscle cramps; unstable blood pressure; rapid and deep respiration; mottled cyanosis; arrhythmias; muscle rigidity; oliguria; anuria; deep tendon reflexes absent.

Merei GJ, Bell RD. Preoperative management of the surgical patient with neurological disease. *Med Clin North Am* 1987; 71:511.

Waterman PM. Malignant hyperthermia syndrome. *Am J Ophthalmol* 1981; 92:461-465.

Waterman PM. Malignant hyperthermia: a case report. *Anesth Analg* 1980;59:220.

Mannosidosis 766

General: Rare; deficiency of alpha-mannosidase activity

Ocular: Lens opacities; corneal opacities; strabismus; late onset retinal dystrophy

Clinical: Coarse facial features; dysostosis multiplex; hearing defects; mental retardation; hepatosplenomegaly

Jolly RD, Shimada, Dalefield RR, et al. Mannosidosis: ocular lesions in the bovine model. *Curr Eye Res.* 1987; 9: 1073-1078.

Springer C, Gutschalk A, Meinck HM, et al. Late-onset retinal dystrophy in alpha mannosidosis. *Graefes Arch Clin Exp Ophthalmol.* 2005; 243: 1277-1279.

Marchesani Syndrome (Weill-Marchesani Syndrome; Inverted Marfan Syndrome; Brachymorphy with Spherophakia; Dystrophia Mesodermalis Congenita Hyperplastica) 767

General: Pattern of inheritance uncertain; manifest at age 9 months to 13 years.

Ocular: Lenticular myopia; secondary glaucoma (rare), caused by luxation of the lens; iridodonesis; ectopia lentis; spherophakia; optic atrophy; megalocornea; corneal opacity; acute pupillary block glaucoma ..

Clinical: Brachydactyly; reduced growth; athletic build with abundant subcutaneous tissue; short neck and large thorax; short and clumsy hands and feet; decreased joint flexibility; hearing defects; inheritable connective tissue disorder, usually inherited as an autosomal recessive.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Jensen AD, et al. Ocular complications in the Weill-Marchesani syndrome. *Am J Ophthalmol* 1974; 77:261.

Marchesani O. Brachydactylie und Angeborene Kugellinse als Systemerkrankung. *Klin Monatsbl Augenheilkd* 1939; 103:392.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Willi M, et al. Pupillary-block glaucoma in the Marchesani syndrome. *Arch Ophthalmol* 1973; 90:504.

Young ID, et al. Weill-Marchesani syndrome in mother and son. *Clin Genet* 1986; 30:475-480.

Marcus Gunn Syndrome (Jaw-Winking Syndrome; Congenital Trigeminooculomotor Synkinesis) 768

General: Familial occurrence rare, although dominant inheritance has been reported; symptoms caused by abnormal connections between external pterygoid muscle and levator palpebrae, with supranuclear or supranuclear-nuclear involvement (see Marin Amat Syndrome).

Ocular: Unilateral congenital ptosis in more than 90% of cases; 10% have spontaneous onset, usually in older persons; lid elevates rapidly when mouth is opened or mandible is moved to one or the other side; left eye seems to be more frequently affected than right eye; high incidences of strabismus (36%); amblyopia (34%); bilateral jaw-winking; decreased abduction.

Clinical: Stimulation of ipsilateral pterygoid with chewing, opening mouth, sucking, or contralateral jaw thrusts.

Doucet TW, Crawford JS. The quantification, natural course, and surgical results in 57 eyes with Marcus Gunn (jaw-winking) syndrome. *Am J Ophthalmol* 1981; 92:702-707.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gunn RM. Congenital ptosis with peculiar associated movements of the affected lid. *Trans Ophthalmol Soc U K* 1883; 3:283.

Pratt SG, Beyer CK, Johnson CC. The Marcus Gunn phenomenon. A review of 71 cases. *Ophthalmology* 1984; 91: 27-30.

Rowland LP. Marcus Gunn syndrome. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1995:533.

Schultz RO, Burian HM. Bilateral jaw winking. Reflex in association with multiple congenital anomalies. *Arch Ophthalmol* 1960; 64:946.

Marfan Syndrome (Dolichostenomelia; Arachnodactyly; Hyperchondroplasia; Dystrophia Mesodermalis Congenita) 769

General: Hypoplastic form of dystrophia mesodermalis congenita; autosomal dominant; affects both sexes; has been demonstrated that an abnormality of the gene coding for the connective tissue protein fibrillin is responsible for chronic Marfan syndrome.

Ocular: Exotropia; nystagmus; paralysis of accommodation; myopia (axial or lenticular); iridodonesis; miosis; persistent pupillary membrane; blue sclera; spherophakia; lens dislocation; cataract; megalocornea; retinal detachment (less frequently); pigmentary retinopathy; colobomata of macula, iris, optic nerve, and uveal tract (less frequently); keratoconus; central retinal artery occlusion; rhegmatogenous retinal detachment; syringoma.

Clinical: Arachnodactyly; skeletal anomalies; asymmetric thorax; dolichocephaly and high-arched palate; dissecting aneurysm; mitral valve prolapse; prominent ears; kyphoscoliosis; pectus excavatum; flat feet; hammer toes; pulmonary and kidney defects.

Butt Z, et al. Central retinal artery occlusion in a patient with Marfan's syndrome. *Acta Ophthalmol* 1992; 70: 281-284.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Cotlier E, Reinglass H. Marfan-like syndrome with lens involvement: hyaloideoretinal degeneration with anterior chamber angle, facial, dental, and skeletal anomalies. *Arch Ophthalmol* 1975; 93:93.

Dietz HC, et al. Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. *Nature* 1991; 352:337-339.

Marfan AB. Un Cas de Deformation Congenitale des Quatre Members Plus Prononcees aux Extremites Caracterisee par l'Allongement des Os avec un Certain Degre d'Amincissement. *Bull Mem Soc Med Hop Pans* 1896; 13:220.

Ramsey MS, et al. The Marfan syndrome: a histopathologic study of ocular findings. *Am J Ophthalmol* 1973; 76: 102.

Marin Amat Syndrome (Inverted Marcus Gunn Phenomenon) 770

General: Intrafacial connection between the orbicularis oculi and external pterygoid muscles; occurs primarily after peripheral facial palsy.

Ocular: When mouth is opened and/or mandible is moved to side opposite ptosis, closure of the eye occurs; increased tearing during mastication.

Clinical: Signs of old facial palsy usually recognizable.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Marin Amat. Contribucion del Estudio de la Curabilidad de las Paralisis Oculares de Origen Traumatico. *Arch Oftalmol Hispanoamo* 1918; 71.

Wartenberg R. Inverted Marcus Gunn phenomenon (so-called Marin Amat syndrome). *Arch Neural Psychiatr* 1948; 60:584.

Marinesco-Sjögren Syndrome (Congenital Spinocerebellar Ataxia- Congenital Cataract-Oligophrenia Syndrome) 771

General: Autosomal recessive trait; onset when child learns to walk; mitochondrial disease.

Ocular: Cataracts; aniridia; rotary and horizontal nystagmus; nystagmus; strabismus; optic atrophy.

Clinical: Cerebellar ataxia; oligophrenia; small stature; scoliosis; genu valgum; restricted extensibility of the knee; defects of fingers and toes; mental retardation; hair sparse; hypersalivation; sensorineural hearing loss.

Dotti MT, et al. Optic atrophy in Marinesco-Sjogren syndrome: an additional ocular feature. Report of three cases in two families. *Ophthalmic Paediatr Genet* 1993; 14:5-7.

Gillespie FDL. Aniridia, cerebellar ataxia, and oligophrenia in siblings. *Arch Ophthalmol* 1965; 73:338.

Lindal S, et al. Mitochondrial diseases and myopathies: a series of muscle biopsy specimens with ultrastructural changes in the mitochondria. *Ultrastruct Pathol* 1992; 16:263-275.

Marinesco G, et al. Nouvelle Maladie Familiale Caracterisee par une Cataracte Congenitale et un Arret du Developpement Somato-NeuroPsychique. *Encephale* 1931; 26:97.

Sjögren T. Hereditary congenital spinocerebellar ataxia combined with congenital cataracts and oligophrenia. *Confinia Neurol* 1950; 10[Suppl 46]:293.

Maroteaux-Lamy Syndrome (Systemic Mucopolysaccharidosis Type VI; MPS VI Syndrome; Mucopolysaccharidosis VI) 772

General: Onset in infancy; etiology unknown; autosomal recessive; excessive urinary excretion of chondroitin sulfate B; lysosomal storage disease; deficiency of the enzyme arylsulfatase B; multiple clinical phenotypes.

Ocular: Corneal haziness and opacities; pupillary membrane remnants.

Clinical: Skeleton deformities; restriction of articular movements; dyspnea; heart murmur; hearing impairment.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kenyon KR, et al. Ocular pathology of the Maroteaux-Lamy syndrome. *Am J Ophthalmol* 1972; 73:718.

Matalon R, et al. Deficiency of chondroitin sulfate N-acetylgalactosamine 4-sulfate sulfatase in Maroteaux-Lamy syndrome. *Biochem Biophys Res Commun* 1974; 61:1450-1457.

Quigley HA, Kenyon KR. Ultrastructural and histochemical studies of a newly recognized form of systemic mucopolysaccharidosis. *Am J Ophthalmol* 1974; 77:809.

Voskoboeva E, et al. Four novel mutant alleles of the arylsulfatase B gene in two patients with intermediate form of mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). *Hum Genet* 1994; 93:259-264.

Marquardt-Loriaux Syndrome (Wolfram Syndrome; Diabetes Insipidus-Diabetes Mellitus-Optic Atrophy-Deafness Syndrome; DIDMOAD Syndrome) 773

General: Autosomal recessive; present from childhood; age of onset varies.

Ocular: Optic nerve atrophy; color blindness; visual field defects; anisocoria; diabetic retinopathy; nystagmus; cataract; pigmentation of retina.

Clinical: Juvenile diabetes mellitus; diabetes insipidus; neurosensory hearing loss; hypertension; cerebellar dysfunction; vertigo; atony of urinary tract; anosmia; peripheral neuropathy; mitochondrial abnormalities; moderate hearing loss.

Bundey S, et al. Mitochondrial abnormalities in the DIDMOAD syndrome. *J Inherit Metab Dis* 1992; 15:315-319.

Higashi K. Otologic findings of DIDMOAD syndrome. *Am J Otol* 1991; 12:57-60.

Mtanda AT, et al. Optic atrophy in Wolfram syndrome. *Ophthalmol Paediatr Genet* 1986; 7:159-165.

Niemeyer G, Marquardt JL. Retinal function in an unique syndrome of optic atrophy, juvenile diabetes mellitus, diabetes insipidus, neurosensory hearing loss, autonomic dysfunction, and hyperalanineuria. *Invest Ophthalmol Vis Sci* 1972; 11:617.

Wolfram DJ. Diabetes mellitus and simple optic atrophy among siblings: report of four cases. *Mayo Clin Proc 1938;* 13:715.

Marshall (D) Syndrome (Atypical Ectodermal Dysplasia) 774

General: Autosomal dominant; variant of ectodermal dysplasia; onset at birth.

Ocular: Myopia; congenital cataract (spontaneous absorption not uncommon); degenerative fluid vitreous; luxation of lens; cataract; shallow orbits.

Clinical: Facial malformation; saddle nose; hypohidrosis; partial deafness; flat or retracted midface.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger. 1976.

Marshall D. Ectodermal dysplasia: report of kindred with ocular abnormalities and hearing defect. *Am J Ophthalmol* 1958; 45[Part 2]:143-156.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Stratton RF. et al. Marshall syndrome. *Am J Med Genet* 1991; 41:35-38.

Marshall (RE) Syndrome 775

General: Present from birth; etiology unknown; death usually from pneumonia before age 20 months.

Ocular: Exophthalmos; blue sclera; megalocornea; thick eyebrows.

Clinical: Underweight for length; long cranium; prominent forehead; hyperextension; small mandible; small upturned nose; broad middle and proximal phalanges; repeated respiratory infections; failure to thrive; mental retardation; accelerated skeletal growth.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Marshall RE, et al. Syndrome of accelerated skeletal maturation and relative failure to thrive: a newly recognized clinical growth disorder. *J Pediatr* 1971; 78:95-101.

Marshall-Smith Syndrome 776

General: Rare congenital condition with advanced bone age, facial anomalies, and relative failure to thrive.

Ocular: Hypertelorism, protuberant eyes with shallow orbits.

Clinical: Feeding and respiratory difficulties; developmental delay; advanced bone age; characteristic facies.

Summers DA, Cooper HA, Butler MG. Marshall-Smith syndrome: case report of a newborn male and review of the literature. *Clin Dysmorphol* 1999; 8:207-210.

Williams DK, et al. Marshall-Smith syndrome: the expanding phenotype. *J Med Genet* 1997; 34:842-845.

Yoder CC, et al. Marshall Smith syndrome: further delineation. *South Med J* 1988; 81:1297-1300.

Martsolf Syndrome 777

General: Autosomal recessive; rare; cardiac abnormalities.

Ocular: Cataracts.

Clinical: Mental retardation; short stature; hypogonadism.

Harbord MG, et al. Microcephaly, mental retardation, cataracts and hypogonadism in sibs: Martsolf's syndrome. *J Med Genet* 1989; 26:397-406.

Martsolf JT, et al. Severe mental retardation, cataracts, short stature, hypogonadism in two brothers. *Am J Med Genet* 1978; 1:291-299.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Sanchez JM, et al. Two brothers with Martsolf's syndrome. *J Med Genet* 1985; 22:308-310.

Masquerade Syndrome 778

General: Chronic blepharoconjunctivitis due to an underlying conjunctival carcinoma.

Ocular: Squamous cell and sebaceous carcinomas that mimic chalazion or other eyelid lesions; lacrimal secretory and excretory systems; lymph node involvement; hematogenous spread (rare).

Clinical: Orbital or regional involvement; intracranial extension and dural invasion.

Brownstein S. Masquerade syndrome. *Ophthalmology* 1980; 87:259-262.

Colak A, et al. Intracranial extension of meibomian gland carcinoma. *J Clin Neuro-Ophthalmol* 1991; 11:39-42.

Irvine AR. Diffuse epibulbar squamous cell epithelioma. *Am J Ophthalmol* 1967; 64:550-554.

Khan J, et al. Sebaceous and meibomian carcinomas of the eyelid. Recognition, diagnosis, and management. *Ophthal Plast Reconstr Surg* 1991; 7:61-66.

Theodore FH. Conjunctival carcinoma masquerading as chronic conjunctivitis. *Eye Ear Nose Throat Monthly* 1967; 46:1419-1420.

Mastocytosis (Urticaria; Mast Cell Leukemia) 779

General: Increased mast cells found in tissues and organs; range from cutaneous to systemic condition.

Ocular: Conjunctival pigmentation; keratitis; pingueculae.

Clinical: Urticarial wheals; mast cells infiltrate into liver, spleen, gastrointestinal system, and bones.

Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Duke-Elder S, MacFaul PA. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Graham JR. Pingueculae in systemic mastocytosis: a possible association. *Aust N Z J Med* 1992; 22:711.

Matsoukas Syndrome (Oculo-Cerebro-Articulo-Skeletal Syndrome) 780

General: Autosomal dominant; some of the features are found in Larsen syndrome, Schwartz syndrome, Hallermann-Streiff syndrome, Mietens syndrome, and Stickler syndrome; both sexes affected; onset at birth.

Ocular: Microphthalmia; myopia; increased pupillary distance; cataract; corneal sclerosis with vascular pericorneal net.

Clinical: Small stature; multiple joint dislocations; mental retardation; high palate; small mouth.

Matsoukas J, et al. A newly recognized dominantly inherited syndrome: short stature, ocular and articular anomalies, mental retardation. *Helv Paediatr Acta* 1973; 28:383.

Schwartz O, Jumpel RS. Congenital blepharophimosis associated with a unique generalized myopathy. *Arch Ophthalmol* 1962; 68:52.

McFarland Syndrome 781

General: Autosomal recessive; duplication of chromosome 16q22 has been proposed; prominent amniotic fluid leakage.

Ocular: Hypertelorism; dystopic canthi.

Clinical: Flat-appearing face; prominent forehead and frontal bossing; dislocation of joints (elbows, knees, hips most commonly); malformation of feet; short metacarpals; heart defects (usually ventricular septum); harelip; cleft palate; micrognathia.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Houlston RS, et al. Duplication of 16q22 qter confirmed by fluorescence in situ hybridization and molecular analysis. *J Med Genet* 1994; 31:884-887.

McFarland BL. Congenital dislocation of knee. *J Bone Joint Surg* 1929; 11:281.

Provenzano RW. Congenital dislocation of the knee: report of a case. *N Engl J Med* 1947; 236:360.

Vedantam R, Douglas DL. Congenital dislocation of the knee as a consequence of persistent amniotic fluid leakage. *Br J Clin Pract* 1994; 48:342-343.

McKusick-Weiblaecher Syndrome 782

General: Cataract; leg absence deformity; rare; two cases reported in Amish females whose parents share same ancestors; autosomal recessive.

Ocular: Congenital cataract; partial paralysis of oculomotor III nerve.

Clinical: Absence or deformity of leg; progressive scoliosis; partial duplication of foot; imperforate anus.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA, Weiblaecher RG. Recessive inheritance of congenital malformation syndrome. *JAMA* 1968; 204: 111-116.

Measles (Morbilli; Rubeola) 783

General: Acute, extremely communicable disease that affects young school-aged children; caused by paramyxovirus; multiple sclerosis.

Ocular: Hypopyon; uveitis; conjunctivitis; Koplik (Hirschberg) spots of conjunctiva; keratitis; corneal ulcer; cellulitis of lid; dacryocystitis; congenital cataract; optic atrophy; optic neuritis; strabismus; pigmentary retinopathy; iris prolapse; hemianopsia; secondary glaucoma; central retinal artery occlusion; orbital cellulitis; accommodative spasm; paralysis of sixth nerve; keratoconus.

Clinical: Maculopapular rash; fever.

Bloch-Michel E, et al. Measles virus antibody in aqueous humor of patients with uveitis associated with multiple sclerosis. *Lancet* 1992; 339:750-751.

Boniuk V. Rubeola (measles, morbilli). *Int Ophthalmol Clin* 1975; 15:243-255.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Frederique G. Corneal ulcers in rubeola. *Am J Ophthalmol* 1969; 68:996-1003.

Peduzzi M, et al. Bilateral pigmented retinopathy following measles: long-term follow-up and possible association with keratoconus. *Eur J Ophthalmol* 1991; 1:148-150.

Tierney LM. Measles. In: Tierney LM, ed. *Current medical diagnosis and treatment*, 33rd annual revision. Stamford: Appleton & Lange, 1994:1107.

MEB Disease (Muscle-Eye-Brain Disease) 784

General: Autosomal recessive; possibly the same as Walker-Warburg syndrome. **Ocular:** Severe congenital myopia; congenital glaucoma; pallor of optic disk; retinal hypoplasia.

Clinical: Congenital muscular dystrophy; mental retardation; hydrocephalus; myoclonic jerks; high serum creatine phosphokinase.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Santauvori P, Leisti J. Muscle, eye and brain disease. In: Eriksson AW, et al., eds. *Population structure and genetic disorders.* Orlando, FL: Academic Press, 1981:647-651.

Santavuori P, et al. Muscle-eye brain disease and Wa1ker-Warburg syndrome [Letter]. *Am J Med Genet* 1990; 36: 371-372.

Meckel Syndrome (Dysencephalia Splanchnocystic Syndrome; Gruber Syndrome) 785

General: Autosomal recessive; ocular manifestations are similar to those of trisomy 13-15 syndrome.

Ocular: Cryptophthalmos; clinical anophthalmos; microphthalmos; mongoloid slant of lid fissures; sclerocornea; microcornea; partial aniridia; cataract; retinal dysplasia; posterior staphyloma; optic nerve hypoplasia.

Clinical: Sloping forehead; posterior encephalocele; short neck; polydactyly and syndactyly (hands and feet); polycystic kidneys; cryptorchidism; cleft lip and palate; central nervous system abnormalities, including the Dandy-Walker malformation.

Herriot R, et al. Dandy-Walker malformation in the Meckel syndrome. *Am J Med Genet* 1991; 39:207-210.

MacRae DW, et al. Ocular manifestations of the Meckel syndrome. *Arch Ophthalmol* 1972; 88:106.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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MELAS Syndrome 786

General: Changing of threonine at amino acid 109 to an alanine; A3245G mitochondrial DNA point mutation Cataracts; RPE abnormalities with age-related maculopathy

Clinical: Migraines; sensorineural hearing loss; grand mal seizures; stroke-like episodes; lactic acidosis; ragged-red muscle fibers

Jones, M, Mitchell P, Wang JJ, et al. MELAS A3245G mitochondrial DNA mutation and age related maculopathy. *Am J Ophthalmol* 2004; 138: 1051-1053.

Latkany P, Ciulla TA, Cacchillo PF, et al. Mitochondrial maculopathy: geographic atrophy of the macula in the MELAS associated A to G 3243 mitochondrial DNA point mutation. *Am J Ophthalmol* 1999; 128: 112-114.

Latvala T, Mustonen E, Uusitalo R, et al. Pigmentary retinopathy in patients with the MELAS mutation 3243A-G in mitochondrial DNA. *Graefes Arch Clin Exp Ophthalmol.* 2002; 240: 795-801.

Melkersson-Rosenthal Syndrome (Melkersson Idiopathic Fibroedema; Miescher Cheilitis Granulomatosis) 787

General: Occurrence in childhood or youth; possible etiologies include viral infection, tuberculosis, sarcoidosis, and allergic reactions (all affecting parasympathetic cells in geniculate ganglia); facial palsy resembles Bell palsy; possible localization of this disorder to the gene at 9p11 has been reported.

Ocular: Lagophthalmos; lid edema; lacrimation secondary to the "crocodile tear" phenomenon from aberrant seventh nerve regeneration; exposure keratitis and corneal ulcers; corneal opacities.

Clinical: Chronic edema of face and lips; peripheral facial palsy (may be bilateral), which may precede edema by weeks to years; furrowed tongue; granulomatous cheilitis and glossitis; lingua plicata.

Cohen HA, et al. Melkersson-Rosenthal syndrome. *Cutis* 1994; 54:327-328.

Hallet JW, Mitchell B. Melkersson-Rosenthal Syndrome. *Am J Ophthalmol* 1968; 65:542.

Paton D. The Melkersson-Rosenthal Syndrome. *Am J Ophthalmol* 1965; 59:705.

Rosenthal C. Klinischer bio1ogischer Beitrag zur Konstitutionspathologie: Gemeinsames Aufreten von (rezidivierender familiarer) Facialislahmung, Angioneurotischem Gesichtsoderm und Lingua Plicata in Arthritismus-Familien. Z *Neurol Psychiatr* 1931; 475:500.

Smeets E, et al. Melkersson-Rosenthal syndrome and de novo autosomal t(9;21)(P 11;p 11) translocation. *Clin Genet* 1994; 45:323-324.

Melnick-Needles Syndrome (Osteodysplasty) 788

General: Bone dysplasia; fewer than 30 cases reported; familial congenital autosomal trait; affect both sexes; onset at birth.

Ocular: Exophthalmos; hypertelorism; bilateral sclerocornea; mild cornea plana; strabismus.

Clinical: Misaligned teeth; micrognathia; multiple symmetric bone deformities; large ears; broad nose; frontal bossing; rosy cheeks; short stature; recurrent respiratory and ear infections; pneumosinus dilatans; abnormalities of the distal phalanges.

Melnick JL, Needles CF. An undiagnosed bone dysplasia: a 2-family study of 4 generations and 3 generations. *Am J Roentgenol* 1966; 97:39-48.

Memis A, et al. Case report 717. Osteodysplasty (Melnick-Needles syndrome). *Skel Radial* 1992; 21:132-134.

Perry LD. et al. Melnick-Needles syndrome. *J Pediatr Ophthalmol Strabismus* 1978; 15:226-230.

Stretch JR, Poole MD. Pneumosinus dilatans as the etiology of progressive bilateral blindness. *Br J Plast Surg 1992;* 45:469-473.

Ménière Syndrome 789

General: Etiology unknown; more common in males between ages 40 and 60 years.

Ocular: Nystagmus (rapid component toward the normal side), mainly during attacks; diplopia possible during and after attacks.

Clinical: Paroxysmal attacks of vertigo; tinnitus; gradually progressing deafness, although not prerequisite for diagnosis; during attacks, pallor, nausea, vomiting, and fainting; allergy; giant cell arteritis; facial paralysis.

Black FO, Kitch R. A review of vestibular test results in Meniere's disease. *Otolaryngol Clin North Am* 1980; 13: 631-642.

Derebery MJ, Valenzuela S. Meniere's syndrome and allergy. *Otolaryngol Clin North Am* 1992; 25:213-224.

McKennan KX, et al. Meniere's syndrome: an atypical presentation of giant cell arteritis (temporal arteritis). *Laryngoscoper* 1993; 103:1103-1107.

Meniere P. *Memoire sur des Lesions de l'Oreille Interne*. Paris: Gaz. Med, 1861.

Wilmot TJ. The effect of beta histine hydrochloride in Meniere's disease. *Acta Otolaryngol Suppl* 1972; 305:18.

Meningioma 790

General: Benign, slow-growing tumors that arise from the arachnoid matter, the middle layer of meninges that lies inside the dura mater, and outside the pia mater; more common in females; peak incidence in the seventh decade of life.

Ocular: Exposure keratopathy; paralysis of extraocular muscles; proptosis; optic nerve atrophy; papilledema; choroidal folds; hyperopia; visual field defect; afferent pupil defect; optociliary shunt veins.

Clinical: Headache; intracranial pressure; vomiting.

Anderson D, Khalil M. Meningioma and the ophthalmologist. A review of 80 cases. *Ophthalmology* 1981; 88: 1004-1009.

Finn JE, Mount LA. Meningiomas of the tuberculum sellae and planum sphenoidale: a review of 83 cases. *Arch Ophthalmol* 1974; 92:23-27.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Miller NR, Solomon S. Retinochoroidal (optociliary) shunt veins, blindness and optic atrophy: a non-specific sign of chronic optic nerve compression. *Aust N* Z *J Ophthalmol* 1991; 19:105-109.

Meningococcemia *(Neisseria Meningitides;* Meningitis) 791

General: Systemic bacterial infection caused by *Neisseria meningitides;* can be present chronically in patients with immune deficiencies including deficient complement levels.

Ocular: Photophobia; conjunctivitis; chemosis; keratitis; uveitis; panophthalmitis; retinal endophlebitis; macular edema; papillitis; optic neuritis; paresis of sixth or seventh nerve; nystagmus; miosis; hippus; cortical blindness; papilledema (rare); conjunctival petechiae; strabismus.

Clinical: Meningitis; fever; malaise; joint pain; splenic enlargement.

Bannatyne RM. Meningococcal ophthalmia. *Can Med Assoc J* 1978; 118:769.

Brinser JH, Hess JB. Meningococcal endophthalmitis without meningitis. *Can J Ophthalmol* 1981; 16:100-101.

Hanna LS, et al. Ocular complications in meningitis "fifteen year study." *Metab Pediatr Syst Ophthalmol* 1988; 11: 160-162.

Kilpi T, et al. Length of prediagnostic history related to the course and sequelae of childhood bacterial meningitis. *Pediatr Infect Dis J* 1993; 12:184-188.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part 1,* 4th ed. Baltimore: Williams & Wilkins, 1995:2983.

Nussbaum E, et al. Primary meningococcal conjunctivitis followed by meningitis. *J Pediatr* 1978; 92:784-785.

Menkes (2) Syndrome (Kinky Hair Syndrome) 792

General: Etiology unknown; sex-linked recessive neurodegenerative disorder; focal cerebral and cerebellar degenerative changes involving the white and gray matter; affects only males; onset in early infancy.

Ocular: Decreasing visual function with progression of the disease.

Clinical: Spasticity; refractory motor seizures; retarded growth; dementia; abnormal pigmentation of hair with kinky, wiry texture; lack of facial expression; thick and dry skin; transient jaundice.

Levy NS, et al. Ocular abnormalities in Menkes' kinky hair syndrome. *Am J Ophthalmol* 1974; 77:319.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Menkes JH, et al. A sex linked recessive disorder with retardation of growth, peculiar hair, and focal cerebral and cerebellar degeneration. *Pediatrics* 1962; 29:764.

Seelenfreund MH. The ocular pathology of Menkes' disease. *Arch Ophthalmol* 1968; 80:718.

Tuppurainen K, et al. Ophthalmological findings in a girl with Menkes-like disease. *Graefes Arch Clin Exp Ophthalmol* 1994; 232:445-447.

Welby J, et al. Isolation of a candidate gene for Menkes' disease that encodes a potential heavy metal binding protein. *Nat Genet* 1993; 3:14-19.

Mercury Poisoning (Minamata Syndrome) 793

General: Both sexes affected; onset several weeks or months after ingestion of fish from contaminated water or animals fed with contaminated grain; symptoms may be mild to severe.

Ocular: Constriction of visual fields; blindness.

Clinical: Paresthesia of mouth, tongue, and extremity; hearing loss; asthenia; fatigue; inability to concentrate; dysarthria; tremors; persistent vegetative state; peripheral neuropathy; cerebella ataxia; gait disturbance; sensory impairment; anosmia; loss of taste; bladder disturbance; mental deterioration.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Takeuchi T, et al. Neuropathology of childhood cases of methylmercury poisoning (Minamata disease) with prolonged symptoms, with particular reference to the decortication syndrome. *Neurotoxicology* 1979; 1:1-20.

Meretoja Syndrome (Finnish Type, FAP IV) 794

General: Lattice corneal dystrophy type II with familial amyloid polyneuropathy type IV; also called primary hereditary systemic amyloidosis.

Ocular: Lattice corneal dystrophy; cranial nerve palsies.

Clinical: Multiple neurologic symptoms such as severe itching, various nerve palsies, and diminished vibratory sensation; patients are said to develop a so-called bloodhound-like appearance due to skin and facial nerve degeneration.

Asaoka T, et al. Lattice corneal dystrophy type II with familial amyloid polyneuropathy type IV. *Jpn J Ophthalmol* 1993; 37:426-431.

Purcell JJ, et al. Lattice corneal dystrophy associated with familial systemic amyloidosis (Meretoja's syndrome). *Ophthalmology* 1983; 90:1512-1517.

Rintala AE, et al. Primary hereditary systemic amyloidosis (Meretoja's syndrome): clinical features and treatment by plastic surgery. *Scand J Plast Reconstr Surg Hand Surg* 1988; 22:141-145.

MERRF Syndrome 795

General: Associated with mitochondrial tRNA [Leu(UUR)] A3243G mutation.

Ocular: Optic neuropathy; pigmentary retinopathy, ophthalmoparesis, and ptosis.

Clinical: Mitochondrial encephalomyopathy; lactic acidosis; strokelike episodes.

Hwang JM, Park HW, Kim SJ. Optic neuropathy associated with mitochondrial tRNA[Leu(UUR)] A3243G mutation. *Ophthalmic Genet* 1997; 18:101-105.

Meshers Macroreticular Dystrophy of Retinal Pigment Epithelium (Butterfly-Shaped Dystrophy of Retinal Pigment Epithelium) 796

General: Autosomal recessive; autosomal dominant inheritance has been reported.

Ocular: Butterfly-shaped dystrophies of retinal pigment epithelium; macular degeneration associated with fundus flavimaculatus; drusen of Bruch membrane; choroidal folds; bull's-eye degeneration of macula; detachment of pigment epithelium.

Clinical: None.

Bastiaensen LA, et al. Patterned anomalies of the retinal pigment epithelium: dystrophy or syndrome? *Doc Ophthalmol* 1983; 5: 17-29.

Girard P, et al. Macroreticular and butterfly shaped dystrophies of the retinal pigment epithelium. *J Fr Ophthalmol* 1980; 3:101-108.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Nichols BE. Butterfly-shaped pigment dystrophy of the fovea caused by a point mutation in codon 167 of the RDS gene. *Nat Genet* 1993; 3:202-207.

Mesodermal Dysgenesis (Anterior Chamber Dysgenesis; Dysembryogenesis; Anterior Segment Ocular Dysgenesis Syndrome) 797

General: Mesodermal abnormalities, including oculocutaneous albinism; autosomal dominant.

Ocular: Capsular cataracts; external ophthalmoplegia; anterior chamber cleavage syndrome; atrophy of iris; ectropion; flat cornea; coloboma of iris and choroid; posterior embryotoxon; Axenfeld anomaly; Rieger anomaly; Peters anomaly; keratoconus; microphthalmos.

Clinical: None.

Ferrell RE, et al. Anterior segment mesenchymal dysgenesis; probable linkage to the MNS blood group on chromosome 4. *Am J Hum Genet* 1982; 24:245-249.

Lubin JR. Oculocutaneous albinism associated with corneal mesodermal dysgenesis. *Am J Ophthalmol* 1981; 91:347-350.

Lubin JR. Oculocutaneous albinism and corneal mesodermal dysgenesis. *Am J Ophthalmol* 1981; 92:587.

Ghose S, et al. Microphthalmos and anterior segment dysgenesis in a family. *Ophthalmic Paediatr Genet* 1991; 12:177-182.

Metaphyseal Chondrodysplasia with Retinitis Pigmentosa 798

General: Autosomal recessive.

Ocular: Retinitis pigmentosa.

Clinical: Defective cartilage and growth of long bones, particularly the metacarpals and phalanges.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Phillips CI, et al. Retinitis pigmentosa, metaphyseal chondrodysplasia and brachydactyly: an affected brother and sister. *J Med Genet* 1981; 18:46-49.

Metastatic Bacterial Endophthalmitis 799

General: Causative agent usually of low pathogenicity (e.g., *Staphylococcus albus, Staphylococcus epidermidis);* occasionally organisms of greater pathogenicity (e.g., *Pseudomonas aeruginosa, Diplococcus pneumoniae);* bilateral 45%; organisms originate in body or are introduced by drug addicts using nonsterile needles.

Ocular: Conjunctival hemorrhages; conjunctivitis; Roth spots; retinal arterial occlusion; uveitis; hypopyon; chorioretinitis; endophthalmitis; retinal hemorrhages.

Clinical: Manifestations are nonspecific.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Pflugfelder SC, Flynn HW Jr. Infectious endophthalmitis. Infect Dis Clin North Am 1992; 6:859-873.

Rogell G. Infectious and inflammatory diseases. In: Duane TD, ed. Clinical *Ophthalmology*, vol. V. Philadelphia: JB Lippincott, 1982: 10-11.

Metastatic Fungal Endophthalmitis 800

General: Usually occurs in immunosuppressed or immunocompromised patients; usually asymmetrical; *Candida albicans* frequent etiologic agent.

Ocular: Anterior uveitis; vitreitis; focal retinitis; Roth spots; chorioretinitis; Fusarium solani also has been isolated from immunocompromised patients with endogenous endophthalmitis.

Clinical: May be evidence of other monocular foci of metastatic fungal disease.

Cohen M, Montgomerie JZ. Hematogenous endophthalmitis due to Candida tropicalis: report of two cases and review. *Clin Infect Dis* 1993; 17:270-272.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Louie T, et al. Endogenous endophthalmitis due to Fusarium: case report and review. *Clin Infect Dis* 1994; 18:585-588.

Methemoglobinemia 801

General: Deficiency of enzyme; inherited or acquired, with acquired most common; caused by contact with drugs and chemicals; disorder disappears when offending chemical is eliminated.

Ocular: Pigmentation of conjunctiva and retina.

Clinical: Cyanosis; mental retardation; central nervous system involvement.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*, 11th ed. New York: McGraw-Hill, 1987.

Duke-Elder S, MacFaul PA. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Gibson QH. Methemoglobinemia-long ago and far away. *Am J Hematol* 1993; 42:3-6.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Meyer-Schwickerath-Weyers Syndrome (Microphthalmos Syndrome; Oculodentodigital Dysplasia) 802

General: Etiology unknown; two types recognized: (I) dysplasia oculodentodigitalis and (II) dyscraniopygophalangie; type I is characterized by microphthalmia with possible iris pathology and glaucoma, oligodontia and brown pigmentation of teeth, camptodactyly, and possible absence of middle phalanx of second to fifth toes; type II consists of severe microphthalmos to anophthalmos, polydactyly, and developmental anomalies of nose and oral cavity; both sexes affected; present from birth; abnormal cerebral white matter.

Ocular: Microphthalmos; hypotrichosis; glaucoma; iris anomalies (eccentric pupil; changes in normal iris texture; remnants of pupillary membrane along iris margins); microcornea; hypertelorism; myopia; hyperopia; keratoconus.

Clinical: Thin, small nose with anteverted nostrils and hypoplastic alae; syndactyly; camptodactyly (fourth and fifth fingers); anomalies of middle phalanx of fifth finger and toe; hypoplastic teeth; wide mandible; alveolar ridge; sparse hair growth; visceral malformations.

Geeraets WK. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gutmann DH, et al. Oculodentodigital dysplasia syndrome associated with abnormal cerebral white matter. *Am J Med Genet* 1991; 41:18-20.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Meyer-Schwickerath, et al. Mikrophthalmus-syndrome. *Klin Monatsbl Augenheilkd* 1957; 131:18.

Weyers H. Zur Dyscephalie mit Cataracta Congenita und Hypotrichose. *Eur J Pediatr* 1954; 74:468.

Micro Syndrome 803

General: Autosomal recessive microcephaly and microcornea; Muslim Pakistani inheritance; present at birth; consanguinity; autosomal recessive.

Ocular: Microcornea; congenital cataract; retinal dystrophy; optic nerve atrophy; ptosis; microphakia; microphthalmos; nuclear cataract; atonic pupils.

Clinical: Severe mental retardation; hypothalamic hypogenitalism; hypoplasia of the corpus callosum; short stature; cortical visual impairment; microcephaly; developmental delay.

Ainsworth JR, et al. Micro syndrome in Muslim Pakistan children. *Ophthalmology* 2001; 108:491-497.

Megarbane A, et al. Microcephaly, microphthalmia, congenital cataract, optic atrophy, short stature, hypotonia. Severe psychomotor retardation, and cerebral malformations: a second family with micro syndrome or a new syndrome? *J Med Genet* 1999; 36:637-640.

Rodriguez C, Rufo M, Gomez de Terreros I. A second family with micro syndrome. *Clin Dysmorphol 1999; 8:* 241-245.

Warburg M, et al. Autosomal recessive microcephaly, microcornea, congenital cataract, mental retardation, optic atrophy, and hypogenitalism: micro syndrome. *Am J Dis Child* 1993; 147:1309-1312.

Microcephaly with Chorioretinopathy 804

General: Autosomal dominant; congenital infection; exposure to irradiation, chemical agents, mother's infection, or injury.

Ocular: Chorioretinopathy usually inactive.

Clinical: Microcephaly; slow growth of brain; mild mental retardation.

Fanconi R, et al. Chorio-retinal dysplasia, microcephaly and mental retardation: an autosomal dominant syndrome. *Clin Genet* 1981; 20:347-351.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Sadler LS, Robinson LK. Chorioretinal dysplasia-microcephaly-mental retardation syndrome: report of an American family. *Am J Med Genet* 1993; 47:65-68.

Microcephaly, Microphthalmia, Cataracts, and Joint Contractures 805

General: Autosomal dominant; ocular features like Hagberg-Santavuori syndrome.

Ocular: Microphthalmia; cataracts; hypopigmented retinal degeneration.

Clinical: Microcephaly; shortening or wasting of muscle fibers, causing excess scar tissue over joints.

Bateman JB, et al. Ocular features of Hagberg-Santavuori syndrome. *Am J Ophthalmol* 1986; 102:262-271.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Microphthalmia and Mental Deficiency 806

General: Autosomal recessive.

Ocular: Microphthalmia; corneal opacities.

Clinical: Severe mental retardation; spastic cerebral palsy; glycinuria; abnormally small head.

Balci L. Corneal opacity, microphthahnos, mental retardation; microcephaly, and generalized muscular spasticity associated with hyperglycinemia. *Clin Genet* 1974; 5:36-39.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Microphthalmos, Myopia, and Corectopia 807

General: Autosomal dominant; characterized by microphthalmos, myopia, and corectopia.

Ocular: Microphthalmos; myopia; corectopia; glaucoma; cataract; hypoplastic macula; spherophakia; microphakia.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Scheie HG. *Text of Ophthalmology*, 10th ed. Philadelphia: WB Saunders, 1986.

Microphthalmos, Pigmentary Retinopathy, Glaucoma 808

General: Autosomal dominant; three disorders combined.

Ocular: Microphthalmos; pigmentary retinopathy; glaucoma.

Clinical: None.

Hermann P. Syndrome: Microphthalmic-retinite pigmentaire-glaucoma. *Arch Ophthalmol* 1958; 18:17-24.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Micropsia Syndrome (Lilliputian Syndrome) 809

General: Psychosensory illusion produced by various mental derangements such as acute infections, alcoholism, toxic delirium, dementia, or trauma.

Ocular: Illusions, with misjudging of distance, position, and size of known objects (regarded as a psychovisual phenomenon).

Clinical: Fixed hallucinations or dreams are expressions of illusions and are misinterpreted by the patient.

Bender MB, Savitzky N. Micropsia and teleopsia limited to the temporal fields of vision. *Arch Ophthalmol* 1943; 29:904.

Savitzky N, Tarachow S. Lilliputian hallucinations during convalescence from scarlet fever in a child. *J Nerv Ment Dis* 1941; 93:310.

Microspherophakia with Hernia 810

General: Autosomal dominant.

Ocular: Microspherophakia; glaucoma.

Clinical: Inguinal hernia.

Johnson VP, et al. Dominant microspherophakia. *Arch Ophthalmol* 1971; 85:534-537.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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MIDAS Syndrome (Microphthalmia, Dermal Aplasia and Sclerocornea) 811

General: X-linked phenotype; male-lethal trait

Ocular: Bilateral microphthalmia; sclerocornea; blepharophimosis

Clinical: dermal aplasia; microcephaly; cardiomyopathy; ventricular fibrillation; congenital heart defect

Cope CJ, Zaidman GW, Beck AD, et al. Phenotypic variation in ophthalmic manifestations of MIDAS syndrome (microphthalmia, dermal aplasia and sclerocornea) *Arch Ophthal*. 2004; 122:1070-1074

Happle R, Daniels O, Koopman RJ,: MIDAS syndrome (microphthalmia, dermal aplasia and sclerocornea): an x-linked phenotype distinct from Goltz syndrome. *Am J Med Genet*. 1993; 47:710-713.

Mietens Syndrome (Mietens-Weber Syndrome) 812

General: Etiology unknown; unclassifiable familial condition.

Ocular: Bilateral corneal opacities; horizontal and rotational nystagmus; strabismus; bushy eyebrows; ptosis.

Clinical: Growth failure; flexion contracture of the elbows; dislocation of the head of the radii; mental retardation; small pointed nose with a depressed root; low hairline; external ear defects; digital defects; hypertrichosis.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981:548.

Mietens C, Weber H. A syndrome characterized by corneal opacity, nystagmus, flexion contracture of the elbows, growth failure and mental retardation. *J Pediatr* 1966; 69:624.

Waring GO, Rodrigues MM. Ultrastructural and successful keratoplasty of sclerocornea in Mietens' syndrome. *Am J Ophthalmol* 1980; 90:469-475.

Migraine (Vascular Headache) 813

General: Recurrent attacks of pain in the head; usually unilateral; often familial.

Ocular: Abnormal visual sensations; scotoma generally restricted to one half of the visual field; complete blindness; unilateral transient visual loss; photopsia; branch retinal artery occlusions; anisocoria.

Clinical: Nausea; vomiting; anorexia; sensory, motor, and mood disturbances; fluid imbalance; headache.

Drummond PD. Cervical sympathetic deficit in unilateral migraine headache. *Headache* 1991; 31:669-672.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Glenn AM, et al. Complicated migraine resulting in blindness due to bilateral retinal infarction. *Br J Ophthalmol* 1992; 76:189-190.

Hykin PG, et al. Bilateral cilioretinal artery occlusion in classic migraine. *Postgrad Med J* 1991; 67:282-284.

Tomsak RL. Ophthalmologic aspects of headache. *Med Clin North Am* 1991; 75:693-706.

Saper JR. Migraine. II. Treatment. *JAMA* 1978; 239:2480-2484.

Smith JL. Migraine. *Neuro-Ophthalmology update*. New York: Masson, 1982.

Mikulicz-Radecki Syndrome (Mikulicz Syndrome; Dacryosialoadenopathy; Mikulicz-Sjögren Syndrome) 814

General: Not an individual disease but a manifestation of tuberculosis, syphilis, leukemia, lymphosarcoma, sarcoidosis, Hodgkin disease, mumps, Waldenström macroglobulinemia, or lymphoma; exhibits a chronic course with frequent recurrences; milder form of Sjögren syndrome (see Schaumann Syndrome).

Ocular: Bilateral painless enlargement of lacrimal glands with bulging of upper lid; decreased or absent lacrimation; conjunctivitis; uveitis; optic atrophy; optic neuritis; phlyctenules; keratoconjunctivitis; dacryoadenitis; retinal candlewax spots; periphlebitis.

Clinical: Symmetrical, perhaps marked, enlargement of salivary glands; dryness of mouth and pharynx; hoarseness; neurologic complications.

Meyer D, et al. Differential diagnosis in Mikulicz's syndrome, Mikulicz's disease, and similar disease entities. *Am J Ophthalmol* 1971; 71:516-524.

von Mikulicz-Radecki J. Ueber eine Eigenartige Symmetrische Erkrankung der Thranen und Mundspeicheldrusen. *Beitr Chirurg Festschr Gewid T Billroth Stuttgart* 1892; 610-630.

Millard-Gubler Syndrome (Abducens-Facial Hemiplegia Alternans) 815

General: Vascular, infectious, or tumorous lesion at the base of the pons affecting the nuclei of the sixth and seventh nerves and fibers of the pyramidal tract; demyelinating disease.

Ocular: Diplopia; esotropia; paralysis external rectus muscle (often bilateral); in unilateral cases, there is deviation of eyes to side opposite lesion and inability to move them toward side of lesion; abduction of eye prevented by destruction of sixth nerve nucleus; opposite eye cannot be voluntarily adducted but can converge and move in this position by rotatory and caloric stimulation.

Clinical: Ipsilateral facial paralysis; contralateral hemiplegia of arm and leg from involvement of pyramidal tract.

Bronstein AM, et al. Abnormalities of horizontal gaze: *Clinical*, oculographic and agenetic resonance findings I. Abducens palsy. *J Neurol Neurosurg Psychiatry* 1990; 53:194.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gubler A. De l'Hemiplegie Alterne Envisagee Comme Signe de Lesion de la Protuberance Annulaire et Comme Preuve de la Decussation des Nerfs Faciaux. *Gaz Hebd Med Chir* 1856; 3:749,789,811.

Minderhoud JM. Diagnostic significance of symptomatology in brain stem ischaemic infarction. *Eur Neurol* 1971; 5:343.

Newman NJ. Third, fourth and sixth nerve lesions and the cavernous sinus. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994:2458.

Miller Syndrome (Postaxial Acrofacial Dysostosis; Genee-Wiedemann Syndrome) 816

General: Cause unknown; sporadic and familial cases known as Genee-Wiedemann Syndrome.

Ocular: Ectropion.

Clinical: Malar hypoplasia; cleft palate and lip; postaxial limb deficiency; cup-shaped ears.

Chrzanowska KH, et al. Phenotype variability in the Miller acrofacial dysostosis syndrome: report of two further patients. *Clin Genet* 1989; 35:157-160.

Genee E. Une forme extensive de dysostose mandibulo-faciale. *J Genet Hum* 1969; 17:45-52.

Miller M, Fineman R, Smith DW. Postaxial acrofacial dysostosis syndrome. *J Pediatr* 1979; 95:970-975.

Wiedemann HR. Missbildungs-Retardierungs-Syndrom mit Fehlen des 5 Strahls an Haenden and Fuessen, Gaumenspalte, dysplastischen Ohren und Augenlidem und radioulnarer Synostose. *Klin Paediatr* 1973; 185: 181-186.

Miller Syndrome (Wilms Aniridia Syndrome; WAGR Syndrome; Wilms Tumor-Aniridia-Genitourinary Abnormalities-Mental Retardation Syndrome) 817

General: Etiology unknown; manifests an association of aniridia, which is inherited as a dominant autosomal trait, and Wilms tumor; this is one of the best studied continuous gene syndromes as defined by Schmickel.

Ocular: Glaucoma; bilateral aniridia (aniridia often not complete, with remnants of iris root present as rudimentary forms); cataract.

Clinical: Wilms tumor; mental retardation with microcephaly; genital malformations with cryptorchidism and hypospadias; hemihypertrophy; kidney anomalies (horseshoe kidney).

Fraumeni JF, Glass AG. Wilms' tumor and congenital aniridia. *JAMA* 1968; 206:825.

Mackintosh TF, et al. Aniridia and Wilms' tumor (nephroblastoma). *Br J Ophthalmol* 1968; 52:846.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Miller RW, et al. Association of Wilms' tumor with aniridia, hemihypertrophy and other congenital malformations. *N Engl J Med* 1964; 270:922.

Schmickel RD. Chromosomal deletions and enzyme deficiencies. *J Pediatr* 1986; 108:244-246.

Mirror Image Syndrome (Autoscopic Syndrome; Lukianowicz Phenomenon) 818

General: Patient's delusion that he or she is seeing a double of himself or herself; seen in patients with schizophrenia, epilepsy, migraine, and even depression; the "double" usually appears suddenly and is of white or gray hue (see Capgras Syndrome, of which this syndrome is a variant).

Ocular: Hallucination in the form of seeing a double of self.

Clinical: Migraine; schizophrenia; epilepsy; depression.

Lipton CW. Hallucinations of physical duality in migraine. *J Nerv Ment* Dis 1953; 117:345.

Lukianowicz N. Autoscopic phenomena. *Arch Neurol Psychiatr* 1958; 80:199.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed Philadelphia: JB Lippincott, 1981.

Misdirected Third Nerve Syndrome 819

General: May occur with a variety of inflammatory infections and parainfections, vascular lesions, tumors, and degenerative and demyelinating diseases that may involve the nerve anywhere; may occur as primary aberrant regeneration without prior history of acute oculomotor nerve palsy.

Ocular: Bizarre eyelid movements that may accompany various eye movements; lid may rise as the medial rectus, the inferior rectus, or the superior rectus muscle contracts; iridoplegia; ptosis.

Clinical: None.

Beard C. Misdirected third nerve syndrome. In: Mosby CV, ed. *Ptosis,* 3rd ed. St. Louis: CV Mosby, 1981:115.

Harley RD, ed. *Pediatric Ophthalmology,* 4th ed. Philadelphia: WB Saunders, 1998.

Lepore FE, Glaser JS. Misdirection revisited: a critical appraisal of acquired oculomotor nerve synkinesis. *Arch Ophthalmol* 1980; 98:2206-2209.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol. III.* 4th ed. Baltimore: Williams & Wilkins, 1988.

Roy FH. *Ocular Differential Diagnosis,* 7th ed. Philadelphia: Lippincott Williams & Wilkins, 2002.

Schatz NJ, et al. Primary aberrant oculomotor regeneration: a Sign of intracavernous meningioma. *Arch Neurol* 1977; 34:29-32.

ML I (Mucolipidosis I; Lipomucopolysaccharidosis; Dysmorphic Sialidosis; Spranger Syndrome) 820

General: Rare storage disease; autosomal recessive; increased sialic acid and deficiency of the enzyme alpha-N-acetylneuraminidase in cultured mucolipidosis I fibroblasts.

Ocular: Variable corneal clouding; macular cherry-red spot; optic atrophy; lens opacity; pupillary reflexes anomaly; grayish area around cherry-red spot.

Clinical: Moderate progressive mental retardation; skeletal changes of dysostosis multiplex; peripheral neuropathy; myoclonic jerks; tremor; cerebellar signs; gait abnormalities.

Carta F, et al. Mucolipidosis 1: ocular signs in three sisters. *Metab Ophthalmol* 1985; 8:21-27.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Shinkai H, et al. Connective tissue naevus with pseudo-Hurler polydystrophy. *Br J Dermatol* 1994; 130:528-533.

ML II (I-Cell Disease; Mucolipidosis II) 821

General: Autosomal recessive mucolipidosis is a Hurler-like disorder with some radiologic features, striking fibroblast inclusions, and no excess mucopolysacchariduria; abnormal N-acetylglucosamine phosphotransferase.

Ocular: Minimal corneal clouding; glaucoma; megalocornea.

Clinical: Congenital dislocation of the hips; thoracic deformities; hernia; hyperplastic gums; retarded psychomotor development and restricted joint mobility; dysmorphic facies; skeletal deformities; organomegaly; short stature; mental retardation.

Ben-Yoseph, et al. Mucolipidoses II and III variants with normal N-acetylglucosamine I-phosphotransferase activity toward alpha-methylmannoside are due to nonallelic mutations. *Am J Hum Genet* 1992; 50:137-144.

Borit A, et al. Ocular involvement in I-cell disease (mucolipidosis II). Light and electron microscopic findings. *Graefes Arch Klin Exp Ophthalmol* 1976; 198:25-32.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Libert J, et al. Ocular findings in I-cell disease (mucolipidosis type II). *Am J Ophthalmol* 1977; 83:617-628.

ML III (Pseudo-Hurler Polydystrophy; Mucolipidosis III) 822

General: Autosomal recessive disorder, almost indistinguishable biochemically from mucolipidosis II; decreased levels of N-acetylglucosamine phosphotransferase.

Ocular: Increased corneal thickness; wrinkled maculopathy; granular pigmentary changes of fundus; papilledema; hyperopic astigmatism; corneal opacities; retinal vascular tortuosity; visual field defects.

Clinical: Joint stiffness; coarse facial feature; short stature; aortic valve disease; arm and hand deformities; self-mutilation of the distal phalanges; carpal tunnel syndrome.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Traboulsi EI, Maumenee IH. Ophthalmologic findings in mucolipidosis III (pseudo-Hurler polydystrophy). *Am J Ophthalmol* 1986; 102:592-597.

Zammarchi E, et al. Self-mutilation in a patient with mucolipidosis III. *Pediatr Neurol* 1994; 11:68-70.

ML IV (Mucolipidosis IV; Berman Syndrome) 823

General: Storage disease in which corneal clouding is an early sign with no evidence of systemic involvement until age 1 year; autosomal recessive; cases seen in Ashkenazi Jews; abnormal neuraminidase.

Ocular: Corneal clouding; corneal opacities; epithelial edema; retinal atrophy; pale optic nerve; diffuse corneal clouding present at birth or in early infancy.

Clinical: Progressive psychomotor retardation; skeletal dysplasia; facial anomalies.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Folkerth RD, et al. Mucolipidosis IV. Morphology and histochemistry of an autopsy case. *J Neuropathol Exp Neurol* 1995; 54:154-164.

Merin S, et al. Mucolipidosis IV. Ocular, systemic, and ultrastructural findings. *Invest Ophthalmol* 1975; 14:437-448.

MMMM Syndrome (Neuhauser Syndrome; Megalocornea, Macrocephaly, Mental and Motor Retardation) 824

General: Rare

Ocular: Megalcornea

Clinical: Mental retardation; hearing loss; sensorineural complications; hypoplasia; corpus callosum; macrocephaly

Tominaga N, Kamimura N, Matsumoto T, et al. A case of megalocornea mental retardation syndrome complicated with bilateral sensorineural hearing loss. *Pediat Int* 1999; 41:392-394.

Tonaki H: Megalcornea-mental retardation syndrome. *Turk J Pediatr*. 2002; 44: 274-277

Möbius I Syndrome (Hemicrania, Hemiplegic; Hemiplegic-Ophthalmoplegic Migraine; Hemiplegic Familial Migraine) 825

General: Etiology unknown; indirect indications of unilateral cerebral edema due to vasomotor phenomena; occurs in young adults; recovery usually follows after a few days; no clear etiology has been determined, including a vascular theory of embryopathogene-sis, a chromosome translocation, and exposure to teratogens.

Ocular: Extraocular palsy; permanent damage of oculomotor nerve III.

Clinical: Hemicrania; hemiparesis; aneurysm of internal carotid; neoplasia; headache.

D'Cruz OF, et al. Mobius syndrome: evidence for a vascular etiology. *J Child Neurol* 1993; 8:260-265.

Donahue SP, et al. Broad-spectrum Mobius syndrome associated with a I; II chromosome translocation. *Ophthalmic Paediatr Genet* 1993; 14:17-21.

Kankirawatana P, et al. Mobius syndrome in infant exposed to cocaine in utero. *Pediatr Neurol* 1993; 9:71-72.

Lipson T, et al. The Mobius syndrome: etiology, incidence of mental retardation and genetics [Letter]. *J Med Genet* 1990; 27:533-535.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Mobius PJ. Ueber Periodishe Wiederkehrende Oculomotoriuslachennung. *Klin Wochenschr* 1884; 21:604-608.

Möbius II Syndrome (Congenital Facial Diplegia; Congenital Paralysis of the Sixth and Seventh Nerves; Congenital Oculofacial Paralysis; Von Graefes Syndrome) 826

General: Congenital; possibly failure of development of facial nerve cells or primary defect of muscles deriving from first two brachial arches or both; recovery in a few weeks or nonprogressive permanent paralysis of face; asymmetrical; if incomplete, usually spares lower face and platysma.

Ocular: Proptosis; ptosis; weakness of abductor muscles; normal convergence; limitation to internal rotation in lateral movements; esotropia.

Clinical: Facial diplegia; deafness; loss of vestibular responses; webbed fingers or toes; clubfoot.

Abbott RL, et al. Saccadic velocity studies in Mobius syndrome. *Ann Ophthalmol* 1978; 10:619-623.

Fenichel GM. Congenital facial asymmetry (aplasia of facial muscles). In: Fenichel GM, ed. *Clinical pediatric neurology*, 2nd ed. Philadelphia: WB Saunders, 1993:341-342.

Kawai M, et al. The syndrome of Mobius sequence, peripheral neuropathy, and hypogonadotropic hypogonadism. *Am J Med Genet* 1990; 37:578-582.

Menkes JH, Kenneth T. Mobius syndrome. In: Menkes JH, ed. *Textbook of child neurology*, 5th ed. Baltimore: Williams & Wilkins. 1995:309-310.

Merz M, Mojtowicz S. The Mobius syndrome. *Am J Ophthalmol* 1967; 63:837.

Mobius PJ. Uber Angeborene Doppelseitige Abducens-Facialislahmung. *Munch Med Wochenschr* 1888; 35:91.

Pucket CL, Beg SA. Facial reanimation in Mobius syndrome. *South Med J* 1978; 71:1498-1501.

Mohr-Claussen Syndrome (Oral-Facial-Digital Syndrome Type II; OFD Syndrome; Orofaciodigital Syndrome II) 827

General: Rare; autosomal recessive; certain features similar to Papillon-Leage-Psaume, Carpenter, Laurence-Moon-Bardet-Biedl, and Ellis-Van Creveld syndromes (see Papillon-Leage-Psaume Syndrome; Laurence-Moon-Bardet-Biedl Syndrome; Ellis-Van Creveld Syndrome).

Ocular: Epicanthus; bridged chorioretinal colobomata.

Clinical: Clefts and fibroma of tongue; polydactylia; broad nasal bridge; narrow-arched palate; short humerus, femur, and tibia; irregular teeth; hypotonia; mental retardation; deafness; thin and fair hair; cerebellar atrophy.

Anneren G, et al. Abnormalities of the cerebellum in oro-facio-digital syndrome II (Mohr syndrome). *Clin Genet* 1990; 38:69-73.

Claussen O. Et Arvelig Syndrome Omfattende Tungemisdannelse og Polydaktyli. *Nord Med* 1946; 30: 1147.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Moller-Barlow Disease 828

General: Vitamin C deficiency in children.

Ocular: Hemorrhages around and, in rare cases, in eyes; yellow-brown discoloration from hemosiderin may remain for some time after resolution of a subconjunctival hemorrhage; hemorrhage in eyelids, conjunctiva, anterior chamber, and retina; proptosis in infantile scurvy.

Clinical: Tenderness of the lower extremities; ecchymoses.

Gabay C, et al. A case of scurvy presenting with cutaneous and articular signs. *Clin Rheumatol* 1993; 12:278-280.

Goskowicza M, Eichenfield LF. Cutaneous findings of nutritional deficiencies. *Curr Opin Pediatr* 1993; 5:441-445.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Molluscum Contagiosum 829

General: Etiologic agent of this disease is a poxvirus that can cause proliferative skin lesions anywhere on the body; commonly found in patients who are immunosuppressed.

Ocular: Lesions of lid, lid margin, conjunctiva, and cornea; conjunctivitis; keratitis; corneal ulcer.

Clinical: Well-defined, pearly appearing papules with umbilicated centers of varying size (3 to 10 mm); eczematization of the surrounding skin.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Hindaal CH, Van Bijsterveld OP. Molluscum contagiosum of the palpebral conjunctiva. *Ophthalmologica* 1979; 178: 137-141.

Kohn SR. Molluscum contagiosum in patients with acquired immunodeficiency syndrome. *Arch Ophthalmol* 1987; 105:458.

Monbrun-Benisty Syndrome (Ocular Stump Causalgia) 830

General: Sympathetic irritation of resected sympathetic fiber to the eye; occurs after trauma of eye.

Ocular: Severe refractory pain of orbital cavity.

Clinical: Pain of face and the corresponding hemicranium; congestion and hyperhidrosis of region involved.

Barre JA, Klein M. Un Cas de Syndrome de Monbrun-Benisty (Causalgie du Moignon Oculaire) Gueri par Gasserectomie. *Revotoneuro-Ophthalmol* 1934; 11:755-758.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Monofixation Syndrome (Blind Spot Syndrome; Primary Monofixation) 831

General: No hereditary factor; uncommon.

Ocular: Deviation of eight prism diopters or less by simultaneous prism and cover test; central scotoma; stereopsis; good fusional vergences found in patients with congenital esotropia; unilateral syphilitic optic perineuritis (rare); congenital esotropia (inherited in a multifactorial fashion).

Clinical: Syphilis (rare).

Bolet RV. Development of mono fixation syndrome in congenital esotropia. *J Pediatr Ophthalmol Strabismus* 1981; 18:49-51.

Fletcher WA, et al. Acute Idiopathic blind spot enlargement. *Arch Ophthalmol* 1988; 106:44-49.

Hahn E, et al. Factors associated with binocular single vision in microtropia/monofixation syndrome. *Can J Ophthalmol* 1991; 26:12-17.

McBurney J, Rosenberg M. Unilateral syphilitic optic perineuritis presenting as the big blind spot syndrome. *J Clin Neuro-Ophthalmol* 1987; 7:167.

Morris RJ, et al. Fusion after surgical alignment of long-standing strabismus in adults. *Ophthalmology* 1993; 100: 135-138.

Parks MM. The monofixation syndrome. *Trans Am Ophthalmol Soc* 1969; 67:809-855.

Scott MJ, et al. Prevalence of primary mono fixation syndrome in parents of children with congenital esotropia. *J Pediatr Ophthalmol Strabismus* 1994; 31:298-301.

Moraxella Lacunata 832

General: Gram-negative rod; causes chronic angular blepharoconjunctivitis; without treatment, may persist for months or years; normally found in flora of respiratory tract; seen more frequently in alcoholics and those with poor sanitary habits; *Moraxella* organisms produce proteases, although those are not related directly to their pathogenetic mechanism.

Ocular: Catarrhal angular conjunctivitis; corneal ulcer; hypopyon, chronic blepharitis; eczema; lateral canthal skin erythema; iridocyclitis.

Clinical: Alcoholism; impaired nutrition; dermatitis.

Baum J, et al. A survey of Moraxella corneal ulcers in a derelict population. *Am J Ophthalmol* 1980; 90:476-480.

Burd EM. Bacterial keratitis and conjunctivitis. In: Smolin G, Thoft RA, eds. *The Cornea*. Boston: Little, Brown and Company, 1994:20-21.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

van Bijsterveld OP. The incidence of Moraxella on mucous membranes and the skin. *Am J Ophthalmol* 1972; 74: 72-76.

Morgagni Syndrome (Hyperostosis Frontalis Interna Syndrome; Intracranial Exostosis; Metabolic Craniopathy) 833

General: Dominant inheritance; onset around age 45 years; occurs almost exclusively in females. Cataract; optic nerve injury within the optic canal by bony protrusions, with resulting blindness.

Clinical: Hyperostosis frontalis interna; obesity (mainly trunk and proximal portions of limbs); hirsutism; menstrual disorders; hypertension; arteriosclerosis; headache; hypertrichosis; no case of male-to-male transmission is known; hyperprolactinemia.

Falconer MA, Pierard BE. Failing vision caused by a bony spike compressing the optic nerve within the optic canal. Report of two cases associated with Morgagni's syndrome benefited by operation. *Br J Ophthalmol* 1950; 34:265.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Morgagni GB. *De Sedibus et Causis Morborum Peranatomen Indagatis Libri Quinque (2 volumes).* Venetiis: Typog. Remondiniana, 1761. Alexander B, English translation, 3 volumes. London, 1769. *(Selections* in *Med Classics* 1940; 4:640).

Pawlikowsky M, et al. Hyperostosis frontalis, galactorrhea, hyperprolactinemia, and Morgani-Stewart-Morel syndrome [Letter]. *Lancet* 1983; 1:474.

Morning Glory Syndrome (Hereditary Central Glial Anomaly of the Optic Disk) 834

General: No hereditary factor; rare; anomaly of optic disk with deep excavation resembling the flower for which syndrome is named.

Ocular: Strabismus; abnormality of embryologic development of anterior chamber (anterior chamber cleavage syndrome); remnants of hyaloid system; chorioretinal pigment surrounding optic disk; narrow branches of retinal arteries at edge of optic

disk; retinal exudates and detachment; subretinal hemorrhages and retinal neovascularization; enlarged pink optic disk, funnel shaped with a central white fluffy dot; nerve head surrounded by elevated annulus of chorioretinal pigment, unilateral.

Clinical: Midline cranial facial defects such as hypertelorism, cleft lip/palate, basal encephalocele, agenesis of corpus callosum, sphenoid encephalocele defects in the floor of the sella turcica; cranial, facial, and neurologic associations; pituitary dwarfism; association with the CHARGE syndrome.

Caprioli J, Lesser RL. Basal encephalocele and morning glory syndrome. *Br J Ophthalmol* 1983; 67:349.

Chang S, et al. Treatment of total retinal detachment in morning glory syndrome. *Am J Ophthalmol* 1984; 97: 596-600.

Eustis HS, et al. Morning glory syndrome in children. Association with endocrine and central nervous system anomalies. *Arch Ophthalmol* 1994; 112:204-207.

Handemann M. Erbliche, Vermutlich Angeborene Zentrale Gliose Entartung des Sehnerven mit Besonderer Beteiligung der Zentralgefasse. *Klin Monatsbl Augenheilkd* 1929; 83:145.

Itakura T, et al. Bilateral morning glory syndrome associated with sphenoid encephalocele. Case report. *J Neurosurg* 1992; 77:949-951.

Kindler P. Morning glory syndrome: unusual congenital optic disc anomaly. *Am J Ophthalmol* 1970; 69:376.

Morphea (Localized Scleroderma; Circumscribed Scleroderma) 835

General: Localized chronic connective tissue disease of unknown etiology; etiology remains unknown, although there is a possible association with *Borrelia burgdorferi* infection.

Ocular: Circumscribed plaque like lesions of the eyelid; prevalent in females; onset usually in second to fourth decades of life; onset occasionally associated with trauma, pregnancy, or menopause.

Clinical: Firm skin plaques over entire body, but most frequently on trunk, lower extremities, upper extremities, face, and genitalia; abdominal pain; migraine; generalized joint pain; renal crisis; Raynaud phenomenon; systemic sclerosis; eosinophilia; positive antinuclear factor; increased immunoglobulin (immunoglobulin G [IgG]); seizures; skin sclerosis; alterations in tryptophan metabolism.

Baba FE, et al. Morphea of the eyelids. *Ophthalmology* 1982; 89:1285-1288.

Ghersetich I, et al. Localized scleroderma. *Clin Dermatol* 1994; 12:237-242.

Maricq HR. Capillary abnormalities, Raynaud's phenomenon, and systemic sclerosis in patients with localized scleroderma. *Arch Dermatol* 1992; 128:630-632.

Trevisan G, et al. Borrelia burgdorferi and localized scleroderma. *Clin Dermatol* 1994; 12:475-479.

West RH, Barnett AJ. Ocular involvement in scleroderma. *Br J Ophthalmol* 1979; 63: 845-847.

Wigley FM. Clinical aspects of systemic and localized scleroderma. *Curr Opin Rheumatol* 1994; 6:628-636.

Morquio Syndrome (Morquio-Brailsford Syndrome; Brailsford-Morquio Dystrophy; Familial Osseous Dystrophy; Keratosulfaturia; MPS IV; Mucopolysaccharidosis IV; Spondyloepiphyseal Dysplasia; Osteochondrodystrophia Deformans; Infantile Hereditary Chondrodysplasia; Hereditary Poly topic Enchondral Dysostosis; Hereditary Osteochondrodystrophy; Eccentro-Osteochondrodysplasia; Dysostosis Enchondralis Meta- Epiphysaria; Morquio-Ullrich Syndrome; Atypical Chondrodystrophy; Chondrodystrophia Tarda; Chondro-Osteodystrophy) 836

General: Autosomal recessive dystrophy of cartilage and bone; slight predilection for males; apparent between ages 4 and 10 years; excess production of keratosulfate (see Hurler Syndrome; Hunter Syndrome; Sanfilippo-Good Syndrome; Scheie Syndrome; Maroteaux-Lamy Syndrome); autosomal recessive; abnormal N-acetylgalactosamine-G-sulfate sulfatase.

Ocular: Enophthalmos; ptosis; excessive tear secretion; ocular hypotony; miosis; occasionally hazy cornea; bushy eyebrows; optic nerve atrophy; moderate-to-late corneal clouding.

Clinical: Dwarfism; skeletal deformities (progressive); delayed ossification of epiphyses; decreased muscle tone; deafness; weak extremities; waddling gait; coarse broad mouth; spaced teeth; aortic regurgitation; normal intelligence.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders. 2000.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kenyon KR, et al. The systemic mucopolysaccharidoses: ultrastructural and histochemical studies of conjunctiva and skin. *Am J Ophthalmol* 1972; 73:811.

MacDessi JJ, et al. Spondylo-epiphyseal dysplasia with ocular changes: report of two "new" variants in two different families. *Pediatr Radiol* 1978; 7:22G-228.

Morquio L. Sur une Forme de Dystrophie Osseuse Familiale. *Bull Soc Pediatr (Paris)* 1929; 27:145-152.

Mort d'Amour Syndrome (Death of Love Syndrome) 837

General: Sudden death during sexual intercourse.

Ocular: Pupillary dilation.

Clinical: Hypertension; arrhythmia; heart ischemia; rupture of cerebral aneurysm.

Heggveit HA. La mort d'amour. *Am Heart J* 1965; 69:287-294.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Mosse Syndrome (Polycythemia-Hepatic Cirrhosis Syndrome) 838

General: Unknown etiology.

Ocular: Scleral icterus; marked retinal venous tortuosity and dilation; retinal artery occlusion (occasionally); papilledema.

Clinical: Thrombosis portal vein secondary to polycythemia; hepatosplenomegaly; ascites; clinical features of liver cirrhosis.

Barbas AP. Surgical problems associated with polycythemia. *Br J Hosp Med* 1980; 23:289-290,292,294.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Mosse M. Uber Policythamie mit Urobilinikterus und Milztumor. *Dtsch Med Wochenschr* 1907; 33:2175.

Moyamoya Disease (Multiple Progressive Intracranial Arterial Occlusion) 839

General: Almost exclusively seen in Japanese infants and children; cerebrovascular disorder that results in occlusion of the large vessels at the base of the brain; slight female prevalence; collateral vascular networks (secondary to bilateral carotid occlusive disease) resembling puffs of smoke (Moyamoya in Japanese).

Ocular: Hemianopsia; nystagmus; papilledema; central retinal vein occlusion; visual field defects; amaurosis fugax; diplopia; optic pallor; ischemic chiasmal syndrome; bilateral renal artery stenosis.

Clinical: Loss of consciousness; seizures; hemiplegia; hemiparesis; intracranial hemorrhage; mental retardation; speech disturbances; unsteady gait; headache; psychiatric manifestations; focal epileptic attacks; chronic cerebrovascular disorder; intracerebral hemorrhage; pituitary adenoma (association).

Ahmadi J, et al. Ischemic chiasmal syndrome and hypopituitarism associated with progressive cerebrovascular occlusive diseases. *Am J Neuroradiol* 1984; 5:367.

Arita K, et al. Moyamoya disease associated with pituitary adenoma-report of two cases. *Neurol Med Chir* 1992; 32:753-757.

Nakano T, et al. Moyamoya disease associated with bilateral renal artery stenosis. *Acta Paediatr Jpn* 35:354-357, 1993.

Noda S, et al. Ocular symptoms of Moyamoya disease. *Am J Ophthalmol* 1987; 103:812-816.

Slamovits TL, et al. Moyamoya disease with central retinal vein occlusion. *J Clin Neuro-Ophthalmol* 1981; 1: 123-127.

Ueki K, et al. Moyamoya disease: the disorder and surgical treatment. *Mayo Clin Proc* 1994; 69:749-757.

Yamada H. Moyamoya disease in monocular twins: case report. *J Neurosurg* 1980; 53: 109-112.

Moynahan Syndrome (XTE; Xeroderma, Talipes, and Enamel Defect) 840

General: Autosomal dominant inheritance; xeroderma, talipes, and enamel defect.

Ocular: Absence of eyelashes of lower lid.

Clinical: Cleft palate; hypohidrosis; defective enamel; nail anomalies; coarse and dry hair; short-lasting skin bullae.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Moynahan E. XTE (xeroderma, talipes and enamel defect): a new heredo-familial syndrome. Two cases. Homozygous inheritance of a dominant gene. *Proc R Soc Med* 1970; 63:447-448.

Mucocele (Pyocele) 841

General: Accumulation and retention of mucoid material within the sinus as a result of continuous or periodic obstruction of the sinus ostium.

Ocular: Paralysis of extraocular muscles; exophthalmos; lacrimation; erosion of bony walls of orbit; decreased visual acuity; diplopia; elevation of lower lid; ptosis; compression optic neuropathy; globe distortion; enophthalmos; epiphora; scleral indentation; choroidal folds; discharging lesion of the upper lid; pseudotelecanthus; spontaneous nontraumatic enophthalmos; local anesthesia.

Clinical: Headaches; epidural abscess; subdural empyema; meningitis; brain abscess; occlusion of nasal passage; loosening of teeth.

Abrahamson IA Jr, et al. Frontal sinus mucocele. *Ann Ophthalmol* 1979; 11:173-178.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kaufman SJ. Orbital mucopyoceles: two cases and a review. *Surv Ophthalmol* 1981; 25:253-262.

Moryyama H, et al. Studies on mucoceles of ethmoid and sphenoid sinuses. *J Laryngol Otol* I992; 106:23-27.

Ormerod LD, et al. Ophthalmic manifestations of maxillary sinus mucoceles. *Ophthalmology* 1987; 94:1013-1019.

Mucocutaneous Lymph Node Syndrome (MLN Syndrome; Kawasaki Disease) 842

General: Multisystem syndrome with worldwide distribution; occurs from age 2 months\ to 9 years; increased incidence in summer; etiology unknown, but allergic reactions to chemicals or abnormal reactions to numerous infections have been suggested.

Ocular: Severe conjunctival congestion and hyperemia (88%); uveitis; dacryocystitis; anterior uveitis (commonly bilateral); punctate keratitis.

Clinical: Fever (1 to 2 weeks; does not respond to antibiotics) "strawberry tongue"; red palms and soles; indurative edema; membranous desquamation from fingertips; polymorphous exanthema; arthritis; myocarditis; enlarged cervical lymph nodes.

Bums JC, et al. Anterior uveitis associated with Kawasaki syndrome. *Pediatr Infect Dis* 1985; 4:258-261.

Kawasaki T, et al. A new infantile acute febrile mucocutaneous lymph node syndrome (MLNS) prevailing in Japan. *Pediatrics* 1974; 54:271.

Mauriello JA, et al. Dacryocystitis following Kawasaki's disease. *Ophthal Plast Reconstr Surg* 1986; 2:209.

Naoe S, et al. Kawasaki disease, with particular emphasis on arterial lesions. *Acta Pathol Jpn* 1991; 41:785-790.

Puglise JV, et al. Ocular features of Kawasaki's disease. *Arch Ophthalmol* 1982; 100:1101-1103.

Raimer SS, Sanchez RL. Vasculitis in children. *Semin Dermatol* 1992; 11:48-56.

Mucormycosis (Phycomycosis) 843

General: Acute, often fatal infection caused by saprophytic fungi; associated with diabetes mellitus and ketoacidosis.

Ocular: Corneal ulcer; striate keratopathy; ptosis; panophthalmitis; proptosis; cellulitis of orbit; immobile pupil; retinitis; optic neuritis; paralysis of extraocular muscles; central retinal artery thrombosis.

Clinical: Epistaxis; nasal discharge; facial pain; facial palsies; anhidrosis; cranial nerve or peripheral motor and sensory nerve deficits may occur.

Albert DM, et al. Orbitofacial mucormycosis with unusual pathological features. *Br J Ophthalmol* 1979; 63:699-703.

Bullock JD, et al. Two cases of orbital phycomycosis with recovery. *Am J Ophthalmol* 1974; 78:811.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gutierrez Diaz A, et al. Orbital phycomycosis. *Ophthalmologica* 1981; 182: 165-170.

Sponsler TA, et al. Ocular invasion in mucormycosis. *Surv Ophthalmol* 1992; 36:345-350.

Mucous Membrane Pemphigoid 844

General: Immune mediated disease characterized by autoantibodies to the basement membrane zone at the sub-epithelial junction of mucous membranes

Ocular: Progressive cicatrizing conjuncitivitis; symblepharon; corneal clouding

Clinical: Nasal and oral mucosa cicitrization; trachea and esophagus cicitrization

Jabs DA, Rosenbaum JT, Foster CS, et al. Guidelines for the use of immunosuppressive drugs in patients with ocular inflammatory disorders: recommendations of an expert panel. *Am J Ophthalmol* 2000; 130: 492-513

Thorne JE, Anhalt GJ, Jabs DA: Mucous Membrane Pemphigoid and Pseudoemphigoid. *Ophthalmology* 2004; 11:45-52.

Mucus Fishing Syndrome 845

General: Excessive mucus production created by mechanical damage of conjunctival epithelium; epithelial injury created by mechanical removal of excess mucus, which leads to further increased mucus production, causing a continuous cycle; initial mucus pro-duction may be associated with an underlying ocular disease, such as pterygium, keratoconus, blepharitis, or keratoconjunctivitis.

Ocular: Excessive mucus production; blepharitis; pterygium; keratoconjunctivitis; keratoconus; corneal foreign body; floppy lid syndrome; map-dot fingerprint dystrophy; squamous cell cancer of conjunctiva.

Clinical: None.

McCulley JP, et al. Mucus fishing syndrome. *Ophthalmology* 1965; 92:1262-1265.

McCulley JP, et al. Classification of chronic blepharitis. *Ophthalmology* 1982; 89:1173-1180.

Muir-Torre Syndrome 846

General: Rare; autosomal dominant; characterized by sebaceous tumors and internal malignancies

Ocular: Sebaceous tumor of the eyelids

Clinical: Gastrointestinal, breast and genitourinary malignancies; sebaceous neoplasms

Font RL, Rishi K: Sebaceous gland adenoma of the tarsal conjunctiva in a patient with Muir-Torre syndrome. *Ophthalmology* 2003; 110: 1833-1836

Mathiak M, Rutten A, Mangold E, et al. Loss of DNA mismatch repair proteins in skin tumors form patients with Muir-Torre syndrome and MSH2 or MLH1 germline mutations: establishment of immunohistochemical analysis as a screening test. *Am J Surg Pathol* 2002; 26: 338-343

Mulibrey Nanism Syndrome (Perheentupa Syndrome) 847

General: Autosomal recessive inheritance; progressive growth failure; myocardial fibrosis.

Ocular: Alternating esotropia and exotropia; yellowish retinal dots and scattered pigment dispersion in clusters (especially in the midperiphery); drusen of Bruch membrane; hypoplasia of choriocapillaries (diagnostic sign).

Clinical: Triangular face with bulging forehead and low, broad nasal bridge; hypotonia; hepatomegaly; ascites; pulmonary congestion; cardiac enlargement with possible heart failure; pericardial constriction; dwarfism; immunoglobulin deficiency; isolated growth hormone deficiency.

Haraldsson A. Antibody deficiency and isolated growth hormone a deficiency in a girl with Mulibrey nanism. *Eur J Pediatr* 1993; 152:509-512.

Lapunzina P, et al. Mulibrey nanism: three additional patients and a review of 39 patients. *Am J Med Genet* 1995; 55: 349-355.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Perheentupa J, et al. Mulibrey nanism dwarfism with muscle, liver, brain and eye involvement. *Acta Paediatr Scand* 1970; 59:74.

Raitta C, Perheentupa J. Mulibrey nanism: an inherited dysmorphic syndrome with characteristic ocular findings. *Acta Ophthalmol Suppl (Copenh)* 1974; 123:162.

Multiple Endocrine Neoplasia 2B or 3 (MEN 2B or 3) 848

General: Autosomal dominant inheritance; multiple endocrine neoplasia type 3 (MEN3) has been separated from MEN2 because of low incidence of associated parathyroid disease in these cases; poor prognosis; several different point mutations in the RET protooncogene on chromosome 10 have been associated with the multiple endocrine neoplasia type 2 syndromes.

Ocular: Prominent corneal nerves in clear stroma; diffuse, nodular thickening of eyelids; nasal displacement of lacrimal puncta; rostral displacement of eyelashes; eversion of eyelids; subconjunctival neuromas; thickened conjunctival nerves; decreased tear formation; prominent eyebrows; impaired pupillary dilation; thickened iris nerves; increased intraocular pressure (rare); localized orbital neurofibromas (orbit, conjunctiva); lesions of the tongue resembling neuromas.

Clinical: May be present at birth or develop later; 50% show complete syndrome of multiple neuromas (lips, tongue, eyelids), bumpy lips, pheochromocytoma, and medullary carcinoma; others exhibit variable combinations of the preceding, without the pheochromocytoma; diarrhea; marfanoid habitus.

Colombo CG, Watson AG. Ophthalmic manifestations of multiple endocrine neoplasia, type three. *Can Ophthalmol* 1976; 11:290-294.

Dennehy PJ, et al. Relationship of familial prominent corneal nerves and lesions of the tongue resembling neuromas to multiple endocrine neoplasia type 2B. *Am J Ophthalmol* 1995; 120:456-461.

Holloway KB, Flowers FP. Multiple endocrine neoplasia 2B (MEN 2B)/MEN3. *Dermatol Clin* 1995; 13:99-103.

Kalina PH, et al. Isolated neurofibromas of the conjunctiva. *Am J Ophthalmol* 1991; 111:694-698.

Meyer DR, Wobig JL. Bilateral localized orbital neurofibromas. *Ophthalmology* 1992; 99: 1313-1317.

Norum RA, et al. Linkage of the multiple endocrine neoplasia type 2B gene (MEN 2B) to chromosome 10 markers linked to MEN2A. *Genomics* 1990; 8:313-317.

Schweitzer NMJ, Van Der Pal BAE. Multiple mucosal neuroma (MMN) or multiple endocrine neoplasia (MEN) type 3 syndrome: ocular manifestations. A case report. *Doc Ophthalmol* 1977; 44:151.

Spector B, et al. Histologic study of the ocular lesions in multiple endocrine neoplasia syndrome type IIb. *Am J Ophthalmol* 1981; 91:204-215.

Multiple Evanescent White-Dot Syndrome (MEWDS) 849

General: Unilateral disease including multiple white dots at the level of the pigment epithelium; etiology unknown; recurrent; no systemic involvement.

Ocular: Retinal pigment granularity in the macula; reduced visual acuity; vitreitis; loss of retinal sensitivity; white dots of pigment epithelium; uveitis; blind spot enlargement; choroidal neovascularization.

Clinical: None.

Aaberg TM, et al. Recurrences and bilaterality in the multiple evanescent white-dot syndrome. *Ophthalmology* 1985; 100:29-37.

Callanan D, Gass JD. Multifocal choroiditis and choroidal neo vascularization associated with the multiple evanescent white dot and acute idiopathic blind spot enlargement syndrome. *Ophthalmology* 1992; 99:1678-1685.

Jampol LM, et al. Multiple evanescent white dot syndrome I. Clinical findings. *Arch Ophthalmol* 1984; 102:671.

Keunen JEE, van Norren D. Foveal densitometry in the multiple evanescent white-dot syndrome. *Ophthalmology* 1988; 105:561-562.

Khorram KD, et al. Blind spot enlargement as a manifestation of multi focal choroiditis. *Arch Ophthalmol* 1991; 109: 1403-1407.

Tejada PP, et al. Multiple evanescent white dot syndrome. *Ann Ophthalmol* 1993; 25:216-218.

Multiple Lentigines Syndrome (LEOPARD Syndrome) 850

General: Familial occurrence; classic features include lentigines (small focal hyperpigmentations of skin), electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonary stenosis, abnormal genitalia, retardation of growth, and deafness (LEOPARD).

Ocular: Hypertelorism; exophthalmos; epicanthal folds; strabismus; nystagmus; keratoconus.

Clinical: Lowset ears; receding chin; deafness; lentigines; pulmonary stenosis; genital abnormalities; growth retardation; skeletal malformations (bony fusion involving cervical vertebrae, ossicles, carpal and tarsal bones, scoliosis); hyposmia; heart murmur; mental retardation; hypospadias; congenital heart defect; thoracic deformities; respiratory insufficiency.

Gorlin RJ, et al. Multiple lentigines syndrome. *Am J Dis Child* 1969; 117:652.

Howard RO. Premature cataracts associated with generalized lentigo. *Ophthalmology* 1980; 87:234-239.

Peter JR, Kemp JS. Leopard syndrome: death because of chronic respiratory insufficiency. *Am J Med Genet* 1990; 37:340-341.

Swanson SL, et al. Multiple lentigines syndrome: new findings of hypogonadotrophism, hyposmia, and unilateral renal agenesis. *J Pediatr* 1971; 78:1037.

Multiple Mucosal Neuromata with Endocrine Tumors Syndrome (Multiple Mucosal Neuromas, Pheochromocytoma, and Medullary Thyroid Carcinoma Syndrome) 851

General: Pheochromocytoma can be inherited and found in combination with neurofibromatosis and other brain tumors (meningioma, spongioblastoma, ependymoma, astrocytoma, cerebellar hemangioblastoma), multiple mucosal neuromas, and medullary carcinoma of the thyroid.

Ocular: Conjunctival neuromas; multiple, white, myelinated nerve fibers in corneal stroma arising at limbus with anastomoses in center of cornea.

Clinical: Neuromas of lips or anterior portion of tongue or, more rarely, of buccal, gingival, or laryngeal mucosa; hypertension; flushing; weakness; sweating; palpitations; headaches.

Burgdorf W. Letter: multiple mucosal neuroma syndrome. *Arch Dermatol* 1975; 111:931.

Paloyan E, et al. familial pheochromocytoma, medullary thyroid carcinoma and parathyroid adenomas. *JAMA* 1970; 214:1443.

Williams ED, Pollock DJ. Multiple mucosal neuromata with endocrine tumors: a syndrome allied to von Recklinghausen's disease. *J Pathol Bacteriol* 1966; 91:71.

Multiple Pterygium Syndrome, Lethal Type (Pterygium, Multiple, Lethal Type) 852

General: Autosomal recessive; nonconsanguineous parents; lethal.

Ocular: Hypertelorism; epicanthal folds.

Clinical: Multiple pterygia involving chin to sternum, cervical, axillary, antecubital, and popliteal areas; flexion contracture of multiple joints; small chest; hydrops; markedly flattened nasal bridge with hypoplastic nasal alae; cleft palate; micrognathia; lowset, malformed ears; short neck with a cystic hygroma on back of neck, head; pulmonary and cardiac hypoplasia; hypoplastic teeth; elongated clavicles; cryptorchidism; foot deformities; arthrogryposis; hypoplasia of the left arm, leg, pelvis, and kidney.

Chen H, et al. Syndrome of multiple pterygia, camptodactyly, facial anomalies, hypoplastic lungs and heart, cystic hygroma, and skeletal anomalies: delineation of a new entity and review of lethal forms of multiple pterygium syndrome. *Am J Med Genet* 1984; 17:809-826.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Spearritt DJ, et al. Lethal multiple pterygium syndrome: report of a case with neurological anomalies. *Am J Med Genet* 1993; 47:45-49.

Willems PJ, et al. Multiple pterygium syndrome with body asymmetry. *Am J Med Genet* 1993; 47:106-111.

Multiple Sulfatase Deficiency 853

General: Autosomal recessive

Ocular Congenital peripheral cataract; retinal degeneration; cherry red-spot of macula

Clinical: Progressive neurologic disease with death in the teenage years; ichthysis

Johnson JL: prenatal diagnosis of molybdenum cofactor deficiency and isolated sulfite oxidate deficiency. *Prenat Diag* 2003; 23: 6-8

Mulvihill-Smith Syndrome 854

General: Progeroid disorder.

Ocular: Keratoconus; conjunctivitis.

Clinical: Patients have short stature, microcephaly, unusual facies, numerous pigmented nevi, hypodontia, sensorineural hearing loss, immunodeficiency (low IgG), and a high-pitched voice.

Bartsch O, et al. Mulvihill-Smith syndrome: case report and review. *J Med Genet* 1994; 31:707-711.

Okashi N, et al. Premature aging and immunodeficiency: Mulvihill-Smith syndrome? *Am J Med Genet* 1997; 45: 597-600.

Rau S, Duncker GI. Keratoconus in Mulvihill-Smith syndrome. *Klin Monatsbl Augenheilkd* 1994; 205:44-46.

Mumps 855

General: Viral infection.

Ocular: Conjunctivitis; keratitis; corneal ulcer; tenonitis; exophthalmos; microphthalmos; optic atrophy; optic neuritis; papillitis; scleritis; uveitis; cortical blindness; congenital punctal occlusion; paralysis of extraocular muscles; dacryoadenitis; iritis; paralysis of accommodation; internal and external ophthalmoparesis.

Clinical: Affects the parotid glands, but infection of other glandular tissue occurs, including the lacrimal gland and testicles; encephalitis; meningitis.

AI Rashid RA, Cress C. Mumps uveitis complicating the course of acute leukemia. *J Pediatr Ophthalmol* 1977; 14: 100-102.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Meyer RF, et al. Mumps conjunctivitis. *Am J Ophthalmol* 1974; 78:331.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol.* 5, *part* 2, 4th ed. Baltimore: Williams & Wilkins, 1982:4062.

Strong LE, et al. Bilateral retrobulbar neuritis secondary to mumps. *Am J Ophthalmol* 1974; 78:331.

Muscular Dystrophy, Congenital, with Infantile Cataract and Hypogonadism 856

General: Autosomal recessive; rare; reported in isolated Norwegian village.

Ocular: Infantile cataract.

Clinical: Ovarian agenesis in females; Klinefelter syndrome in males; congenital muscular dystrophy.

Bassoe HH. Familial congenital muscular dystrophy with gonadal dysgenesis. *J Clin Endocrinol* 1956; 16:1614-1621.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Myasthenia Gravis, Neonatal or Infantile (Infantile Myasthenia Gravis; Neonatal Myasthenia Gravis) 857

General: Occurs in newborn of myasthenic mother; caused by compound in circulation received through placenta (see Erb-Goldflam Syndrome).

Ocular: Transient diplopia; ptosis of upper eyelids; internuclear ophthalmoplegia.

Clinical: Excessive fatigue musculature; symptoms appear and increase as day progresses; expressionless face; sagging jaw; sucking, swallowing, and respiratory difficulties.

Jay WM, et al. Internuclear ophthalmoplegia with downshoot in myasthenia gravis. *J Clin Neuro-Ophthalmol* 1987; 7:74.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981,577.

Papazian O. Transient neonatal myasthenia gravis. *J Child Neurol* 1992; 7:135-141.

Stickroot FL, et al. Myasthenia gravis occurring in an infant born of a myasthenic mother. *J Am Med Assoc* 1942; 120: 1207-1209.

Mycosis Fungoides Syndrome (Sézary Syndrome; Malignant Cutaneous Reticulosis Syndrome) 858

General: Lymphoma characterized by abnormal lymphocytes having hyperchromatic, hyperconvoluted nuclei; malignant, cutaneous T-cell lymphoma, which initially presents as a nonspecific erythematous cutaneous eruption that progresses to form plaques and tumors.

Ocular: Thick and swollen eyelids; ectropion; blepharitis; loss of eyelashes; keratoconjunctivitis; uveitis; retinal edema and exudates; papilledema; exophthalmos; retinal hemorrhage; endophthalmitis; pupillary dilatation; scleritis; corneal opacity; optic disk swelling.

Clinical: Pruritus followed by thickening and edema of the skin; dermatitis exfoliativa; eczema; pyoderma; pigmentary changes of body and extremities (mottled appearance); hyperhidrosis.

Deutsch AR, Duckworth JK. Mycosis fungoides of upper lid. *Am J Ophthalmol* 1968; 65:884-888.

Forester HC. Mycosis fungoides with intraocular involvement. *Trans Am Acad Ophthalmol Otolaryngol* 1960; 64:308.

Fraunfelder FT, Roy FT. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Leitch, R. J, et al. Ocular involvement in mycosis fungoides. *Br J Ophthalmol* 1993; 77: 126-127.

Meekins B, et al. Cutaneous T-cell lymphoma presenting as a rapidly enlarging ocular adnexal tumor. *Ophthalmology* 1985; 92:1288-1293.

Myelinated Optic Nerve Fibers 859

General: Autosomal dominant; transmission has been reported as autosomal recessive.

Ocular: White area adjacent to the disk caused by myelin sheath; pseudopapilledema.

Clinical: None.

Francois J. *Heredity* in *Ophthalmology.* St. Louis: CV Mosby, 1961:945.

Mann I. *Developmental abnormalities of the eye,* 2nd ed. Philadelphia: JB Lippincott, 1957.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 1, 4th ed. Baltimore: Williams & Wilkins, 1982.

Myopathy, Mitochondrial, with Cataract 860

General: Autosomal dominant.

Ocular: Early-onset bilateral cataracts; severe ophthalmoplegia; mitochondrial abnormalities in the inferior oblique muscle.

Clinical: Facial weakness; myocardial and skeletal myopathy of the mitochondrial type.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pepin B, et al. Familial mitochondrial myopathy with cataract. *J Neurol* Sci 1980; 45: 191-203.

Myopia, Infantile Severe 861

General: Autosomal recessive; consanguineous parents.

Ocular: High myopia; optic disk cupping.

Clinical: None.

Drack AV. Myopia. In: Wright KW; ed. *Pediatric Ophthalmology and Strabismus*. St. Louis: Mosby, 1995:555.

Karlsson JL. Evidence for recessive inheritance of myopia. *Clin Genet* 1975; 7:197-202.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Myopia, Sex-Linked 862

General: Sex-linked; linkage to factor VIII gene; probable location Xq28.

Ocular: Myopia; hemeralopia; deuteranopia in males; hypoplasia of the optic nerve head.

Clinical: Short stature.

Bartsocas CS, Kastrantas AD. Sex-linked form of myopia. *Hum Hered* 1981; 31:199-200.

Haim M, et al. X-linked myopia in a Danish family. *Acta Ophthalmol* 1988; 66:450-456.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Schwarz M, et al. Bornholm eye disease-linkage to DNA markers on the distal part of Xq. *Clin Genet* 1990; 38: 281-286.

Myopia-Ophthalmoplegia Syndrome 863

General: Sex-linked; characteristics seen in males; carried by females.

Ocular: Ptosis; myopia; complete or partial ophthalmoplegia; abnormal pupil; progressive degeneration of retina and choroid.

Clinical: Patellar reflex absent; Achilles reflex absent; spina bifida; cardiac defects; absent deep tendon reflex in carriers only.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Ortiz de Zarate JC. Recessive sex-linked inheritance of congenital external ophthalmoplegia and myopia coincident with other dysplasias. *Br J Ophthalmol* 1966; 50:606-607.

Myotonic Dystrophy Syndrome (Myotonia Atrophica Syndrome; Dystrophia Myotonica; Curschmann-Steinert Syndrome) 864

General: Rare autosomal dominant disease; onset about age 20 years; condition is worsened by administration of neostigmine (Prostigmin); associated with an unstable deoxyribonucleic acid (DNA) sequence composed of varying numbers of CTG triplet repeats (which allows a specific molecular test for this disorder).

Ocular: Mild ptosis (occasionally); myotonic cataract with small, dotlike subcapsular cortical opacities during early stage, with polychromatic properties on biomicroscopic examination; corneal epithelial dystrophy; loss of corneal sensitivity; tapetoretinal degeneration; macular red spot; macular degeneration; chorioretinitis; pilomatrixomas; ocular hypotony; pattern pigmentary changes; abnormal saccades.

Clinical: Progressive muscular atrophy with selection of certain muscles (mainly stern-ocleidomastoid, temporalis, dorsiflexor muscles of the ankle, anterior oblique); myotonia; bland facial expression; speech disturbance due to involvement of vocal cords and palatal muscles; dysphagia; endocrine disturbances.

Brooke NM, Cwik VE. Myotonic dystrophy. In: Bradley WG. et al., eds. *Neurology in Clinical practice*, 2nd ed. Boston: Butterworth-Heinemann, 1995:2020-2022.

Gjertsen IK, Sandvig KU, Eide N, et al. Recurrence of secondary opacification and development of a dense posterior vitreous membrane in patients with myotonic dystrophy. *J Cataract Refract Surg* 2003; 29: 213-216.

Kimizuka Y, et al. Retinal changes in myotonic dystrophy. Clinical and follow-up evaluation. *Retina* 1993; 13: 129-135.

Koca MR, et al. Alterations of saccadic eye movements in myotonic dystrophy. *Graefes Arch Clin Exp Ophthalmol* 1992; 230:437-441.

Kuwabara T, Lessell S. Electron microscopic study of extraocular muscles in myotonic dystrophy. *Am J Ophthalmol* 1976; 82:303-339.

Mausolf FA, et al. Morphologic and functional retinal changes in myotonic dystrophy unrelated to quinine therapy. *Am J Ophthalmol* 1972; 74:1141.

Meyer E, et al. Myotonic dystrophy: pathological study of the eyes. *Ophthalmologica* 1980; 181:215-220.

Reardon W, et al. Cataract and myotonic dystrophy: the role of molecular diagnosis. *Br J Ophthalmol* 1993; 77: 579-583.

Myxomas, Spotty Pigmentation, and Endocrine Overactivity Syndrome (NAME Syndrome) 865

General: Autosomal dominant.

Ocular: Eyelid myxomas; pigmented lesions of the caruncle or conjunctival semilunar fold.

Clinical: Cardiac, cutaneous, or mammary myxomas; acromegaly; adrenal, pituitary, and testicular neoplasms; Cushing syndrome; sexual precocity; spotty skin pigmentation.

Danoff A, et al. Adrenocortical micronodular dysplasia, cardiac myxomas, lentigines and spindle cell tumors: report of a kindred. *Arch Intern Med* 1987; 147:443.

Kennedy RH, et al. Ocular pigmented spots and eyelid myxomas. *Am J Ophthalmol* 1987; 104:533-538.

Koopman RJJ, et al. Autosomal dominant transmission of the NAME syndrome. *Hum Genet* 1991; 86:300-304.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Naegeli Syndrome (Melanophoric Nevus Syndrome; Franceschetti-Jadassohn Syndrome; Naegeli Incontinentia Pigmenti; Reticular Pigmented Dermatosis) 866

General: Autosomal dominant; separate entity from incontinentia pigmenti (Bloch-Sulzberger syndrome) based on mode of inheritance; blisters and inflammatory lesions of skin as seen in incontinentia pigmenti not present in Naegeli syndrome; skin pigmentation appears at age 2 years.

Ocular: Nystagmus; strabismus; pseudoglioma; papillitis; optic atrophy.

Clinical: Reticular pigmentary skin changes; hypohidrosis; defective teeth with yellow spotting; keratoderma of palms and soles; disturbed regulation of temperature by reduction of number of sweat glands.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Greither A, Haensch R. Anhidrotic reticular pigment dermatosis with a pustular erythematous initial phase. Transitional cases, failing between Bloch-Sulzberger incontinentia pigmenti and the reticular pigment dermatosis of Naegeli-Franceschetti-Jadassohn. *Schweiz Med Wochenschr* 1970; 100:228.

Naegeli O. Familiarer Chromatophorennaevus. *Schweiz Med Wochenschr* 1927; 8:48.

Naffziger Syndrome (Scalenus Anticus Syndrome) 867

General: Compression of brachial plexus and subclavian artery by scalenus anticus muscle; symptoms vary from mild, with remissions and exacerbations, to severe.

Ocular: Enophthalmos; ptosis (unilateral); small pupil.

Clinical: Weakness of ipsilateral hand grip; reduced ipsilateral biceps reflex; diminution of pulse volume on affected side; numbness and coldness in hand and fingers.

Aligne C, Barral X. Rehabilitation of patients with thoracic outlet syndrome. *Ann Vasc Surg* 1992; 6:381-389.

Atasoy E. Thoracic outlet compression syndrome. *Orthop Clin North Am* 1996; 27:265-303.

Davies AH, et al. Surgical management of the thoracic outlet syndrome. *Br J Surg* 1991; 78:1193-1195.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Karas SE. Thoracic outlet syndrome. *Clin Sports Med* 1990; 9:297-310.

Naffziger HD. Quoted in: Ochsner A, Gage M, DeBakey M. Scalenus anticus (Naffziger) syndrome. *Am J Surg* 1935; 28:669.

Pfaltz CR, Richter HR. Syndrome of vertebrobasilar insufficiency: etiology and pathogenesis of the cochleovestibular symptoms m cerebral circulation disorders. 1969; 193:190.

Nager Syndrome (Nager Acrofacial Dyostosis) 868

General: Rare congenital syndrome characterized by mandibulofacial dyostosis with associated radial defects.

Ocular: Downward slanting palpebral fissures; absent eyelashes in the medial third of the lower lids.

Clinical: Mandibular and malar hypoplasia; dysplastic ears with conductive deafness; variable degrees of palatal clefting; upper limb malformation (often bilateral hand deformities).

Danziger I, et al. Nager's acrofacial dysostosis. Case report and review of the literature. *Int J Pediatr Otorhinolaryngol* 1990; 20:225-240.

Friedman RA, et al. Nager acrofacial dysostosis: management of a difficult airway. *Int J Pediatr Otorhinolaryngol* 1996; 35:69-72.

Pfeiffer RA, Stoess H. Acrofacial dysostosis (Nager syndrome): synopsis and report of a new case. *Am J Med Genet* 1983; 15:255-260.

Nance-Horan Syndrome 869

General: Rare; X-linked oculodental trait; occurs in both sexes; present from birth.

Ocular: Posterior sutural cataracts in females; zonular cataracts in males; decreased visual acuity; nystagmus; microcornea; punctate opacities of cornea; affected males have dense nuclear cataracts and frequently microcorneas; carrier females may show posterior Y sutural cataracts with small corneas and only slightly reduced vision.

Clinical: Dental anomalies, including supernumerary central incisors; short fourth metacarpals; diastemata in females.

Bixler D, et al. The Nance-Horan syndrome: a rare X-linked ocular-dental trait with expression in heterozygous females. *Clin Genet* 1984; 26:30-35.

Horan MB, et al. X-linked cataract and hutchinsonian teeth. *Aust Paediatr J* 1974; 10:98-102.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Nance WE, et al. Congenital sex-linked cataract, dental anomalies, and brachymetacarpalia. *Birth Defects* 1974; 10: 285-287.

van Dorp DB, Delleman JW. A family with X-chromosomal recessive congenital cataract, microphthalmia: a peculiar form of the ear and dental anomalies. *J Pediatr Ophthalmol Strabismus* 1979; 16:166-171.

NARP Syndrome 870

General: Neurogenic weakness (N), ataxia (A), and retinitis pigmentosa (RP) syndrome.

Ocular: Retinitis pigmentosa; bull's-eye maculopathy; salt-and-pepper retinopathy.

Clinical: NARP syndrome patients develop ataxia, weakness, and have retinitis pigmentosa, causing gradual visual field constriction.

Choweres I, et al. Cone and rod dysfunction in the NARP syndrome. *Br J Ophthalmol* 1999; 83:190-193.

Holt IJ, et al. A new mitochondrial disease associated with mitochondrial DNA heteroplasmy. *Am J Hum Genet* 1990; 46:428-433.

Kerrison JB, Biousse V, Newman NJ. Retmopathy of NARP syndrome. *Arch Ophthalmol* 2000; 118:298.

Ortiz, et al. Variable retinal and neurologic manifestations in patients harboring the mitochondrial DNA 8993 mutation. *Arch Ophthalmol* 1993; 111:1525-1530.

Negative Acceleration Syndrome (Hydrostatic Pressure Syndrome; Supersonic Bailout Syndrome) 871

General: At rapid deceleration, blood volume is shifted to the brain and face of the individual due to rapid elevation of intravascular blood pressure; the abrupt rise in hydrostatic pressure simultaneous with the decelerative force results in the signs of cerebral concussion, confusion, retrograde amnesia, and circulatory shock.

Ocular: Periorbital edema; ecchymosis of lids; transient visual loss; subconjunctival hemorrhages; retinal hemorrhages and exudates; retinal arteriolar spasm.

Clinical: Mental confusion; shock; possible perforation of internal organs.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger. 1976.

Lyle DJ, et al. Ophthalmologic hydrostatic pressure syndrome. *Am J Ophthalmol* 1957; 44:652.

Stokes WH. Unusual retinal vascular changes in traumatic injury to the chest. *Arch Ophthalmol* 1932; 7:101.

Nelson Syndrome 872

General: Caused by elevated, incompletely suppressible, or nonsuppressible levels of circulating adrenocorticotropic hormone (ACTH) and possibly melanocyte-stimulating hormone; originally reported as hyperpigmentation with associated pituitary inactivity following bilateral adrenalectomy for Cushing syndrome; appears that age at the time of adrenalectomy is an important predictive factor for development of this disorder.

Ocular: Progressive visual deterioration; recurrent visual field defects with bilateral hemianopsia caused by a pituitary mass diagnosed as chromophobe adenoma.

Clinical: Hyperpigmentation; intermittent headaches.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hoffmann JE, et al. Dissociation of plasma and spinal fluid ACTH in Nelson syndrome. *JAMA* 1974; 228:491.

Kemink L, et al. Patient's age is a simple predictive factor for the development of Nelson's syndrome after total adrenalectomy for Cushing's disease. *J Clin Endocrinol Metab* 1994; 79:887-889.

Nelson DH, et al. ACTH-producing tumor of the pituitary gland. *N Engl J Med* 1958; 259:161.

Nematode Ophthalmia Syndrome (Visceral Larva Migrans Syndrome; Toxocariasis) 873

General: Usually found in children; invasion by larvae of *Toxocara canis* and *Toxocara cati* of viscera and eyes; pronounced eosinophilia; as many as 30% of asymptomatic children demonstrate serologic evidence of prior *Toxocara* infestations.

Ocular: Leukocoria; uveitis; cataract; marked vitreous reaction with large floaters; choroiditis; large, cystlike white masses extending into vitreous; optic neuritis; papillitis; strabismus; hemorrhagic, exudative, or granulomatous retinitis; retinal detachment; endophthalmitis; larvae present in the cornea.

Clinical: Hepatosplenomegaly; pulmonary infiltration; fever; cough; lack of appetite.

Biglan AW, et al. Serum and vitreous Toxocara antibody in nematode endophthalmitis. *Am J Ophthalmol* 1979; 88: 898,901.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Maguire AM, et al. Recovery of intraocular Toxocara Canis by pars plana vitrectomy. *Ophthalmology* 1990; 97: 675-680.

Raistrick ER, Hart JCD. Ocular toxocariasis in adults. *Br J Ophthalmol* 1976; 60:365-370.

Wilder HC. Nematode endophthalmitis. *Trans Am Acad Ophthalmol Otolaryngol* 1950; 55:99.

Wilkinson C. Ocular toxocariasis. In: Ryan SJ, ed. *Retina, vol. II*, 2nd ed. St. Louis: Mosby, 1994:1545-1552.

Neonatal Hemolytic Disease of Hyperbilirubinemia (Kernicterus; Bilirubin Encephalopathy) 874

General: Condition with severe neural symptoms associated with high levels of bilirubin in the blood.

Ocular: Disturbance of supranuclear vertical gaze; mild ocular motility defects; involuntary levator palpebrae inhibition.

Clinical: Athetosis; hearing loss; possible central nervous system (CNS) damage, characterized by deep yellow staining of the basal nuclei, globus pallidus, putamen and caudate nucleus, cerebellar and bulbar nuclei, and gray substance of the cerebrum.

Aramideh M, et al. Abnormal eye movements in blepharospasm and involuntary levator palpebrae inhibition. Clinical and pathophysiological considerations. *Brain* 1994; 117[Part 6]:1457-1474.

Hoyt CS, et al. The supranuclear disturbances of gaze in kernicterus. *Ann Ophthalmol* 1978; 10:1487-l492.

Hyman CB, et al. CNS abnormalities after neonatal hemolytic disease of hyperbilirubinemia. *Am J Dis Child* 1969; 117:395.

Nephrotic Syndrome (Lipoid Nephrosis; Idiopathic Nephrotic Syndrome; Epstein Syndrome) 875

General: Unknown etiology, although some connection seems to exist between use of various drugs (penicillamine, heavy metals, trimethadione) and certain disease entities, such as intercapillary glomerulosclerosis, renal vein thrombosis, renal amyloidosis, and congestive heart failure; age at onset usually 1 to 6 years.

Ocular: Diffuse periorbital swelling; lid edema (mainly upper lids) more pronounced in the morning; uveitis; retinal edema around optic disk.

Clinical: Impaired renal function (depending on severity of renal involvement); anasarca (generalized and changing with time).

Drummond KN, et al. The nephrotic syndrome of childhood: immunologic, *Clinical*, and pathologic correlations. *J Clin Invest* 1966; 45:620.

Epstein AA. Concerning the causation of edema in chronic parenchymatous nephritis; methods for its alleviation. *Am J Med* Sci 1917; 154:638.

Forman AR, et al. Reversibility of corticosteroid-associated cataracts in children with the nephrotic syndrome. *Am J Ophthalmol* 1977; 84:75.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Iwasaki M, et al. Uveitis in nephrotic syndrome. *Metab Pediatr Syst Ophthalmol* 1993; 16:46-48.

Neurilemoma (Schwannoma; Neurinoma) 876

General: Slow-growing encapsulated neoplasm from the Schwann cells of nerves; seen most frequently with patients with von Recklinghausen disease.

Ocular: Ptosis; exophthalmos; visual loss; pupillary dilation; lacrimal sac mass.

Clinical: Facial numbness; retroorbital headaches; intermittent pain radiating from the distribution of the appropriate sensory nerve branch.

Allman MI, et al. Orbital neurilemoma. *Ann Ophthalmol* 1977; 9:1409-1413.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Katsumata Y, et al. Neurinoma of the oculomotor nerve: CT and MRI features. *J Comput Assist Tomogr* 1990; 14:658.

Sutula FC. Tumors of the lacrimal gland and sac. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. III.* Philadelphia: WB Saunders, 1994.

Trese MT, et al. Orbital embryonal rhabdomyosarcoma and intracranial schwannoma. *Arch Ophthalmol* 1979; 97: 1929-1930.

Neuroblastoma 877

General: Highly malignant solid tumor arising from undifferentiated sympathetic neuroblasts of the adrenal medulla, sympathetic ganglia, ectopic adrenal, and theoretically the ciliary ganglion; autosomal dominant.

Ocular: Ptosis; exophthalmos; proptosis; optic atrophy; optic neuritis; papilledema; metastatic tumor of orbit; retinal hemorrhage; convergent strabismus; paralysis of sixth or seventh nerve; nonreactive pupil; primary differentiated neuroblastoma of the orbit also has been reported; tonic pupils; microphthalmia; choroidal metastases (rare); iris metastases (rare).

Clinical: Skeletal metastasis to the cranium.

Blake J, Fitzpatrick C. Eye signs in neuroblastoma. *Trans Ophthalmol Soc UK* 1972; 92:825-833.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Haupt R, et al. Neuroblastoma IV-S in a patient with bilateral microphthalmia. *Acta Paediatr* 1993; 82:1085-1086.

Knudsen AG Jr, Meadows AT. Developmental genetics of neuroblastoma. *J Natl Cancer Inst* 1976; 57:675-682.

Volpe NJ, Albert DM. Metastases to the uvea. In: Albert D, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3267.

West, Repka MX. Tonic pupils associated with neuroblastoma. *J Pediatr Ophthalmol Strabismus* 1992; 29:382-383.

Neurocutaneous Syndrome 878

General: Triad of linear nevus sebaceous; seizures; mental retardation.

Ocular: Colobomas of irides and choroid; nystagmus; keratoconus; corneal vascularization; optic glioma; epibulbar choristomas; connective tissue nevi of the eyelids.

Clinical: Multiple nevi; seizures; mental retardation; failure to thrive; hydrocephalus; deformities of skull; lipoma of the cranium; alopecia of the scalp.

Kodsi SR, et al. Ocular and systemic manifestations of encephalocraniocutaneous lipomatosis. *Am J Ophthalmol* 1994; 118:77-82.

Kucukoduk S, et al. A new neurocutaneous syndrome: nevus sebaceus syndrome. *Cutis* 1993; 51:437-441.

Mardon PM, Venters HD. A new neurocutaneous syndrome. *Am J Dis Child* 1966; 67:79-81.

Sato K, et al. Linear sebaceous nevus syndrome (sebaceous nevus of Jadassohn) associated with abnormal neuronal migration and optic glioma: case report. *Neurosurgery* 1994; 35:318-320.

Neurodermatitis (Lichen Simplex Chronicus) 879

General: Skin altered due to chronic rubbing or scratching.

Ocular: Keratoconjunctivitis; lid edema; lid pigmentation; lid lichenification; atopic cataracts; keratoconus.

Clinical: Pruritus; dermatitis; seborrheic dermatitis; contact dermatitis; lichenification of skin; skin hyperpigmentation.

Arndt KA. *Manual of dermatologic therapeutics, with essentials of diagnosis*, 2nd ed. Boston: Little, Brown and Company, 1978:55-57.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Neuronal Ceroid Lipofuscinosis (Neuronal Intranuclear Inclusion Disease; NIID) 880

General: Probably autosomal recessive.

Ocular: Nystagmus; sluggish pupil reaction; restricted ocular movements; optic disk pallor; loss of nerve fiber around macula; loss of pigment; widespread loss of photoreceptor function with abnormal electroretinogram.

Clinical: Slurred speech; extrapyramidal and lower motor neuron abnormalities; atrophy of skeletal muscles; bronchopneumonia; cerebral deterioration.

Haltia M, et al. Neuronal intranuclear inclusion disease. *Acta Ophthalmol (Copenh)* 1986; 64:637-643.

Michand J, Gilbert JJ. Multiple systems atrophy with neuronal intranuclear hyaline inclusions: report of a case with light and electron microscopic studies. *Acta Ophthalmol (Copenh)* 1981; 54: 113-118.

Patel H, et al. Multiple system atrophy with neuronal intranuclear hyaline inclusions. *J Neurol Sci* 1985; 67:57-65.

Weleber RG. Retinitis pigmentosa and allied disorders. In: Ryan SJ. ed. *Retina. vol.* 1. St. Louis: CV Mosby, 1989.

Neu Syndrome 881

General: Microcephaly-growth retardation-flexion deformities; both sexes affected; autosomal recessive; early death.

Ocular: Hypertelorism; absent eyelids.

Clinical: Flexion deformities; overlapping fingers; rocker-bottom feet; protruding heels; toe syndactyly; microcephaly; short neck; tiny nose; brain atrophy.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Neu RL, et al. A lethal syndrome of microcephaly with multiple congenital anomalies in three siblings. *Pediatrics* 1971; 47:611-612.

Newcastle Disease (Fowlpox) 882

General: Acquired directly by people handling chickens (see Parinaud Oculoglandular Syndrome); self-limiting conjunctivitis caused by a paramyxovirus.

Ocular: Acute follicular conjunctivitis, unilateral; keratitic precipitates; lid edema; decreased accommodation and visual acuity.

Clinical: Fatigue; fever; headache; pulmonary complications; preauricular lymphadenopathy.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gordon S. Viral keratitis and conjunctivitis. Adenovirus and other nonherpetic viral diseases. In: Smolin G, Thoft RA, eds. *The Cornea.* Boston: Little, Brown and Company, 1994.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Niacin Overdose (Nicotinic Acid Overdose) 883

General: B vitamin used in large doses to lower serum cholesterol and triglyceride levels or as a vasodilator.

Ocular: Cystoid maculopathy without leakage on fluorescein angiography, usually resolves when patient stops taking niacin.

Clinical: Facial flushing; gastric irritation; liver function impairment.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. Clinical *Ophthalmology* Philadelphia: JB Lippincott, 1987.

Menna VJ. Index of suspicion. Case 2. Diagnosis: niacin overdose. *Pediatr Rev* 1993; 14:433-435.

Nicolau Syndrome (Nicolau-Hoigne Syndrome) 884

General: First described as a nonallergic reaction following injection of bismuth; assumed to be caused by emboli of medication inadvertently introduced into an artery.

Ocular: Visual loss, depending on degree of involvement of retinal arteries.

Clinical: Tachycardia; acoustic sensations; somnolence; motoric irritation; sudden pain in extremities and abdomen; pallor, cyanosis, and edema of extremities; shock; paresis-paralysis; arterial hypotension.

Geeraets WJ *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hoigne R. Akute Nebenreaktionen auf Pencillinpraparate. *Acta Med Scand* 1962; 171:201.

Nicolau S. Dermite Livedoide et Gangreneuse de la Fesse, Consecutive aux Injections Intramusculaires, dans la Syphilis. A Propos d'un Cas d'Embolie Arterielle Bismuthique. *Ann Mal Vener (Paris)* 1925; 20:321.

Nieden Syndrome (Telangiectasia-Cataract Syndrome) 885

General: Etiology unknown; familial occurrence; onset from birth.

Ocular: Sparse eyebrows; glaucoma; dyscoria; defects of iris mesenchyme; bilateral cataract (cortical or mature).

Clinical: Telangiectasia of face and upper extremities; pigmentary changes of the neck; thick, atrophic skin; heart enlargement; congenital valvular defect.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Nieden A. Cataractbildung bei Teleangiectatischer Ausdehnung der Capillaren der Ganzen Gesichtshaut. *Zentbl Parkt Augenheilkd* 1887; 11:353.

Petersen HP. Telangiectasis and cataract. *Acta Ophthalmol* 1954; 32:565.

Nielsen Syndrome (Exhaustive Psychosis Syndrome; Neuromuscular Exhaustion Syndrome) 886

General: Chronic infections; postoperative phases frequently are associated with extreme stress and fatigue; similar manifestations often have been reported during and after prolonged combat; develops subacutely after severe overexertion during a period of euphoria.

Ocular: Paralysis of extraocular muscles; diplopia.

Clinical: Physical exhaustion; extreme weakness; delirium; fascicular twitching; poor attention; restlessness; lack of concentration; irritability; depression; anxiety; pain in muscles; weight loss; muscle flaccidity; absence of deep reflexes.

Nielsen JM. Subacute generalized neuromuscular exhaustion syndrome. *Bull Los Angeles Neurol Soc* 1940; 5:128, and *Calif Med* 1947; 66:338.

Strassman HD, et al. A prisoner of war syndrome: apathy as a reaction of severe stress. *Am J Psychiatry* 1956; 112:998.

Niemann-Pick Syndrome (Essential Lipoid Histiocytosis; Sea-Blue Histiocytosis; Sphingomyelinase Deficiency) 887

General: Phospholipidosis with degeneration of ganglion cells of CNS and lipid storage involving entire reticuloendothelial system and parenchymatous tissue; autosomal recessive; occurs during first few months of life; similar to Tay-Sachs syndrome; predisposition for Jews (see Tay-Sachs Syndrome); lysosomal storage disease resulting from diminished activity or deficiency of sphingomyelinase.

Ocular: Vision may be reduced but usually not complete blindness; cherry-red spot of macula, similar to that of Tay-Sachs syndrome; progressive optic atrophy; supranuclear ophthalmoplegia; periorbital fullness; macular halo syndrome; abnormal visual evoked potentials.

Clinical: Mental retardation; extensive hepatosplenomegaly; epileptic seizures; progressive physical deterioration; deafness; skin pigmentation; abdominal enlargement.

Breen L, et al. Juvenile Niemann Pick disease with vertical supranuclear ophthalmoplegia: two case reports and review of the literature. *Arch Neurol* 1981; 38:388-390.

Filling-Katz MR, et al. Ophthalmologic manifestations of type B Niemann-Pick diseases. *Metab Pediatr Syst Ophthalmol* 1992; 15:16-20.

Libert J, et al. Ocular findings in Niemann-Pick disease. *Am J Ophthalmol* 1975; 80:991-1002.

Merin S, et al. Conjunctival ultrastructure in Niemann-Pick disease type C. *Am J Ophthalmol* 1980; 90:708-714.

Sperl W, et al. A family with visceral course of Niemann-Pick disease, macular halo syndrome and low sphingomyelin degradation rate. *J Inherit Metab Dis* 1994; 17:93-103.

Walton DS, et al. Ocular manifestations of group A Niemann-Pick disease. *Am J Ophthalmol* 1978; 85:174-180.

9p- Syndrome 888

General: Congenital mental retardation syndrome due to a 9p deletion.

Ocular: Mongoloid (down-slanting) eyes; usually glaucoma.

Clinical: Mental retardation; sociable personality; trigonocephaly; wide, flat nasal bridge; anteverted nostrils; long upper lip; short neck; long digits; predominance of finger whorls; choanal atresia; usually seizures.

Alfi OS, et al. The 9p- syndrome. *Ann Genet* 1976; 19:11-16.

Chaves-Carballo E, et al. Neurologic aspects of the 9p- syndrome. *Pediatr Neurol* 1985; 1:57-59.

Shashi V; Golden WL, Fryburg JS. Choanal atresia in a patient with the deletion (9p) syndrome. *Am J Med Genet* 1994; 49:88-90.

Nocardiosis 889

General: Aerobic Actinomycetaceae that may cause a chronic suppurative process; aerobic Gram-positive filamentous bacteria with branching pattern which resemble fungi.

Ocular: Conjunctivitis; keratitis; corneal ulcer; uveitis; lid involvement; orbital cellulitis; endophthalmitis; glaucoma; external ophthalmoplegia; scleritis; canaliculitis; preseptal cellulitis.

Clinical: Granuloma; draining sinuses; brain abscess; meningitis.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Kattan HM. Nocardia scleritis. *Am J Ophthalmol* 1990; 110:446-447.

Lass JH, et al. Exogenous Nocardia asteroides endophthalmitis associated malignant glaucoma. *Ann Ophthalmol* 1981; 13:317-321.

Lissner GS, et al. Endogenous intraocular Nocardia asteroides in Hodgkin's disease. *Am J Ophthalmol* 1978; 36: 388-394.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol.* 5. *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995:3081.

Sher NA, et al. Bilateral intraocular Nocardia asteroides infection. *Arch Ophthalmol* 1977; 95:1415-1418.

Nonne-Milroy-Meige Disease (Chronic Hereditary Lymphedema; Milroy Disease; Meige Disease; Meige-Milroy Syndrome; Nonne-Milroy Syndrome; Chronic Hereditary Edema; Chronic Hereditary Trophedema; Chronic Trophedema; Elephantiasis Congenita Hereditaria; Familial Hereditary Edema; Hereditary Edema; Idiopathic Hereditary Lymphedema; Pseudoedematous Hypodermal Hypertrophy; Pseudoelephantiasis Neuroarthritica; Oromandibular Dystonia; Blepharospasm-Oromandibular Dystonia; Congenital Trophedema; Tropholymphedema; Trophoneurosis; Elephantiasis Arabum Congenita) 890

General: Autosomal dominant; prevalent in females; two types: praecox at birth to 35 years and tarda after age 35 years.

Ocular: Lid and conjunctival edema; blepharoptosis; distichiasis; strabismus; buphthalmos; ectropion.

Clinical: Lymphedema; mandibulofacial dysostosis; unilateral or bilateral edema of ankle ascending to the knee and eventually above; rough, pigmented skin over swollen parts.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Meige H. Le Trophoedeme Chronique Hereditaire. *Nouv Salpetrieie* 1899; 12:453-480.

Milory WF. An undescribed variety of hereditary edema. *NY Med J* 1892; 56:505-508.

Nonne M. Vier Falle von Elephantiasis Congenita Hereditaria. *Arch Pathol Anat* 1891; 125:184-196.

Tabbara KF, Baghdassarian SA. Chronic hereditary lymphedema of the legs with congenital conjunctival lymphedema. *Am J Ophthalmol* 1972; 73:531-532.

Noonan Syndrome (Male Turner Syndrome) 891

General: Similar to Turner syndrome, but with normal chromosomal analysis; X-linked dominant inheritance; X-linked dominant phenotype.

Ocular: Hypertelorism; exophthalmos; ptosis (unilateral or bilateral); antimongoloid-slanting palpebral fissure; myopia; keratoconus; optic disk coloboma.

Clinical: Valvular pulmonary stenosis; short stature; webbed neck; low hairline in the back; cubitus valgus; deformed chest wall; micrognathia; low-set ears; mild mental retardation.

Ahmed ML, et al. Noonan's syndrome abnormalities of the growth hormone/IGF-I axis and the response to treatment with human biosynthetic growth hormone. *Acta Paediatr Scand* 1991; 80:446-450.

Daoud MS, Dahl PR, Su WP. Noonan syndrome. *Semin Dermatol* 1995; 14:140-144.

Goens NM, Campbell D, Wiggins JW. Spontaneous chylothorax in Noonan syndrome. Treatment with prednisone. *Am J Dis Child* 1992; 146:1453-1456.

Lee NB, Kelly L, Sharland M. Ocular manifestations of Noonan syndrome. *Eye* 1992; 6[Part 3]:328-334.

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Miller M, Motulsky AC. Noonan syndrome in an adult family presenting with chronic lymphedema. *Am J Med* 1978; 65:379-383.

Noonan JA. Hypertelorism with Turner phenotype: a new syndrome with associated congenital heart disease. *Am J Dis Child* 1968; 116:373.

Schwartz DE. Noonan's syndrome associated with ocular abnormalities. *Am J Ophthalmol* 1972; 73:955.

Sharland M, et al. A *Clinical* study of Noonan syndrome. *Arch Dis Child* 1992; 67:178-183.

Wiedemann HR. Otto Ullrich and his syndromes. *Am J Med Genet* 1991; 41:128-133.

White SW Lymphedema in Noonan's syndrome. *Int J Dermatol* 1984; 23:656-657.

Nothnagel Syndrome (Ophthalmoplegia-Cerebellar Ataxia Syndrome) 892

General: Lesion of superior cerebellar peduncle, red nucleus, and emerging oculomotor fibers, such as pineal tumor, or tumor or vascular disturbance in corpora quadrigemina or vermis cerebelli (see Bruns Syndrome).

Ocular: Oculomotor paresis; gaze paralysis most frequently upward, combined with some degree of internal or external ophthalmoplegia.

Clinical: Cerebellar ataxia; poor upper extremity movements; neoplasia; infarction; midbrain lesion.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Nothnagel CWH. *Topische Diagnostik der Gehirnkrankheiten.* Berlin: A. Hirschwald, 1879:220.

Nystagmus Blockage Syndrome (NBS) 893

General: Congenital (see Ethan Syndrome, Primary; Ethan Syndrome Secondary; Nystagmus Compensation Syndrome); convergence is used to diminish nystagmus.

Ocular: Esotropia; nystagmus; amblyopia; most patients with this syndrome prefer to fixate with one eye, but others show alternating fixation.

Clinical: Abnormal head position.

Isenberg SJ, Yee RD. The Ethan syndrome. *Ann Ophthalmol* 1986; 18:358-361.

von Noorden GK, Avilla CW. Nystagmus blockage syndrome: revisited. *Strabismus II.* New York: Grune & Stratton, 1984:75-82.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol.* 2, 4th ed. Baltimore: Williams & Wilkins, 1985:897.

Nystagmus Compensation Syndrome (NCS) 894

General: Congenital (see Ethan Syndrome, Primary; Ethan Syndrome, Secondary; Nystagmus Blockage Syndrome).

Ocular: Esotropia; amblyopia; onset may be preceded by manifest nystagmus; abnormal head posture toward the adducted fixing eye; nystagmus reduced or absent, with the fixing eye adducted.

Clinical: Abnormal head position.

Frank JW. Diagnostic signs in the nystagmus compensation syndrome. *J Pediatr Ophthalmol Strabismus* 1979; 16: 317-320.

von Noorden GK. The nystagmus compensation (blockage) syndrome. *Am J Ophthalmol* 1976; 82:287.

Nystagmus, Congenital (Congenital Idiopathic Nystagmus) 895

General: Autosomal dominant; pattern of inheritance in congenital nystagmus, whether of the "motor" or "sensory" type, may be autosomal dominant, recessive, or sex-linked.

Ocular: Vertical and horizontal nystagmus; this nystagmus occasionally is vertical or torsional; in addition, periodic alternating, downbeat, and see-saw nystagmus may be present at birth; normal electroretinogram.

Clinical: None.

Allen M. Three pedigrees of eye defects: primary hereditary nystagmus. Case study with genealogy. *J Hered* 1942; 33:454-456.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 2, 4th ed. Baltimore: Williams & Wilkins, 1985:894.

Nystagmus, Hereditary Vertical 896

General: Autosomal dominant.

Ocular: Motor-type vertical and horizontal nystagmus; hyperactive vestibuloocular response; strabismus.

Clinical: Mild ataxia; poor line walking.

Marmor MF. Hereditary vertical nystagmus. *Arch Ophthalmol* 1973; 90: 101-111.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Nystagmus, Primary Hereditary (Congenital Nystagmus) 897

General: Autosomal recessive, sex-linked or irregular dominant; may be associated with albinism.

Ocular: Horizontal nystagmus; myopia.

Clinical: Head spasms; carpopedal spasms (Trousseau sign); Chvostek sign.

Allen M. Primary hereditary nystagmus: case study with genealogy. *J Hered* 1942; 33:454-456.

Guttman DH, et al. Congenital nystagmus in a (40,XX/45,X) mosaic woman from a family with X-linked congenital nystagmus. *Am J Med Genet* 1991; 39:167-169.

Lavin PJM. Congenital nystagmus. In: Bradley WG, ed. *Neurology in Clinical practice*, 2nd ed. Boston: Butterworth-Heinemann, 1995:200.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Stromberg A, Pavan-Langston D. Extraocular muscles, strabismus, and nystagmus. In: Pavan-Langston D, ed. *Nystagmus: manual of ocular diagnosis and therapy*, 4th ed. Boston: Little, Brown and Company, 1995:333-336.

Nystagmus, Voluntary 898

General: Autosomal dominant; usually is purely horizontal, but it may be vertical or-torsional.

Ocular: Voluntary rapid to-and-fro synchronous movements of eyes.

Clinical: Simultaneous head tremor has been associated with this condition.

Keyes MJ. Voluntary nystagmus in two generations. *Arch Neurol* 1973; 29:63-64.

Lee J, Gresty M. A case of "voluntary nystagmus" and head tremor. *J Neurol Neurosurg Psychiatry* 1993; 56: 1321-1322.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 2, 4th ed. Baltimore: Williams & Wilkins, 1985:4554.

O

Obesity-Cerebral-Ocular-Skeletal Anomalies Syndrome 899

General: Rare, autosomal recessive disease; similar to Prader-Willi and Laurence-Moon-Bardet-Biedl syndromes (see Prader-Willi Syndrome; Laurence-Moon-Bardet-Biedl Syndrome).

Ocular: Microphthalmia; antimongoloid slant of lid fissure; asymmetrical size of fissure; strabismus; myopia; iris and chorioretinal colobomata; mottled retina; prominent choroidal vessels.

Clinical: Obesity (mid-childhood onset); hypotonia; mental retardation; craniofacial anomalies with microcephaly; tapering extremities; hyperextensibility at elbows and proximal interphalangeal joints; cubitus valgus; genu valgum; simian creases; syndactyly.

Cohen MM, et al. A new syndrome with hypotonia, obesity, mental deficiency, and facial, oral, ocular, and limb anomalies. *J Pediatr* 1973; 83:280.

Hall BD, Smith DW. Prader-Willi syndrome. *J Pediatr* 1972; 81:286.

Ocular Dominance 900

General: Autosomal dominant; 97% of people with normal vision have a sighting-dominant eye; experimental data suggest a relationship between eye dominance and head tilt.

Ocular: Sighting-dominant eye.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Previc FH. The relationship between eye dominance and head tilt in humans. *Neuropsychologia* 1994; 32: 1297-1303.

Zoccolotti P. Inheritance of ocular dominance. *Behav Genet* 1978; 8:377-379.

Ocular Myopathy with Curare Sensitivity 901

General: Autosomal recessive.

Ocular: Static ophthalmoparesis.

Clinical: Limb weakness; sensitivity to tubocurarine.

Matthew NT, et al. Familial ocular myopathy with curare sensitivity. *Arch Neurol* 1970; 22:68-74.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Ocular Toxoplasmosis (Toxoplasmic Retinochoroiditis; Toxoplasmosis) 902

General: Parasite infestation caused by *Toxoplasma gondii;* cell-mediated immunity is believed to be the major defense mechanism against *Toxoplasma* infection; ocular toxoplasmosis occurs in approximately 1% of patients with acquired immunodeficiency syndrome (AIDS); AIDS-related toxoplasma retinochoroiditis may have several atypical clinical manifestations.

Ocular: Keratitis; uveitis; optic atrophy; papillitis; anisocoria; persistent pupillary membrane; focal retinochoroiditis; scleritis; cataract; microphthalmos; myopia; nystagmus; esotropia.

Clinical: Cysts are seen in many organs, including brain and muscle; hydrocephalus; intracerebral calcification; various CNS complaints.

Binkhorst CD. Toxoplasmic infection and toxoplasmosis of the eye. *Int Ophthalmol Clin* 1970; 10:621-638.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Friedman AH. The retinal lesions of the acquired immune deficiency syndrome. *Trans Am Ophthalmol Soc 1984;* 82:447.

O'Connor GR. Manifestations and management of ocular toxoplasmosis. *Bull NY Acad Med* 1974; 50: 192-210.

Williams DM, et al. Genetic control of immune resistance to Toxoplasma gondii. *Infect Immunol* 1978; 19:416.

Ocular Vaccinia 903

General: Occurs from virus used for smallpox vaccination; transferred from vaccination site to eye.

Ocular: Chemosis; nonfollicular, catarrhal, or purulent conjunctivitis; punctate epithelial keratitis; corneal vascularization; lid edema; vesicles of lid; uveitis; choroiditis; extraocular muscle palsies; optic neuritis; orbital cellulitis; ocular vaccinia may mimic signs of herpes simplex virus, varicella-zoster virus, and acanthamoeba keratitis.

Clinical: Dermatitis; encephalitis.

Hyndiuk RA, et al. Treatment of vaccinial keratitis with vidarabine. *Arch Ophthalmol* 1976; 94:1363-1364.

Lee SF, et al. Vaccinia keratouveitis manifesting as a masquerade syndrome. *Am J Ophthalmol* 1994; 117:480-487.

Ruben FI, Lane JM. Ocular vaccinia: an epidemiologic analysis of 348 cases. *Arch Ophthalmol* 1970; 84:45-48.

Oculocerebellar Tegmental Syndrome 904

General: Vascular lesion of mesencephalon with softening in peduncular area.

Ocular: Paralysis of associated ocular movements (internuclear anterior ophthalmoplegia).

Clinical: Sudden onset of hemiplegia with rapid recovery; bilateral cerebellar syndrome.

Fournier A, et al. Oculocerebellomyoclonic syndrome and Neuroblastoma. *J Sci Med Lille* 1972; 90:189.

Rodriquez B, et al. A new type of peduncular syndrome: internuclear ophthalmoplegia and bilateral cerebellar syndrome from a tegmental lesion. *Arch Urug Med* 1945; 10:353, *Am J Ophthalmol* 1946; 29:511(abst).

Oculocerebral Syndrome with Hypopigmentation (Amish Oculocerebral Syndrome; Cross Syndrome) 905

General: Autosomal recessive.

Ocular: Spastic ectropion; microphthalmos; enophthalmos; microcornea; corneal opacification; corneal vascularization; palpebral conjunctival injection; narrow lid fissures; aniridia; nystagmus; bilateral optic atrophy.

Clinical: Spastic diplegia; cutaneous hypopigmentation; mental retardation; hypogonadism; growth retardation; developmental defects of the CNS, such as cystic malformation of the posterior fossa of the Dandy-Walker type.

Cross HE, et al. A new oculocerebral syndrome with hypopigmentation. *J Pediatr* 1967; 70:398-406.

De Jong G, Fryns JP. Oculocerebral syndrome with hypopigmentation (Cross syndrome): the mixed pattern of hair pigmentation as an important diagnostic sign. *Genet Couns* 1991; 2:151-155.

Lerone M, et al. Oculocerebral syndrome with hypopigmentation (Cross syndrome): report of a new case. *Clin Genet* 1992; 41:87-89.

Pinsky L, et al. Microphthalmos, corneal opacity, mental retardation and spastic cerebral palsy: an oculocerebral syndrome. *J Pediatr* 1965; 67:387.

Oculocerebrocutaneous Syndrome (Delleman Syndrome) 906

General: Congenital; possibly autosomal recessive or a result of an environmental problem; Dutch descent appears to be a predisposing factor; in most cases, involvement is unilateral, with left to right 2: 1.

Ocular: Orbital cyst; absence of orbital structures; microphthalmos; anophthalmia.

Clinical: Cerebral malformations; focal dermal hypoplasia; aplasia; epilepsy; developmental delay; cranial skin appendages; cutaneous punched-out lesions.

Delleman W, et al. Orbital cyst in addition to congenital cerebral and focal dermal malformations. *Clin Genet* 1984; 25:470.

Hoo JJ. Oculocerebrocutaneous (Delleman) syndrome: a pleiotropic disorder affecting ectodermal tissues with unilateral predominance. *Am J Med Genet* 1991; 40:290-293.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Wilson RD, et al. Oculocerebrocutaneous syndrome. *Am J Ophthalmol* 1985; 99:142-148.

Oculodental Syndrome (Peters Syndrome; Rutherford Syndrome) 907

General: Similar to Rieger syndrome and Meyer-Schwickerath-Weyer syndrome; Peters syndrome inherited as autosomal recessive with defect of corneogenetic mesoderm characterized by incomplete separation of lens vesicle, causing central opacities of cornea, shallow anterior chamber, synechiae, and remnants of pupillary membrane; anterior pole cataract; Rutherford syndrome inherited as autosomal dominant; exhibits iris and dental anomalies and mental retardation.

Ocular: High myopia; corneoscleral staphyloma; aniridia; macrocornea; opacities of the corneal margin; ectopia lentis with deposits of pigment; macular pigmentation; large excavation of optic nerve with atrophy.

Clinical: Oligodontia; microdontia; hypoplasia of enamel; abnormal tooth positions; hypertrophy of gums; failure of tooth eruption.

Houston IB. Rutherford's syndrome. A familial oculo-dental disorder and electrophysiological study. *Acta Paediatr Scand* 1966; 55:233-238.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Peters A. Uber Angeborene Defektbildung der Descemetschen Mem bran. *Klin Monatsbl Augenheilkd* 1906; 44:27.

Reisner SH, et al. Oculodentodigital dysplasia. *Am J Dis Child* 1969; 118:600.

Rutherford ME. Three generations of inherited dental defects. *Br Med J* 1931; 11:9.

Oculogastrointestinal Muscular Dystrophy 908

General: Autosomal recessive; visceral myopathy with external ophthalmoplegia; intestinal pseudo obstruction with external ophthalmoplegia.

Ocular: Ptosis; ophthalmoplegia.

Clinical: Chronic diarrhea; abdominal distention; diffuse abdominal pain; impaired motility of the lower esophagus; demyelinating and axonal neuropathy with focal spongiform degeneration of the posterior columns; proximal muscle weakness and atrophy; primary myopathy of the smooth muscles of the stomach and intestine; dilated duodenum and jejunum; recurrent intestinal obstruction in childhood or adolescence.

Alstead EM, et al. Familial autonomic visceral myopathy with degeneration of muscularis mucosae. *J Clin Pathol* 1988; 41:424-429.

Ionasescu V, et al. Late-onset oculogastrointestinal muscular dystrophy. *Am J Med Genet* 1983; 15:103-112.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Oculo-Orogenital Syndrome (Riboflavin Deficiency Syndrome; Gopalan II Syndrome; Jolliffe Syndrome) 909

General: Vitamin B deficiency and possible vitamin A deficiency; prognosis good with diet therapy.

Ocular: Conjunctivitis, varying from mild to severe; keratitis; optic atrophy; corneal vascularization.

Clinical: Stomatitis; glossitis; scrotal dermatitis with pruritus, erythema, erythema of pharynx and soft palate; small sensitive ulcers of buccal membranes; diarrhea; fatigue; muscular weakness; painful feet with erythema, exfoliation, and ulceration; burning; itching; mental depression; dizziness; oral mucosa becomes pale and macerated with fissuring of skin.

Bajart AM. Lid inflammations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. 1.* Philadelphia: WB Saunders, 1994:114.

Bloom SM, et al. Nutritional amblyopia in American prisoners. *Am J Ophthalmol* 1946; 29: 1248.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Jacobs EC. Oculo-oro-genital syndrome: a deficiency disease. *Ann Intern Med* 1951; 35:1049.

McLaren DS. Cutaneous changes in nutritional disorders. In: Fitzpatrick TB, Eisen AZ, Wolff K, et al., eds. *Dermatology in general medicine*, 3rd ed. New York: McGraw-Hill, 1987:1601-1613.

Oculo-Osteocutaneous Syndrome 910

General: Autosomal recessive.

Ocular: Strabismus; myopia; distichiasis; nystagmus; lenticular opacities.

Clinical: Short stature; brachydactyly; hypoplastic maxilla; scanty hair; hypopigmentation; mental retardation.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Tumaala P, Haapanen E. Three siblings with similar anomalies in the eyes, bones, and skin. *Acta Ophthalmol* 1968; 46:365-371.

Oculo-Oto-Oro-Reno-Erythropoietic Disease 911

General: Etiology unknown; slowly progressive course; ocular lesions similar to those of tuberous sclerosis, but no other signs of that disorder.

Ocular: Exotropia; progressive visual loss to complete blindness; secondary glaucoma with iris bombe; iridocyclitis; chronic uveitis; vitreous opacities; cataract; retinal hemorrhages and exudates.

Clinical: Anemia; atypical development of periodontia and teeth with caries; high-arched palate; abdominal colic; genu valgum and pectus excavatum; dizziness.

Jacobsen CD, Brodwall EK. A *Clinical* syndrome with inborn defect in erythropoiesis, dysplastic kidneys, eye lesions, malformation of the teeth and impaired hearing: a new syndrome in a 28 year old woman. *Acta Med Scand* 1974; 195:231.

McCance RA, et al. The cerebro-ocular-renal dystrophies: a new variant. *Arch Dis Child* 1960; 35:24.

Oculopalatocerebral Dwarfism (OPC Dwarfism) 912

General: Autosomal recessive; persistent hyperplastic primary vitreous.

Ocular: Persistent hypertrophic primary vitreous; microphthalmos; leukocoria; retrolental fibrovascular membrane.

Clinical: Microcephaly; mental retardation; spasticity; cleft palate; short stature.

Frydman M, et al. Oculo-palato-cerebral dwarfism: a new syndrome. *Clin Genet* 1985; 27:414-419.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Oculopalatoskeletal Syndrome 913

General: Autosomal recessive; developmental defect of eye and occipital bone; skeletal abnormalities.

Ocular: Triad of blepharophimosis, blepharoptosis, and epicanthus inversus; corneal stromal opacities; limitation of upward gaze.

Clinical: Cleft lip/palate; spina bifida occulta; cranial asymmetry; radioulnar synostosis; short fifth finger; abnormality of occipital bone.

Cunniff C, Jones KL. Craniosynostosis and lid anomalies: report of a girl with Michels syndrome. *Am J Med Genet* 1990; 37:28-30.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Michels VV, et al. A clefting syndrome and ocular anterior chamber defect and lid anomalies. *J Pediatr* 1978; 93: 444-446.

Oculopharyngeal Syndrome (progressive Muscular Dystrophy with Ptosis and Dysphagia; Oculopharyngeal Muscular Dystrophy) 914

General: Etiology unknown; autosomal dominant inheritance; no CNS pathology; muscles of pharynx, hypopharynx, and proximal third of esophagus involved with myopathy; onset late in life; progressive hereditary myopathy in which the levator palpebrae and pharyngeal muscles are selectively involved; progressive usually symmetrical blepharoptosis with or without dysphagia appears in the fifth decade.

Ocular: Ptosis.

Clinical: Dysphagia; occasionally, weakness of facial muscles.

Codere F. Oculopharyngeal muscular dystrophy. *Can J Ophthalmol* 1993; 28: 1-2.

Duranceau A, Forand MD, Fauteaux JP. Surgery in oculopharyngeal muscular dystrophy. *Am J Surg* 1980; 139:33-39.

Jordan DR, Addison DJ. Surgical results and pathological findings in the oculopharyngeal dystrophy syndrome. *Can J Ophthalmol* 1993; 28:15-18.

Molgat YM, Rodrigue D. Correction of blepharoptosis in oculopharyngeal muscular dystrophy: review of 91 cases. *Can J Ophthalmol* 1993; 28:11-14.

Murphy SF, Drachman DB. The oculopharyngeal syndrome. *JAMA* 1968; 203:1003.

Taylor EW. Progressive glossopharyngeal paralysis with ptosis: contribution to a group of family diseases. *J Nerv Ment Dis* 1915; 42:129.

Oculorenocerebellar Syndrome (ORC Syndrome) 915

General: Autosomal recessive.

Ocular: Progressive tapetoretinal degeneration with loss of retinal vessels.

Clinical: Mental retardation; continuous jerky movements; spastic diplegia; glomerulopathy with most renal glomeruli completely sclerosed.

Hunter AG, et al. Absence of cerebellar granular layer, mental retardation, tapetoretinal degeneration and progressive glomerulopathy: an autosomal recessive oculo-renal-cerebellar syndrome. *Am J Med Genet* 1982; 11: 383-395.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Oguchi Disease 916

General: Autosomal recessive; usually Japanese; form of congenital hemeralopia; one form of essential, congenital night blindness.

Ocular: Fundus has white-gray coloration, especially around the optic disk and in macular region after light exposure (30 minutes to 1 hour); color sometimes is more brown; after 2 to 3 hours in the dark (Mizuo-Nakamura phenomenon), the posterior pole appears normal; the pathogenesis of Mizuo phenomenon has been postulated to be secondary to an excess of extracellular potassium in the retina as a result of a decreased potassium scavenging capacity of retinal Muller cells; central visual acuity and visual fields normal; night blindness always present.

de Jong PT, et al. Mizuo phenomenon in X-linked retinoschisis. Pathogenesis of the Mizuo phenomenon. *Arch Ophthalmol* 1991; 109:1104-1108.

Magalini S, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Pau H. *Differential diagnosis of eye diseases* New York: Thieme, 1987.

OHAHA Syndrome (Ophthalmoplegia, Hypotonia, Ataxia, Hypoacusis, Athetosis) 917

General: Ophthalmoplegia, hypotonia, ataxia, hypoacusis, athetosis (OHARA) are distinguishing symptoms; sudden onset of deafness at an age after patient has learned to speak.

Ocular: Kernicterus; strabismus; nystagmus; ocular migraine; ophthalmoplegia; vascular spasm in branches of the ophthalmic artery; intact convergence.

Clinical: Hemiplegia; athetosis; choreoathetosis; tremor; hypoxia; corticospinal tract disease; diabetes mellitus; ataxia; medulloblastoma; asynergia; dysdiadochokinesis; Holmes rebound phenomenon; acute cerebellar lesion; dysmetria; dysarthria; Fox syndrome; vascular occlusion; congenital athetosis.

Kallis AK, et al. A new syndrome of ophthalmoplegia, hypoacusis, ataxia, hypotonia, and athetosis (OMAHA). *Adv Audiol* 1985; 3:84-90.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Woody RC, et al. Ophthalmoplegic migraine in infancy. *Clin Pediatr* 1986; 25:82-84.

Okihiro Syndrome 918

General: Autosomal recessive syndrome of Duane syndrome (retraction syndrome).

Ocular: Narrowing of palpebral fissures on adduction, widening on abduction; primary global retraction.

Clinical: May be associated with craniofacial abnormalities and various associated syndromes, such as Duane syndrome, cervico-oculo-acoustic syndrome, acrorenoocular syndrome, cat's-eye syndrome; association with cardiac defects, urinary tract anomalies, Duane anomaly associated with mental retardation, thenar hypoplasia, and radial ray abnormalities.

Collins A, Baraitser M, Pembrey M. Okihiro syndrome: thenar hypoplasia and Duane anomaly in three generations, *Clin Dysmorphol* 1993;2:237-240,

McGowan KF, Pagon RA. Okihiro syndrome [Letter]. *Am J Med Genet* 1994; 51:89.

Stoll C, Alembik Y, Dott B. Association of Duane anomaly with mental retardation, cardiac and urinary tract abnormalities: a new autosomal recessive condition? *Ann Genet* 1994; 37:207-209.

Oliver-McFarlane Syndrome (Trichomegaly Syndrome) 919

General: Rare syndrome.

Ocular: Trichomegaly; pigmentary retinal degeneration.

Clinical: Prenatal onset growth failure; anterior pituitary deficiencies; peripheral neuropathy; mental retardation, sparse scalp hair, endocrinologic deficiencies, and koilonychias may be found.

Sampson JR, Tolmie JL, Cant JS. Oliver McFarlane syndrome: a 25 year follow-up. *Am J Med Genet* 1989; 34: 199-201.

Shaker AG, et al. Ovarian stimulation in an infertile patient with growth hormone-deficient Oliver-McFarlane syndrome. *Hum Reprod* 1994; 9:1997-1998.

Zaun H, et al. The long-eyelash syndrome (trichomegaly syndrome, Oliver-McFarlane). *Hautarzt* 1984; 35: 162-165.

Olivopontocerebellar Atrophy III (OPCA III; OPCA with Retinal Degeneration) 920

General: Autosomal dominant; neurologic lesion; dominant with variable penetration.

Ocular: Retinopathy variable: peripheral, macular, and circumpapillary; retinal degeneration; blindness; external ophthalmoplegia; variable electroretinogram function.

Clinical: Ataxia.

Hamilton SR, et al. Cone dysfunction in a subgroup of patients with autosomal dominant cerebellar ataxia. *Arch Ophthalmol* 1990; 108:551.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Traboulsi EI, et al. Olivopontocerebellar atrophy with retinal de generation: a *Clinical* and ocular histopathological study. *Arch Ophthalmol* 1988; 106:801.

Weiner LP, et al. Hereditary olivopontocerebellar atrophy with retinal degeneration. Report of a family through six generations. *Arch Neurol* 1967; 16:364-376.

Onchocerciasis Syndrome (River Blindness; Onchocerca Volvulus Infestation) 921

General: Nematode infestation; positive diagnosis made with microfilariae from a skin biopsy and the presence of microfilariae in anterior chamber; infection results from the bite of the blackfly genus *Simulium;* recent findings suggest an autoimmune etiology for the occurrence of chorioretinopathy.

Ocular: Punctate keratitis (fluffy opacities); sclerosing keratitis; chronic iritis; pear-shaped pupil deformation; chorioretinitis; optic atrophy; uveitis; iris atrophy; papillitis; glaucoma; retinal degeneration; perivascular sheathing; microfilariae present in the anterior chamber, cornea, and vitreous; pannus; synechiae; cataracts; secondary glaucoma.

Clinical: Subcutaneous nodules; pruritus; atrophic skin changes; pretibial pigmentation; dermatitis; lymphadenitis.

Anderson J, Fuglsang H. Further studies on the treatment of ocular onchocerciasis with diethylcarbamazine and suramin. *Br J Ophthalmol* 1978; 62:450-457.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hibberd PL, Ryan ET, Baker S. Pneumocystis carinii, parasitic infections and the eye. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V* Philadelphia: WB Saunders, 1994:307l.

Meilof JF, et al. Autoimmunity and filariasis. Autoantibodies against cytoplasmic cellular proteins in sera of patients with onchocerciasis. *J Immunol* 1993; 151:5800-5809.

Neumann E, Gunders AE. Pathogenesis of the posterior segment lesion of ocular onchocerciasis. *Am J Ophthalmol* 1973; 75:82.

Roy FH. Onchocerciasis. *Ann Ophthalmol* 1974; 6: 12.

One-and-a-Half Syndrome 922

General: Lesions in medial longitudinal fasciculus and paramedian pontine reticular formation; lateral gaze palsy on the side of the lesion and contralateral internuclear ophthalmoplegia; most common etiologies are ischemic infarction, demyelinating lesions, compressive lesion, and infections.

Ocular: Paralysis of abduction; lateral gaze palsy; nystagmus; ocular bobbing; exotropia; esotropia.

Clinical: Dysarthria; dysphagia; hemiparesis; multiple sclerosis frequently associated in young patients with this condition.

Bogousslavsky J, et al. One-and-a-half syndrome in ischemic locked in state: a clinicopathological study. *J Neurol Neurosurg Psychiatry* 1984; 47:927-935.

Hommel M, et al. Magnetic resonance imaging and the one-and-a half syndrome: a case report. *J Clin Neuro-Ophthalmol* 1987; 7:161-l64.

Wall M, Wray SH. The one and one-half syndrome: a unilateral disorder of the pontine tegmentum: a study of 20 cases and review of the literature. *Neurology* 1983; 33:971.

Ophthalmodynia Hypertonica Copulationis Syndrome 923

General: Provocation of a diagnosed and otherwise controlled angle-closure glaucoma by prolonged sexual intercourse in the prone position.

Ocular: Acute attacks of angle-closure glaucoma during copulation in prone position.

Clinical: Nausea; vomiting.

Hyams SW, et al. Elevated intraocular pressure in prone position: a new provocative test for angle-closure glaucoma. *Am J Ophthalmol* 1968; 66:661.

Markovits AS. Ophthalmodynia hypertonica copulationis. *Can J Ophthalmol* 1974; 9:484.

Ophthalmopathic Syndrome (Thyrohypophysial Syndrome) 924

General: Exophthalmos, usually bilateral; pituitary hypersecretion or enhanced tissue sensitivity to exophthalmos-producing factor that has been separated from thyroid-stimulating hormone.

Ocular: Exophthalmos; paralysis of ocular muscles.

Clinical: Thyroid disorders.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Werner SC, et al. New and simple test for hyperthyroidism employing 1-triiodothyronine and 24-hour 1131 uptake method. *Bull NY Acad Med* 1955; 31:137-145.

Ophthalmoplegia, Familial Static 925

General: Autosomal dominant; forms include internal, external, and total ophthalmoplegia.

Ocular: Ptosis; almost completely fixed eyes; nystagmoid movements; unequal pupils; pupil paralysis.

Clinical: None.

Lees F. Congenital Static Familial Ophthalmoplegia. *J Neurol Neurosurg Psychiatry* 1960; 23:44-51.

Mace JW, et al. Congenital hereditary nonprogressive external ophthalmoplegia. *J Neurol Neurosurg Psychiatry* 1960; 23:46-51.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Ophthalmoplegia, Progressive External 926

General: Autosomal recessive; progressive limitation of ocular motility with clinical sparing of pupillary function; underlying pathogenesis is secondary to a mitochondrial cytopathy; appearance of ragged red fibers in the abnormal muscles is primarily caused by mitochondrial accumulations beneath the plasma membrane and between the myofibrils.

Ocular: Oculopharyngeal muscular dystrophy; retinitis pigmentosa; progressive external ophthalmoplegia; some patients show only ptosis and ophthalmoplegia; most patients have multisystem involvement.

Clinical: Heart block; ataxia.

Drachman DA. Ophthalmoplegia plus: a classification of the disorders associated with progressive external ophthalmoplegia. *Clin Neurol* 1976; 22:203-216.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 2, 4th ed. Baltimore: Williams & Wilkins, 1985:817.

Seibel P, et al. Chronic progressive external ophthalmoplegia is associated with a novel mutation in the mitochondrial tRNA(Asn) gene. *Biochem Biophys Res Commun* 1994; 204:482-489.

Ophthalmoplegia, Progressive External, with Ragged Red Fibers 927

General: Autosomal dominant.

Ocular: Progressive ophthalmoplegia.

Clinical: Ragged red fibers in skeletal muscle from the extremities; subsarcolemmal clusters of mitochondria containing paracrystalline inclusions.

Iannoccone ST, et al. Familial progressive external ophthalmoplegia and ragged-red fibers. *Neurology* 1974; 24: 1033-1038.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Ophthalmoplegia, Progressive External, with Scrotal Tongue and Mental Deficiency 928

General: Autosomal dominant.

Ocular: Progressive external ophthalmoplegia; progressive chorioretinal sclerosis; bilateral ptosis; convergence paresis; myopia; optic atrophy; retinitis pigmentosa.

Clinical: Bilateral facial weakness; lingua scrotalis; mental retardation; cerebellar ataxia; weakness and spasticity of the limbs.

Leuic ZM, et al. Progressive nuclear ophthalmoplegia associated with mental deficiency, lingua scrotalis, and other neurologic and ophthalmologic signs in a family. *Neurology* 1975; 25:68-71.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Ophthalmoplegic Migraine Syndrome 929

General: Symptoms produced by ipsilateral herniation of hippocampal gyrus of temporal lobe through incisura tentorii; dependent upon unilateral cerebral edema due to vascular or vasomotor phenomena, intracranial aneurysm, or tumor; incidence may be greater in women with the initial attack in the first decade of life; pathogenesis is unclear, but it is likely secondary to ischemia of the ocular motor nerve.

Ocular: Severe unilateral supraorbital pain; ptosis; transitory partial or complete homolateral oculomotor paralysis; fourth or sixth nerve occasionally involved; retinal hemorrhages; papilledema (may be bilateral); moderate to severe headache with partial to complete cranial nerve III paresis including the pupil; more than one ocular nerve may be affected.

Clinical: Migraine headache, not present in all instances; dizziness; diminution in sense of smell; hypalgesia contralateral side of face; nausea/vomiting may be present; recurrent sinus arrest.

Bazak I, et al. Ophthalmoplegic migraine and recurrent sinus arrest. *J Neurol Neurosurg Psychiatry* 1991; 54:935.

Ehlers H. On pathogenesis of ophthalmoplegic migraine. *Acta Psychiatr Neurol (Scand)* 1928; 3:219.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Raskin NH. Migraine and other headaches. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1995:837-845.

Stommel EW; et al. Ophthalmoplegic migraine or Tolosa-Hunt syndrome? *Headache* 1994; 34:177.

Van Pelt W, Andermann F. On the early onset of ophthalmologic migraine. *Am J Dis Child* 1964; 107:628.

Vijayan N. Ophthalmoplegic migraine: ischemic or compressive neuropathy? *Headache* 1980; 20:300-304.

Ophthalmoplegic Neuromuscular Disorder with Abnormal Mitochondria 930

General: Autosomal recessive.

Ocular: Ptosis; external ophthalmoplegia.

Clinical: Involvement of cranial nerves and skeletal muscles; morphologic alterations of mitochondria; diffuse low-density deep cerebral white matter; weak limbs.

Okamoto T, et al. Ophthalmoplegia plus: its occurrence with periventricular diffuse low density on computed tonography scan. *Arch Neurol* 1981; 38:423-426.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Ophthalmoplegic Retinal Degeneration Syndrome (Barnard-Scholz Syndrome) 931

General: Onset at all ages (see Kearns-Sayre Syndrome).

Ocular: Unilateral or bilateral progressive weakness of muscles of eyelids, up to severe ptosis; progressive ocular myopathy up to complete ophthalmoplegia; retinitis pigmentosa.

Clinical: Facial, neck, and shoulder muscle weakness; hearing defects; heart block.

Barnard RI, Scholz RO. Ophthalmoplegia and retinal degeneration. *Am J Ophthalmol* 1944; 27:621.

Geeraets WJ *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kiloh LG, Nevin S. Progressive dystrophy of the external ocular muscles (ocular myopathy). *Brain* 1951; 774: 115-143.

Optic Atrophy, Cataract, and Neurologic Disorder 932

General: Autosomal dominant; similar to syndromes of Behr, Marinesco, Sjögren, and Friedreich, which are autosomal recessive.

Ocular: Cataract; optic atrophy.

Clinical: Neurologic disorder.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Optic Atrophy with Demyelinating Disease of CNS 933

General: Autosomal dominant; demyelinated optic nerves appear smaller than normal and are pale white or gray in color; blood vessels may seem to be less prominent than normal.

Ocular: Optic neuritis; Leber optic atrophy.

Clinical: Ataxia; leg weakness; dysarthria; hemiparesis.

Lees F. Leber's disease with symptoms resembling disseminated sclerosis. *J Neurol Neurosurg Psychiatry* 1964; 27: 415-421.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, 4th ed. Baltimore: Williams & Wilkins, 1994: 4272.

Optic Atrophy, Juvenile (Optic Atrophy, Congenital; Kjer-Type Optic Atrophy; Optic Atrophy, Kjer-Type; OAK Syndrome [Optic Atrophy, Kjer Type]) 934

General: Autosomal dominant; dominant pattern distinguishes it from Leber optic atrophy; insidious onset; onset in childhood; pathogenetic mechanism may be primary degeneration of retinal ganglion cells.

Ocular: Central scotoma; color defects; choroidal sclerosis; optic neuritis; temporal optic atrophy; aggregation of retinal pigment epithelium; tortuosity of retinal arteries and veins; reduced central vision; retinal lesions; may present with mild-to-moderate reduction of visual acuity with 50% of patients having vision between 20/60 and 20/200; visual field defect associated may be a central, paracentral, or cecocentral scotoma.

Clinical: Keratosis pilaris on the extremities; approximately 10% of patients present with mental abnormalities and approximately 80% of patients with neural hearing loss.

Kivlin JD, et al. Linkage analysis in dominant optic atrophy. *Am J Hum Genet* 1983; 35:1190-1195.

Kjer P. Infantile optic atrophy with dominant mode of inheritance. Copenhagen: Bogtrykkeriet Forum, 1959.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol. 1,* 4th ed. Baltimore: Williams & Wilkins, 1982:320.

Optic Atrophy, Nerve Deafness 935

General: Autosomal recessive.

Ocular: Degeneration of optic nerves.

Clinical: Degeneration of the acoustic nerves; progressive polyneuropathy, distally.

Kollarits CR, et al. The autosomal dominant syndrome of progressive optic atrophy and congenital deafness. *Am J Ophthalmol* 1979; 87:789-792.

Konigsmark BW; Gorlin RJ. *Genetic and metabolic deafness.* Philadelphia: WB Saunders, 1976:108-110.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology. vol. 1,* 4th ed. Baltimore: Williams & Wilkins, 1982:321.

Optic Atrophy, Non-Leber-Type, with Early Onset 936

General: Sex-linked; onset early in life.

Ocular: Optic atrophy.

Clinical: Mental retardation; hyperactive knee jerks; absent ankle jerks, extensor plantar reflexes; dysarthria; tremor; dysdiadochokinesia; difficulty with tandem gait.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Volker-Dieben HJ, et al. A family with sex-linked optic atrophy (ophthalmological and neurological aspects). *Doc Ophthalmol* 1974; 37:307-326.

Optic Atrophy-Spastic Paraplegia Syndrome 937

General: Degenerative disorder of CNS associated with optic atrophy; sex-linked.

Ocular: Optic atrophy.

Clinical: Neurologic spastic paraplegia,

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Went LN. A sex-linked heredodegenerative neurological disorder associated with Leber's optic atrophy. Genetic aspects. *Acta Genet Stat Med* 1964; 14:220-239.

Optic Canal Syndrome 938

General: Caused by severe blow to the head, usually the face or occiput.

Ocular: Unilateral blindness (rarely bilateral); amaurotic pupillary paralysis; pathogenesis is a shearing or tearing injury to the nerve at the transition point from where it is fixed in the optic canal and the free intracranial portion.

Aitken PA, Sofferman RA. Traumatic optic neuropathy. *Ophthalmol Clin North Am* 1991; 4:479.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Snebold NG. Neuroophthalmic manifestations of trauma. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3463-3477.

Optic Disk Traction Syndrome 939

General: Optic disc traction with elevation associated with posterior vitreous detachment or vitreopapillary fibrous membrane may be associated with retinal surgery

Ocular: Central retinal vein occlusion; optic disk traction (vitreopupillary); localized retinal detachment

Rumelt S, Karatar M, Pikkel J, et al. Optic disk traction syndrome associated with central retinal vein occlusion. *Arch Ophthalmol*. 2003; 121: 1093-1097.

Optic Nerve Hypoplasia, Familial (Bilateral, Unilateral) 940

General: Autosomal dominant; congenital defect of optic nerve and retina that occurs in both unilateral and bilateral forms; onset at birth; majority of cases are sporadic, although there are reports of familial cases; association of this condition with maternal ingestion of various substances, including quinine, lysergic acid diethylamide (LSD), and anticonvulsants.

Ocular: Optic nerve hypoplasia; diameter of optic disk is one third normal size; nystagmus; peripapillary halos; situs inversus of disk; strabismus; choroidal atrophy; microphthalmos; coloboma of choroid and/or optic disk; blepharophimosis; ptosis; aniridia; ocular motor nerve palsy.

Clinical: CNS defects; chromosomal abnormalities; cerebral malformations; vascular hypertension.

Hackenbruch Y, et al. Familial bilateral optic nerve hypoplasia. *Am J Ophthalmol* 1975; 79:314-320.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol. 1,* 4th ed. Baltimore: Williams & Wilkins, 1982:346.

Optic Pit Syndrome 941

General: Congenital.

Ocular: Serous detachment of macula; situs inversus; peripapillary chorioretinal changes; cilioretinal vessels; large optic disk; tortuous retinal vessels; retinoschisis.

Clinical: None.

Apple DJ, et al. Congenital anomalies of the optic disc. *Surv Ophthalmol* 1982; 27:3-41.

Apple DJ. New aspects of colobomas and optic nerve anomalies. In: Kivlin J, ed. Developmental abnormalities of the *eye. Int Ophthalmol Clin* 1984; 24:109.

Gass JDM. Serous detachment of the macula secondary to con genital pit of the optic nerve head. *Am J Ophthalmol* 1969; 67:821-841.

Giuffre G. Optic pit syndrome. *Doc Ophthalmol* 1986; 64:187-199.

Lineoff H, et al. Retinoschisis associated with optic nerve pits. *Arch Ophthalmol* 1988; 106:61.

Oral-Facial-Digital Syndrome (OFDS) 942

General: Group of syndromes characterized by congenital anomalies of the oral cavity, face, and digits; see-saw walking; hypertelorism strabismus.

Ocular: Chorioretinal coloboma, optic nerve coloboma, retinal hamartoma.

Clinical: Patients with OFDS have been known to have various congenital anomalies, including cleft palate, tongue hamartomas, bifid tongue, multiple hyperplastic frenula, hypoplastic nasal cartilage, syndactyly, polydactyly, brachydactyly, clinodactyly, and the ocular ophthalmologic findings; nine distinct types of OFDS have been described.

Gurrieri F, et al. Possible new type of oral-facial-digital syndrome with retinal abnormalities: OFDS type (VIII). *Am J Med Genet* 1992; 42:789-792.

Jamieson R, Collins F. Oral-facial-digital syndrome and retinal abnormalities with autosomal recessive inheritance. *Am J Med Genet* 1993; 47:304-306.

Nevin NC, et al. Oral-facial-digital syndrome with retinal abnormalities, OFDS type IX. A further case report. *Am J Med Genet* 1994; 51:228-231.

Sigaudy S, et al. Oral-facial-digital syndrome with retinal abnormalities: report of a new case. *Am J Med Genet* 1996; 61:193-194.

Stevens JL, Marsh JL. Ocular anomalies in the oral-facial-digital syndrome. *J Pediatr Ophthalmol Strabismus* 1994; 31:397-398.

Toriello HV. Heterogeneity and variability in the oral-facial-digital syndromes. *Am J Med Genet* 1988; 4[Suppl]: 149-159.

Tsai PS, O'Brien JM. Retinal hamartoma in oral-facial-digital syndrome. *Arch Ophthalmol* 1999; 117:963-966.

ORF Syndrome (Ecthyma Infectiosum) 943

General: Infectious disease; transmitted between animals; worldwide distribution; human infection from sheep.

Ocular: Pigmentation of lids.

Clinical: Single or multiple lesions of hands and other parts of body; itching; fever; concurrent aseptic meningitis caused by enterovirus.

Duke-Elder S, MacFaul PA, eds. *System of Ophthalmology. vol. XIII.* St. Louis: CV Mosby. 1974.

Gilbert GL, et al. Outbreak of enterovirus 71 infection in Victoria, Australia, with a high incidence of neurologic involvement. *Pediatr Infect Dis J* 1988; 7:484-488.

Tindall JP, Miller GD. Hand-foot and mouth disease. *Cutis* 1972; 9:457.

Ormond Syndrome (Retroperitoneal Fibrosis) 944

General: Periureteral fibrosis that constricts ureter; most frequent in males; average age at onset is 46 years in males and 32 years in females; etiology possibly is a fasciculitis of collagen disease; clinical variant of multiple fibrosclerosis.

Ocular: Exophthalmos; orbital pseudotumor; nongranulomatous panuveitis.

Clinical: Pain in back progressing to ureteral colic or abdominal pain without specific localization; vomiting; nausea; anorexia; malaise; fatigue; weight loss; constipation; diarrhea; pyelonephritis.

Aylward GW et al. Orbital involvement in multifocal fibrosclerosis. *Br J Ophthalmol* 1995; 79:246-249.

Doi M, Uji Y. A case of uveitis associated with idiopathic retroperitoneal fibrosis. *Am J Ophthalmol* 1994; 117: 358-362.

Levine MR, et al. Multifocal fibrosclerosis. Report of a case of bi lateral idiopathic sclerosing pseudotumor and retroperitoneal fibrosis. *Arch Ophthalmol* 1993; 111:841-843.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Ormond JK. Bilateral ureteral obstruction due to envelopment and compression by an inflammatory retroperitoneal process. *J Urol* 1935; 59:1072-1079.

Orzechowski Syndrome (Truncal Ataxia-Opsoclonia; Encephalitis-Opsoclonia- Tremulousness) 945

General: Etiology unknown; follows a benign upper respiratory infection; spontaneous resolution; opsoclonus has been described in patients with encephalitis with Epstein-Barr virus, mumps, and coxsackievirus infections.

Ocular: Opsoclonia; excessive blinking.

Clinical: Incapacitating postural tremulousness of the body; fever; headache.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Orzechowski K. De I'Ataxie Dysmetrique des Yeux; Remarques sur I'Ataxie des Yeux Dite Myoclonique. *J Psychol Neurol* 1927; 35:1-18.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol. 4, part* 2, 4th ed. Baltimore: Williams & Wilkins, 1995:3924,4064.

Osteogenesis Imperfecta Congenita, Microcephaly, and Cataracts 946

General: Autosomal recessive.

Ocular: Cataracts; blue sclera; keratoconus.

Clinical: Brain abnormally small; multiple prenatal bone fractures; calvaria soft; shortening and bowing of lower limbs.

Buyse M, Bull MJ. A syndrome of osteogenesis imperfecta, microcephaly, and cataracts. *Birth Defects* 1978; 14: 95-98.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Ota Syndrome (Nevus of Ota; Oculodermal Melanocytosis; Nevus Fuscoceruleus-Ophthalmomaxillaris Syndrome) 947

General: Affects mainly black and Japanese populations; female preponderance (4:1); mode of transmission unknown; most frequently unilateral; pigmentary changes frequently spread during puberty, but no malignant transformation occurs; malignant transformation to melanoma in the uvea and orbit has been reported.

Ocular: Congenital benign periorbital pigmentation of brown, slate to bluish-black coloration, involving area of first and second (rarely third) division of trigeminal nerve; unilateral hyperchromic heterochromia iridis; possible scleral and conjunctival pigmentation; trabeculae heavily pigmented; slate-gray hyperpigmentation of fundus; optic disk pigmentation (occasionally).

Clinical: Pigmentation of temples, nose, forehead, and malar region; "Mongolian spot" in sacral area (present at birth but usually disappears after puberty).

Bordon AF, et al. Choroidal malignant melanoma in association with oculodermal melanocytosis in a black patient. *Br J Ophthalmol* 1995; 79:191-192.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Cowan TH, Balistocky M. The nevus of Ota or oculodermal melanocytosis: the ocular changes. *Arch Ophthalmol* 1961; 65:483.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Ota M. Nevus fusco-caeruleus ophthalmomaxillaris. *Tokyo Med J* 1939; 63:1243.

Seregard S, et al. Multiple melanocytic tumors in a case of ocular melanocytosis. *Acta Ophthalmol* 1993; 71:562-565.

Otocephaly 948

General: Birth defect; extreme malformation of first brachial arch characterized by almost complete aplasia of its parts.

Ocular: Bilateral anophthalmos.

Clinical: Mouth deformities; absence of lower jaw; joining of ears on the neck.

Duke-Elder S, MacFaul PA. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Glange WD, ed. *The Mosby medical encyclopedia.* St. Louis: CV Mosby, 1985.

Outer Retinal Ischemic Infarction Syndrome 949

General: Complication that may occur during the course of cataract extraction or closed vitrectomy due to obstruction of choroidal circulation.

Ocular: Acute loss of central and paracentral vision; whitening of the outer retinal layers in posterior fundus; mottled changes in the pigment epithelium.

Clinical: None.

Gass DM, Parrish R. Outer retinal ischemic infarction: a newly recognized complication of cataract extraction and closed vitrectomy. *Ophthalmology* 1982; 89:1467-1471.

Weiter JJ, Zuckerman R. The influence of the photoreceptor RPE complex on the inner retina. *Ophthalmology* 1980; 87:113-119.

Oxalosis (Primary Hyperoxaluria; Lepoutre Syndrome) 950

General: Autosomal recessive and acquired forms; metabolic disorders with accumulation of oxalic acid in tissues; type I: glycolic aciduria, defect of 2-oxoglutarate/ glyoxylate carboligase; type II: glyceric aciduria, defect of D-glyceric dehydrogenase.

Ocular: Calcium oxalate deposits may be found in the retina; retinitis punctate albescens; macular degeneration of Stargardt; pigmentary retinopathy; black, geographic central macular subretinal patches; oxalate may deposit in the retinal pigment epithelium, outer plexiform layer, and nuclear layers of the retina.

Clinical: Recurrent calcium oxalate nephrolithiasis and nephrocalcinosis; progressive renal insufficiency; continuous excessive synthesis and excretion of oxalic acid; nausea; vomiting; abdominal pain; renal colic; calculi in urine; tetany.

Francois J. Metabolic tapetoretinal degenerations. *Surv Ophthalmol* 1982; 26:293-333.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Rowe S, et al. Ophthalmologic manifestations of genitourinary diseases. *Urology* 1992; 39:523-525.

Sakamoto T, et al. Ocular histopathologic findings in a 46-year-old man with primary hyperoxaluria. *Arch Ophthalmol* 1991; 109:384-387.

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Page Syndrome (Hypertensive Diencephalic Syndrome) 951

General: Irritation of parasympathetic and sympathetic centers in diencephalon; intradermal histamine 0.25 mg reproduces syndrome; prognosis unpredictable; more frequent in women; onset age 18 to 30 years; spontaneous attacks lasting several minutes.

Ocular: Excessive lacrimation; arteriosclerotic and hypertensive fundus changes.

Clinical: Vasomotor blush over face, neck, and trunk, followed by perspiration; tachycardia; palpitations; elevated baseline blood pressure with additional 20 to 30 mm Hg rise during attacks; reduced respiration, perhaps labored; increased bowel sounds; salivation; sexual frigidity; tremor of hands and generalized tremulousness.

Page IH. A syndrome simulating diencephalic stimulation occurring in patients with essential hypertension. *Am J Med Sci* 1935; 190:9.

Schroeder HA, Goldman ML. Test for the presence of the hypertensive diencephalic syndrome using histamine. *Am J Med Sci* 1949; 6:162.

Paget Disease (Osteitis Deformans; Congenital Hyperphosphatemia; Hyperostosis Corticalis Deformans; Pozzi Syndrome; Chronic Congenital Idiopathic Hyperphosphatemia; Osteochalasis Desmalis Familiaris; Familial Osteoectasia) 952

General: Autosomal dominant; more frequent in men, but more severe in women; onset after age 40 years; characterized by diffuse cortical thickening of involved bones with osteoporosis, bowing deformities, and shortening of stature; osteogenic sarcoma not infrequent.

Ocular: Shallow orbits with progressive unilateral or bilateral proptosis palsy of extraocular muscles; corneal ring opacities; cataract; retinal hemorrhages; pigmentary retinopathy; macular changes resembling Kuhnt-Junius degeneration; angioid streaks; papilledema; optic nerve atrophy; blue sclera; exophthalmos.

Clinical: Skull deformities; kyphoscoliosis; hypertension and arteriosclerosis; muscle weakness; waddling gait; hearing impairment; osteoarthritis.

Eretto P, et al. Optic neuropathy in Paget's disease. *Am J Ophthalmol* 1984; 97:505-510.

Hamdy RC, et al. Clinical presentation of Paget's disease of the bone in older patients. *South Med J* 1993; 86: 1097-1100.

Kheterpal S, et al. Paget's disease presenting with exophthalmos. *Eye* 1994; 8[Part 4]:480-481.

Paget J. On a form of chronic inflammation of bones (osteitis deformans). *Med Chir Trans (Lond)* 1877; 60:37.

Shilling JS, Blach RK. Prognosis and therapy of angioid streaks. *Trans Ophthalmol Soc UK* 1975; 95:301-306.

Simon RM. Paget's disease in the head and neck. *Gerontology* 1980; 26:155-159.

Paine Syndrome 953

General: Microcephaly-spastic diplegia; occurs only in males; onset from birth; sex-linked inheritance; poor prognosis, death usually within first year.

Ocular: Optic atrophy.

Clinical: Microcephaly; poor swallowing; retarded physical and mental development; seizures; lack of interest in environment; hyperreflexia; cerebellar hypoplasia; myoclonic fits; elevated level of amino acids in the cerebrospinal fluid; hypoplasia of cerebellar inferior olives and pons.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Paine RS. Evaluation of familial biochemically determined mental retardation in children, with special reference to amino aciduria. *N Engl J Med* 1960; 262:658-665.

Palatal Myoclonus Syndrome 954

General: Vascular disorders of brainstem that involve inferior olive and olivodentate connection; continuous rhythmic contractions of the palate that occur 100 to 180 times per minute.

Ocular: Opsoclonia; ocular bobbing; nystagmus; torsional, pendular nystagmus; gaze-evoked nystagmus.

Clinical: Contraction of pharynx, larynx, tongue, floor of the mouth, neck, and diaphragm; nodding of head; tremor of hand; sleep apnea; multiple sclerosis.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, 4th ed. Baltimore: Williams & Wilkins, 1995: 4371.

Sakurai N, et al. Sleep apnea and palatal myoclonus in a patient with neuro-Behcet syndrome. *Intern Med* 1993; 32: 336-339.

Yao CB, et al. Ocular bobbing in palatal myoclonus. *Arch Neurol* 1968; 18:304-310.

Pallidal Degeneration, Progressive, with Retinitis Pigmentosa (Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, and Pallidal Degeneration; HARP Syndrome) 955

General: Autosomal recessive; destruction of global pallida and reticular portions of substantia nigra; also may be associated with hypoprebetalipoproteinemia and acanthocytosis; various combinations of components of HARP syndrome may be caused by several distinct genetic diseases or may represent variable manifestations of a contiguous gene defect.

Ocular: Retinitis pigmentosa.

Clinical: Progressive extrapyramidal rigidity; dysarthria.

Higgins JJ, et al. Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP syndrome). *Neurology* 1992; 42:194-198.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Orrell RW, et al. Acanthocytosis, retinitis pigmentosa, and pallidal degeneration: a report of three patients, including the second reported case with hypoprebetalipoproteinemia (HARP syndrome). *Neurology* 1995; 45[3 Part 1]: 487-492.

Winkelman NW Progressive pallidal degeneration: a new clinicopathologic syndrome. *Arch Neurol Psychiatry* 1932; 27:1-21.

Pallister-Killian Syndrome (PKS; Mosaic Tetrasomy) 956

General: Variable condition caused by a mosaic tetrasomy of chromosome 12p.

Ocular: Reported manifestations include hypertelorism, narrow palpebral fissures, prominent epicanthal folds, lower eyelid entropion, ptosis, sparse eyebrows, sparse eyelashes, hypoplastic supraorbital ridge, miotic pupils, sluggish pupils, iris atrophy/hypoplasia, iris heterochromia, cataracts, nystagmus, esotropia, exotropia, optic nerve atrophy, oval optic disks, fundus hypopigmentation, amblyopia, and retinal pigmentary mosaicism.

Clinical: Patients have "coarse" facial features, midface malformations, psychomotor delay, hypotonia, scalp hair sparsity, and variegated lightly and darkly pigmented skin.

Bielanska MM, Khalifa MM, Duncan AM. Pallister-Killian syndrome, a mild case diagnosed by fluorescence in situ hybridization: review of the literature and expansion of the phenotype. *Am J Med Genet* 1996; 65:104-108.

Birch M, Patterson A, Fryer A. Hypopigmentation of the fundi associated with Pallister-Killian syndrome. *J Pediatr Ophthalmol Strabismus* 1995; 32:128-131.

Graham W, et al. Retinal pigment mosaicism in Pallister-Killian syndrome (mosaic tetrasomy 12p). *Arch Ophthalmol* 1999; 117:1648-1649.

Horn D, et al. Pallister-Killian syndrome: normal karyotype in prenatal chorionic villi, in postnatal lymphocytes, and in slowly growing epidermal cells, but mosaic tetrasomy 12p in skin fibroblasts. *J Med Genet* 1995; 32:68-71.

Horneff G, et al. Pallister-Killian syndrome in older children and adolescents. *Pediatr Neurol* 1993; 9:312-315.

Pallister PD. The Pallister mosaic syndrome. *Birth Defects* 1977; 13:103-110.

Reynolds JF, et al. Isochromosome 12p mosaicism (Pallister mosaic aneuploidy or Pallister-Killian syndrome): report of 11 cases. *Am J Med Genet* 1987; 7:257-274.

Schaefer GB, et al. Clinical variability of tetrasomy 120. *Clin Genet* 1997; 51:102-108.

Teschler-Nicola M, Killian W. Case report 72: mental retardation, unusual facial appearance, abnormal hair. *Syndr Ident* 1981; 7:6-7.

Warburton D, Anyane-Yeboa K, Francke U. Mosaic tetrasomy 12p: four new cases, and confirmation of the chromosomal origin of the supernumerary chromosome in one of the original Pallister-Mosaic syndrome cases. *Am J Med Genet* 1987; 27:275-283.

Palpebral Coloboma-Lipoma Syndrome (Nasopalpebral Lipoma-Coloboma) 957

General: Autosomal dominant; described in a Venezuelan family.

Ocular: Coloboma of upper and lower lids at junction between their middle and inner thirds; fat deposits of both upper lids; malposition of lacrimal puncta; hypertelorism; telecanthus.

Clinical: Broad nasal bridge; fatty accumulations on nasal bridge and nasolabial area; maxillary hypoplasia.

Akarsu AN, Sayli BS. Nasopalpebral lipoma-coloboma syndrome. *Clin Genet* 1991; 30:342-344.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Penchaszadeh VB, et al. The nasopalpebral lipoma-coloboma syndrome: a new autosomal dominant dysplasia-malformation syndrome with congenital nasopalpebral lipomas, eyelid colobomas, telecanthus, and maxillary hypoplasia. *Am J Med Genet* 1982; 11:397-410.

Pancoast Syndrome (Hare Syndrome; Superior Pulmonary Sulcus Syndrome) 958

General: Mass occupying lesion in pulmonary apex; erosion of first three ribs frequent; primary bronchogenic carcinoma most frequent cause; symptomatology similar to lower radicular (Dejerine-Klumpke) syndrome and scalenus anticus (Naffziger) syndrome; Horner syndrome caused by involvement of sympathetic chain (see Dejerine-Klumpke Syndrome; Horner Syndrome; Naffziger Syndrome); also can be caused by locally invasive fungus such as *Cryptococcus neoformans* or lymphomatoid granulomatosis.

Ocular: Mild enophthalmos; ptosis; narrowing of the palpebral fissure; miosis.

Clinical: Pulmonary apical tumor; severe shoulder pain; paresthesias, pain, and paresis of the homolateral arm with atrophy of arm and hand muscles.

Brenner B, et al. Pancoast's syndrome in multiple myeloma. *Acta Haematol* 1984; 71:5, 1984, 353-355.

Dolan G, et al. Extrapulmonary lymphomatoid granulomatosis presenting as Pancoast's syndrome. *Postgrad Med J* 1991; 67:914-915.

Esposito S, et al. Therapeutic approach to the Pancoast pain syndrome. *Appl Neurophysiol* 1985; 48:262.

Gallagher KJ, et al. Pancoast syndrome: an unusual complication of pulmonary infection by Staphylococcus aureus. *Ann Thorac Surg* 1992; 53:903-904.

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Lacquet LK. Present views of the surgical treatment of non-cell lung cancer. *Verh K Acad Geneeskd Belg* 1994; 56: 473-493.

Maggi G, et al. Combined radiosurgical treatment of Pancoast tumor. *Ann Thorac Surg* 1994; 57:198-202.

Mantell BS. Superior sulcus (Pancoast) tumors: results of radiotherapy. *Br J Dis Chest* 1973; 67:315.

Mitchell DH, Sorrell TC. Pancoast's syndrome due to pulmonary infection with Cryptococcus neoformans variety gattii. *Clin Infect Dis* 1992; 14:1142-1144.

Okubo K, et al. Treatment of Pancoast tumors. Combined irradiation and radical resection. *Thorac Cardiovasc Surg* 1995; 43:284-286.

Pancoast H. Importance of careful roentgen-ray investigation of apical chest tumor. *J Am Med Assoc* 1924; 83: 1407.

Remmen HJ, et al. Surgical treatment of Pancoast tumor. *J Cardiovasc Surg (Torino)* 1993; 34:157-161.

Shamji FM, et al. Acute Pancoast's syndrome caused by fungal infection. *Can J Surg* 1988; 31:441-443.

Simpson FG, Morgan M, Cooke NI Pancoast's syndrome associated with invasive aspergillosis. *Thorax* 1986; 41: 156-157.

Vandeplas O, et al. Pancoast's syndrome due to Pseudomonas aeruginosa infection of the lung apex. *Thorax* 1991; 46:683-684.

Wang JC, et al. Pancoast syndrome in a patient with malignant lymphoma. *Cancer* 1989; 64:2588-2590.

Pancreatitis 959

General: Inflammation of pancreas.

Ocular: Xerosis; night blindness; multiple branch retinal artery occlusions; cotton-wool patches; retinal edema; striate and blot hemorrhages; retinopathy of pancreatitis has been considered to indicate multiple-organ failure and poor prognosis in severe acute pancreatitis; mechanism may be secondary to granulocyte aggregation and leukoembolization due to activated complement.

Clinical: Chronic pancreatitis; lipid emboli; malabsorption; vitamin A deficiency.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Hollo G, et al. Retinopathy of pancreatitis indicates multiple-organ failure and poor prognosis in severe acute pancreatitis. *Acta Ophthalmol* 1994; 72:114-117.

Horn HB, Ranson J. Evidence of complement catabolism in acute pancreatitis. *Am J Med Sci* 1978; 275:257.

McCoy LS, Morse PH. Retinopathy associated with acute pancreatitis. *Am J Ophthalmol* 1985; 100:246-251.

Papilloma (Wart; Verruca) 960

General: Cutaneous or mucosal tumor of proliferating epithelial and fibrovascular tissues; viral etiology or noninfectious.

Ocular: Papillary conjunctivitis; pseudopterygium; corneal opacity; epithelial keratitis; corneal vascularization; lid ulcers; lacrimal system obstruction; hemorrhages of conjunctiva, lids, and lacrimal system.

Clinical: Mulberry- or cauliflower-like tumors that may occur on any cutaneous or mucosal surface.

Schachat A, et al. Carbon dioxide laser therapy of recurrent squamous papilloma of the conjunctiva. *Ophthalmol Surg* 1982; 13:916-918.

Shields JA, Shields CL. Tumors of the conjunctiva and cornea. In: Smolin G, Thoft RA, eds. *The Cornea*. Boston: Little, Brown and Company, 1994:585.

Wilson FM II, Ostler HB. Conjunctival papillomas in siblings. *Am J Ophthalmol* 1974; 77:103-107.

Papillon-Leage-Psaume Syndrome (Oro-Digital-Facial Syndrome; Linguofacial Dysplasia of Grob; Gorlin Syndrome; Dysplasia Linguofacialis; OFD Syndrome; Oro-Digital-Facial Dysostosis; Grob Linguofacial Dysplasia) 961

General: Familial with strong female preponderance; transmitted as a dominant; partial trisomy has been suggested for the 6-12 (C) chromosome.

Ocular: Hypertelorism; displaced medial and lateral canthi; antimongoloid slanting of palpebral fissures; exotropia; see-saw winking.

Clinical: Clefts of jaws and tongue due to abnormalities in development of frenulum; syndactyly; polydactyly; alopecia; white, hamartomatous patches of midline of tongue; mental retardation; bradydactyly; hypoplastic nasal cartilages; seborrheic changes; dystopia canthus; pseudocleft of upper lip; alopecia; missing mandibular lateral incisors.

Gorlin RJ, Psaume J. Orodigitofacial dysostosis: a new syndrome. *J Pediatr* 1962; 61:520

Grob M. Dysplasia linguo-facialis. In: *Lehrbuch der kinderheikunde.* Stuttgart: Thieme, 1957:98.

Papillon-Leage E, Psaume J. Une malformation hereditaire de la mugueuse buccale brides et freins anormaux: Generalites. *Rev Stomatol* 1954; 55:209-227.

Sugarman GI, et al. See-saw winking in a familial oral-facial digital syndrome. *Clin Genet* 1971; 2:248.

Papillon-Lefevre Syndrome (Hyperkeratosis Palmoplantaris with Periodontosis) 962

General: Autosomal recessive; onset between ages 1 and 4 years.

Ocular: Nystagmus.

Clinical: Hyperkeratosis of the palms and soles associated with destruction of the periodontal ligament and premature exfoliation of the teeth; bad breath; loose teeth; loss of teeth.

Mhaiskar D, et al. Papillon-Lefevre syndromes with ocular nystagmus (a case report). *J Postgrad Med* 1980; 26: 267-268.

Papillon MM, Lefevre P. Deux Cas de Keratodermie Palmaire et Plantaire Symetrique Familiale (Maladie de Meleda) Chez le Frere et la Soeur. Coexistence dans les Deux Cas d' Alterations Dentaires Graves. *Bull Soc Fr Dermatol Syphil* 1924; 31:82-87.

Pappataci Fever (Phlebotomus Fever; Sandfly Fever) 963

General: Viral etiology; transmitted by the sandfly Phlebotomus *papatasii.*

Ocular: Pick sign of conjunctiva (conjunctival injection limited to the exposed portion of the conjunctiva); uveitis; optic neuritis; papilledema; papillitis; blepharospasm; retinal venous engorgement; vitreal exudates.

Clinical: Fever; headaches; myalgia; pain; stiffness of the neck and back.

Duke-Elder S, ed. *System of Ophthalmology, vol. XV.* St. Louis: CV Mosby, 1976:142.

Johnson KM. Sandfly fever. In: Beeson, PB, McDermott W, Wyngaarden JB, eds. *Textbook of medicine.* 15th ed. Philadelphia: WB Saunders, 1979:282-283.

Paragonimiasis (Distomiasis; Endemic Hemoptysis) 964

General: Chronic lung infection; causative agent is *Paragonimus trematode;* transmitted by eating undercooked crabs or crayfish and drinking contaminated water; affects all ages; most severe in children.

Ocular: Convergence paralysis; optic atrophy; lid cysts; dacryocystitis; papilledema; homonymous hemianopsia.

Clinical: Hemoptysis; dyspnea; fever; anorexia; weight loss; pleural effusion; pneumothorax; diarrhea; epileptic seizures; hemiplegia; leukocytosis; eosinophilia; anemia; cystic bronchiectasis; encephalitis; chronic bronchitis; intestinal or peritoneal infections; central nervous system (CNS) paragonimiasis occurs predominantly in persons under age 30 years.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Kirkpatrick JB. Neurologic infections due to bacteria, fungi and parasites. In: Davis RL, Robertson DM, eds. *Textbook of neuropathology,* 2nd ed. Baltimore: Williams & Wilkins, 1991:719-803.

Kusner DJ, King CH. Cerebral paragonimiasis. *Semin Neurol* 1993; 13:201-208.

Romeo DP, et al. Pulmonary paragonimiasis: diagnostic value of pleural fluid analysis. *South Med J* 1986; 79: 241-243.

Parinaud Oculoglandular Syndrome (Parinaud Conjunctiva-Adenitis Syndrome; Catscratch Oculoglandular Syndrome; Catscratch Disease; Bartonella henselae) 965

General: Most frequently seen in children; incubation time 7 to 10 days; caused by small pleomorphic Gram-negative bacillus; good prognosis; affects both sexes; about 90% of patients with this condition have serologic evidence of infection by *Rochalimaea henselae.*

Ocular: Conjunctivitis; retrotarsal conjunctival granulations; formation of granulomata in anterior segment about 3 mm high and 2 to 6 mm in diameter; inferior fornix usually affected; ulceration common; neuroretinitis; optic neuritis.

Clinical: Tender, red papule at the site of a cat scratch; regional preauricular and cervical lymphadenitis (often only one gland involved); irregular fever for 4 to 5 days and malaise; fever; parotid gland swelling.

Carithers HA. Oculoglandular disease of Parinaud: a manifestation of cat-scratch disease. *Am J Dis Child* 1978; 132: 1195-1200.

Chrousos Ga, et al. Neuroretinitis in cat scratch disease. *J Clin Neuro-Ophthalmol* 1990; 10:92-94.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Golden SE. Hepatosplenic cat-scratch disease associated with elevated anti Rochalimaea antibody titers. *Pediatr Infect Dis J* 1993; 12:868-871.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 2, 4th ed. Baltimore: Williams & Wilkins, 1995.

Parinaud H, Galezowski X. Conjonctivite Infectieuse Transmise par les Animaux. *Ann Ocul (Brux)* 1889; 101:252.

Wear DJ, et al. Cat scratch disease bacilli in the conjunctiva of patients with Parinaud's oculoglandular syndrome. *Ophthalmology* 1985; 92: 1282-1287.

Parinaud Syndrome (Divergence Paralysis; Subthalamic Syndrome; Paralysis of Vertical Movements; Pretectal Syndrome) 966

General: Various causes, including pineal tumor, supranuclear lesions, vascular lesions, inflammation, hemorrhages, midbrain lesions, lesion of posterior white commissure of pons, red nucleus, or superior cerebellar peduncle; combination of Parinaud and von Monakow syndromes is known as Gruner-Bertolotti syndrome, which consists of paralysis in upward gaze, tremors, hemiplegia, and sensory disturbances.

Ocular: Retraction of lids with lesion in mesencephalic gray matter and ptosis with lesions more anteriorly; paralysis of conjugate upward movement of the eye without paralysis of convergence; occasionally paralysis of upward and downward movement; spasm with convergence insufficiency; contralateral hemianopsia occurs when the lateral geniculate body becomes involved in case of infiltrating tumor; wide pupils that fail to react to light but sometimes react during accommodation (Holmes); papilledema (usually severe).

Clinical: Vertigo; contralateral cerebellar ataxia and choreoathetoid movement if lesion involves superior cerebellar peduncle after decussation.

Avman N, et al. Symptomatology, evaluation and treatment of aqueductal stenosis. *Neurol Res* 1984; 6:194-198.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Keane JR. The pretectal syndrome: 206 patients. *Neurology* 1990; 40:684-690.

Oppenheim JS, et al. Ependymomas of the third ventricle. *Neurosurgery* 1994; 34:350-353.

Parinaud H. *Le Strabisme et Son Traitement.* Paris: G. Doin, 1899.

Solomon RA, Stein BM. Interhemispheric approach for the surgical removal of thalamocaudate arteriovenous malformations. *J Neurosurg* 1987; 66:345-351.

Waltman SR. Ocular signs in midbrain disease. In: Gay AJ, Burde RM, eds. Clinical concepts of neuro-ophthalmology. *Int Ophthalmol Clin* 1967; 7:807.

Parkinson Syndrome (Paralysis Agitans; Shaking Palsy) 967

General: Late stages of epidemic encephalitis; present with arteriosclerosis and with manganese and carbon monoxide poisoning; widespread destruction of pigmented cells in substantia nigra.

Ocular: Decreased blinking; lid fluttering; blepharospasm; oculogyric crises; ocular hypotony; blepharoplegia; ptosis; nystagmus; paralysis of convergence; paralysis of lateral rectus muscle; absent or sluggish pupillary reactions to light or convergence; mydriasis or anisocoria; optic neuritis; papilledema; abnormal saccades.

Clinical: Slowness of movements; loss of facial expression; "cogwheel" rigidity of the arms; rhythmical tremors; drooling; shuffling gait; stooping; monotonous voice.

Bronstein AM, Kennard C. Predictive ocular motor control in Parkinson's disease. *Brain* 1985; 108:925.

Cannon SZ. Basic mechanisms of ocular motor control. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994.

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Guiloff RJ, et al. Reversible supranuclear ophthalmoplegia associated with parkinsonism. *J Neurol Neurosurg Psychiatry* 1980; 44:552-554.

Parkinson J. *An essay on the shaking palsy.* London: Sherwood, Nesly and Jones, 1817.

Shibasaki H, et al. Oculo-motor abnormalities in Parkinson's disease. *Arch Neurol* 1979; 36:360-364.

Shimizu N, et al. Eye-head co-ordination in patients with parkinsonism and cerebellar ataxia. *J Neurol Neurosurg Psychiatry* 1981; 44:509-515.

Parotid Aplasia or Hypoplasia (Salivary Gland Absence; Lacrimal Puncta Absence) 968

General: Autosomal dominant.

Ocular: Lacrimal gland aplasia; absence or severe dysfunction of lacrimal glands.

Clinical: Aplasia of parotid salivary glands; hemifacial microsomia; mandibulofacial dysostoses; xerostomia; rampant caries; edentulous, salivary gland dysfunction; parotid agenesis or hypoplasia; impalpable parotid gland; absence of the orifice of Stensen duct; bilateral parotid gland aplasia.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Wiesenfeld D, et al. Familial parotid gland aplasia. *J Oral Med* 1985; 40:84-85.

Partial Trisomy 16q Syndrome 969

General: Partial trisomy 16q with chromosome banding; rare.

Ocular: Narrow palpebral fissures; antimongoloid lid slant; hypertelorism; strabismus; epicanthus; congenital glaucoma; corneal edema; shallow anterior chamber; Rieger anomaly.

Clinical: Dry skin; periorbital edema; hydrocele; general hypotonia; low-set ears; micrognathia; hypoplastic lower lip; long philtrum; stumpy hands with short fingers; pectus excavatum.

Balestrazzi P, et al. Partial trisomy 16q resulting from maternal translocation. *Hum Genet* 1979; 49:229-235.

Ferguson JG, et al. Rieger's anomaly and glaucoma associated with partial trisomy 16q. *Arch Ophthalmol* 1987; 105:323.

Passow Syndrome (Bremer Status Dysraphicus; Status Dysraphicus Syndrome; Syringomyelia; Syringobulbia) 970

General: Congenital nonclosure of the neural tube; familial occurrence or may be sporadic; insidious onset in second to third decade of life.

Ocular: Enophthalmos; ptosis; rotatory nystagmus; heterochromia iridis; anterior uveitis; corneal anesthesia; neuroparalytic keratitis; paralysis of third, fifth, sixth, and seventh cranial nerves; Horner syndrome; anisocoria; papilledema; optic atrophy; zonular cataract (see Horner Syndrome).

Clinical: Anesthesia over area of first division of trigeminal nerve; facial hemiatrophy; facial nerve paralysis; muscular weakness; cervical ribs; kyphoscoliosis; spina bifida; unilateral numbness of fingers; loss of deep reflexes; insensitivity to pain and temperature in affected areas; neurogenic bladder.

Duke-Elder S, ed. *System of Ophthalmology, vol. III, part* 2. St. Louis: CV Mosby, 1976:1034.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Mancall EL, McCormick PC. Syringomyelia. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1991:752-753.

Menkes JH, Till Kenneth. Syringomyelia. In: Menkes JH, ed. *Textbook of child neurology,* 5th ed. Baltimore: Williams & Wilkins, 1995:265-267.

Minzter RM, Buckley EG. Neurocranial defects with neuro-ophthalmic significance. In: Wright KW, ed. *Pediatric Ophthalmology and Strabismus*. St. Louis: Mosby, 1995:850.

Passow A. Status dysraphicus. *Klin Monatsbl Augenheilkd* 1935; 94:102.

Ropper AH, Martin JB. Syringomyelia. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine,* 13th ed. New York: McGraw-Hill, 1994:2358.

Patterned Dystrophy of Retinal Pigment Epithelium 971

General: Autosomal dominant; probably there is a migration of pigment granules in the pigment epithelium, resulting in a specific configuration.

Ocular: Reticular fishnet-like dystrophy; macroreticular (spider shaped); butterfly-shaped pigment dystrophy of fovea.

Clinical: None.

Deutman AF. Macular dystrophies. In: Ryan SJ. ed. *Retina, vol. II*, 2nd ed. St. Louis: Mosby, 1994.

Hsieh RC, et al. Patterned dystrophies of retinal pigment epithelium. *Arch Ophthalmol* 1977; 95:429-435.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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O'Donnel FE, et al. Autosomal dominant dystrophy of the retinal pigment epithelium. *Arch Ophthalmol* 1979; 97:680.

Pediculosis and Phthiriasis 972

General: Infestation of lice on head, body, or pubic area.

Ocular: Conjunctivitis; keratitis; infestation of lice or nits glued to shafts of eyelashes and eyebrow.

Clinical: Pruritus; skin excoriation; impetigo; pyoderma with lymphadenitis and febrile episodes.

Awan KJ. Cryotherapy in phthiriasis palpebrarum. *Am J Ophthalmol* 1977; 83:906-907.

Couch JM, et al. Diagnosing and treating phthiriasis pubis palpebrarum. *Surv Ophthalmol* 1982; 26:219.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Mathew M, et al. A new treatment of phthiriasis palpebrarum. *Ann Ophthalmol* 1982; 14:439-441.

Wilhelmus KR. Parasitic keratitis and conjunctivitis, *Clinical* disease. In: Smolin G, Thoft RA, eds. *The Cornea.* Boston: Little, Brown and Company, 1994:272.

Pelizaeus-Merzbacher Disease (Aplasia Axialis Extracorticalis Congenita; Sudanophilic Leukodystrophy) 973

General: Rare; subdivision of diffuse cerebral sclerosis predominantly involving the white matter of CNS; onset in infancy or childhood; X-linked recessive; male members affected through normal-appearing carrier mothers; abnormal myelin/myelin sheath structure secondary to abnormal gene on the X chromosome.

Ocular: Lateral, rotatory, or vertical nystagmus or nonrhythmic wandering eye movements; visual impairment from occipital lobe involvement; tapetoretinal degeneration characteristic of retinitis pigmentosa; attenuated arterioles; optic atrophy; papilledema.

Clinical: Retarded development; gait instability, ataxia; intention tremor and athetosis; hearing and speech disturbances; atrophic disturbances; spastic paralysis; weight gain.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hudson A, et al. Mutation of the proteolipid protein gene PLP in a human X chromosome-linked myelin disorder. *Proc Natl Acad* Sci *USA* 1989; 86:8128-8131.

Merzbacher L. Eine Eigenartige Familiarhereditare Erkrankungsform Caplasia Axialis Extracorticals Congenital. *Z Ges Neurol Psychiatr* 1910; 3:1.

Pelizaeus F. Uber eine Eigentumliche Form Spastischer Laahrnung mit Zerebralerschmungen auf Hereditarer Grundlage (multiple sclerose). *Arch Psych Netvenkr* 1885; 16:698.

Pemphigus Foliaceus (Cazenave Disease) 974

General: Attacks individuals of any race, age, or sex; high incidence in Brazil; characterized by bullous skin lesions resulting in generalized exfoliation.

Ocular: Exfoliative or bullous lesions of lid and conjunctiva; pannus; infiltration of cornea and iris; cataract.

Clinical: Cutaneous manifestations that progress to scaling, crusted patches; simulates lupus erythematosus and exfoliative erythroderma.

Amendola F. Ocular manifestations of pemphigus foliaceus. *Am J Ophthalmol* 1949; 32:35.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Falk ES. Pemphigus foliaceus in a patient with rheumatoid arthritis and Sjogren's syndrome: a case report. *Dermatologica* 1979; 158:348-354.

Mondino BJ. Clinical immunologic diseases. Dermatologic disease. In: Smolin G, Thoft RA, eds. *The Cornea*. Boston: Little, Brown and Company, 1994:425.

Pemphigus Vulgaris 975

General: Primarily in middle-aged people; prognosis varies, from poor to chronic; generalized bullous eruption; blistering autoimmune disease that affects the skin and mucous membranes; association between particular HLA-DR4 and pemphigus vulgaris has been reported.

Ocular: Conjunctival bullae; catarrhal conjunctivitis; scarring and adhesions of conjunctiva.

Clinical: Cutaneous blisters, which may be clear, pustular, or hemorrhagic.

Duane TD, et al. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Scharf SJ, et al. Specific HLA-DQB and HLA-DRB1 alleles confer susceptibility to pemphigus vulgaris. *Proc Natl Acad Sci USA* 1989; 86:6215-6219.

Pendred Syndrome (Sporadic Goiter with Deafness) 976

General: Autosomal recessive; defect in thyroxine biosynthesis.

Ocular: Retinal pigmentary degenerative changes; pendular nystagmus; bull's-eye-type macular degeneration.

Clinical: Thyroid enlargement; sensorineural hearing loss; mental retardation; thyroid carcinoma.

Fraser GR. Deafness, congenital with sporadic goiter (Pendred syndrome). In: *Handbook of Clinical Neurology.* Amsterdam, North Holland, 1981:368-370.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pendred V. Deaf mutism and goitre. *Lancet* 1896; 2:532.

Pericentric Syndrome (pericentric Inversion of Chromosome 11) 977

General: Etiology unknown; leukocyte chromosomes show a pericentric inversion of chromosome 11.

Ocular: Strabismus; hypertelorism; congenital glaucoma; aniridia; corneal disease; epicanthic folds.

Clinical: Microcephaly; broad nasal bridge; arched palate; hyperextensibility of elbows; left hand shows single transverse crease, and right hand shows three palmar creases radiating from radial border.

Broughton WL, et al. Congenital glaucoma and other ocular abnormalities associated with pericentric inversion of chromosome 11. *Arch Ophthalmol* 1983; 101:594-597.

Pergament E. The pericentric syndrome. *Lancet* 1967; 2:777.

Periocular Metastatic Tumors (Ocular Metastatic Tumors) 978

General: Neoplasms that develop from malignant cells and are carried from a primary site of malignancy.

Ocular: Retinal detachment; retinal hemorrhages; enophthalmos; exophthalmos; proptosis; rubeosis iridis; uveitis; papilledema; orbital hemorrhages; hyphema; paralysis of extraocular muscles; secondary glaucoma.

Clinical: Metastasis in the bloodstream and lymphatic system common; tumors of the lung or breast metastasize to globe; neoplasms that most commonly metastasize to the or-bit are neuroblastomas of suprarenal medulla and retroperitoneal ganglia; Wilms tumor may involve the orbit.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Ferry AP, Font RL. Carcinoma metastatic to the eye and orbit. I. A clinicopathologic study of 227 cases. *Arch Ophthalmol* 1974: 92:276-286.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Leys AM, et al. Metastatic carcinoma to the retina: clinicopathologic findings in two cases. *Arch Ophthalmol* 1990; 108:1448.

Peyster RG, et al. Orbital metastasis: role of magnetic resonance imaging and computed tomography. *Radiol Clin North Am* 1987; 25:647.

Peroneal Muscular Atrophy (PMA; Neuropathy, Hereditary Sensorimotor, with Upper Motor Neuron, Visual Pathway, and Autonomic Disturbance) 979

General: Peroneal muscular atrophy with involvement of other parts of nervous system; autosomal dominant; upper motor neuron and visual pathway lesions.

Ocular: Visual pathway lesions; ptosis; irregular pupils; iris atrophy; lack of response to light or near vision.

Clinical: Distal weakness and muscle atrophy; absent ankle jerks; foot drop; stocking-type sensory loss; diminished sweating in distal limbs.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Rechthand E, et al. Hereditary neuropathy with upper motor-neuron, visual pathway, and autonomic disorders. *Neurology* 1983; 33:1495-1497.

Pertussis (Whooping Cough) 980

General: Causative agent *Haemophilus pertussis (Bordetella pertussis);* not all patients who develop pertussis encephalopathy are children.

Ocular: Conjunctivitis; severe cortical blindness; papilledema; choroiditis; retinal ischemia; ocular muscle palsies; hemorrhages of eyelids, conjunctiva, orbit, anterior chamber, and retina; chronic papilledema; optic neuritis; retinal and vitreous hemorrhages, and even intracranial and subarachnoid hemorrhage associated with increased intrathoracic and intraabdominal pressures during coughing.

Clinical: Respiratory tract infection; nasal discharge; cough ending with a loud crowing; inspiratory noise (the "whoop"); thick mucoid sputum; soreness over trachea; ulcer of glottis; vomiting; tetany; encephalopathy; cortical blindness.

Halperin SA, Marcie TJ. Pertussis encephalopathy in an adult: case report and review. *Rev Inject* Dis 1991; 13: 1043-1047.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Hewlett EL: Bordetella species. In: Mandell GL, Douglas RG Jr, Bennett JE, eds. *Principles and Practice of infectious diseases*, 3rd ed. New York: Churchill Livingstone, 1990: 1756-1762.

Keller MA. Adenovirus infection and pertussis-like syndrome. *J Pediatr* 1980; 96:177-178.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995:3087.

Peters Anomaly 981

General: Autosomal recessive; may be morphologic entity with several eye syndromes, including Rieger syndrome, Mietens syndrome, and fetal alcohol syndrome; may be due to a developmental field defect, a contiguous gene syndrome, or a defective homeotic gene controlling development of the eye and other body structures.

Ocular: Corneal opacification; lenticulocorneal adherence; iris adhesions; glaucoma; cataract; narrow lid fissures; colobomatous microphthalmia; persistent hyperplastic primary vitreous; retinal detachment; iris nodules.

Clinical: Short-limbed dwarfism; broad face; thin upper lip; hypoplastic columnella; hypospadias; cleft lip and palate; craniofacial abnormalities; congenital heart disease; horseshoe kidney; polycystic kidneys; Wilms tumor; mental retardation; external ear anomalies; camptodactyly.

Bateman FB, et al. Peters' anomaly associated with partial deletion of the long arm of chromosome 11. *Am J Ophthalmol* 1984; 98:11-15.

Heon E, et al. Peters' anomaly. The spectrum of associated ocular and systemic malformations. *Ophthalmic Paediatr Genet* 1992; 13:137-143.

Kivlin JD, et al. Peters' anomaly as a consequence of genetic and nongenetic syndrome. *Arch Ophthalmol* 1986; 104: 61-64.

Peters A. Ueber Angeborene Defektbildung der Descemetshem Membrane. *Klin Monatsbl Augenheilkd* 1906; 44: 27-41.

Pillay VK. Ophthalmolo-mandibulo-melic dysplasia. *J Bone Joint Surg Am* 1964; 46:858-862.

Traboulsi EI, Maumenee IH. Peters' anomaly and associated congenital malformations. *Arch Ophthalmol* 1992; 110: 1739-1742.

Petzetakis-Takos Syndrome (Phlyctenular Keratoconjunctivitis) 982

General: Malnutrition; lack of hygiene.

Ocular: Superficial keratitis; palpebral edema; cornea hyperesthesia; photophobia; blepharospasm; decreased pupillary response; xerophthalmia.

Clinical: Lymph node hypertrophy.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Petzetakis M. Les Troubles Oculaires Pendant la Trophopenie et l'Epidemic de la Pellagre. La Keratopathie Superficielle Trophopenique. *Presse Med* 1950; 58:1082-1084.

Peutz-Touraine Syndrome (Peutz-Jeghers Syndrome) 983

General: Recognized in infants; autosomal dominant; gastrointestinal polyps; jejunal polyps are consistent feature; these lesions are benign, melanin spots of the lips; buccal mucosa and digits represent second part of syndrome.

Ocular: Brownish speckled dots along border of skin and mucosa of eyelids; freckles similar to those seen on eyelids may appear on conjunctiva, primarily adjacent to limbus and along area of lid fissure; pigment speckles of sclera and iris; brown-pigmented corneal spot.

Clinical: Brownish to black dotlike pigmentation similar to that seen on eyelids is present in the perinasal, perioral, and periorbital regions, fewer on fingers and toes at birth; polyposis of gastrointestinal tract (primarily small intestine) with associated bleeding; potential for malignant transformation in second decade of life.

Jeghers H, et al. Generalized intestinal polyposis and melanin spots of the oral mucosa, lips and digits: syndrome of *Clinical* significance. *N Engl J Med* 1949; 241:993.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Peutz JLA. Over een Zeer Merkvaardige, Gecombineerde Familiaire Polypossi van de Slijmvliezen, van den Tractus Intestinalis met die van de Neuskeelholte en Gepaard met Eigenaardige Pigmentaties van Huid en Slijmvliezen. *Ned Mschr Geneesk* 1921; 10:134.

Reid JD. Intestinal carcinoma in the Peutz-Jeghers syndrome. *JAMA* 1974; 229:833.

Rustgi AK, Marcus DM. Gastrointestinal and nutritional disorders. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:2978.

Pfeiffer Syndrome 984

General: Congenital craniosynostotic syndrome with a high rate of mortality shortly after birth.

Ocular: Severe proptosis (also described as extreme exophthalmic midface hyperplasia).

Clinical: Cloverleaf skull, elbow ankylosis, broad thumbs, and/or broad halluces; variable additional abnormalities including pulmonary problems, brain abnormalities, and prematurity that frequently lead to early death.

Barone CM, et al. Craniofacial, limb, and abdominal anomalies in a distinct syndrome: relation to the spectrum of Pfeiffer syndrome type 3. *Am J Med Genet* 1993; 45:745-750.

Muller D, Steinberger D, Kunze S. Molecular genetics of craniosynostotic syndromes. *Graefes Arch Clin Exp Ophthalmol* 1997; 235:545-550.

Plomp AS, et al. Pfeiffer syndrome type 2: further delineation and review of the literature. *Am J Med Genet* 1998; 75:245-251.

Photosensitivity and Sunburn 985

General: Enhanced responsiveness to natural or artificial nonionizing electromagnetic radiation; photosensitivity induced by exogenous agents accounts for an increasing portion of the total undesirable effects caused by environmental chemicals.

Ocular: Photo-keratoconjunctivitis; conjunctivitis; erythema of periorbital skin; photophobia; lacrimation; blepharospasm.

Clinical: Erythema of skin; edema; vesiculation of skin; fever; nausea; chills; delirium; irregular pigmentation of skin.

Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine,* 11th ed. New York: McGraw-Hill, 1987.

Lerman S. *Radiant energy and the eye.* New York: Macmillan, 1980.

Potter TS, Hashimoto K. Cutaneous photosensitivity to medications. *Compr Ther* 1994; 20:414-417.

Pierre-Robin Syndrome (Robin Syndrome; Micrognathia-Glossoptosis Syndrome) 986

General: Etiology unknown; manifestations at birth; pathogenesis based on arrested fetal development; history of intrauterine disturbance in early pregnancy (25% of cases); also increased incidence in offspring of mothers age 35 years or older; pathogenesis is thought to be incomplete development of the first brachial arch, which forms the maxilla and mandible.

Ocular: Microphthalmos; proptosis; ptosis; high myopia; glaucoma; cataract (rare); retinal disinsertion; megalocornea; iris atrophy; blue sclera; esotropia; conjunctivitis; distichiasis; vitreoretinal degeneration; retinal detachments.

Clinical: Micrognathia; cleft palate; glossoptosis; cyanosis; facial expression birdlike with flat base of nose and high-arched deformed palate with or without cleft; difficulty breathing.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fong DS, Pruett RC. Systemic associations with myopia. In: Albert DM, Jakobiec FA, eds. Principles *and Practice* of *Ophthalmology*, vol. V. Philadelphia: WB Saunders, 1994:3142.

Fraunfelder FT, Roy PH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Robin P. La Chute de la Base de la Langue Consideree Comme une Nouvelle Cause de Gene dans Respiration Naso-Pharyngienne. *Bull Acad Med* 1923; 88:377.

Schreiner RL, et al. Stickler syndrome in a pedigree of Pierre Robin syndrome. *Am J Dis Child* 1973; 126:86-90.

Smith JL, et al. Ocular manifestations of the Pierre Robin syndrome. *Arch Ophthalmol* 1960; 63:984.

Pigmentary Ocular Dispersion Syndrome (Pigmentary Glaucoma) 987

General: Polygenic inheritance; onset at average age 52 years; distribution of pigment in chamber angle; atrophy of posterior iris epithelium; most commonly affects young Caucasian males with myopia; mechanism is likely to be mechanical rubbing between iris pigment epithelium and packets of lens zonules, resulting in aqueous flow obstruction secondary to accumulation of pigment granules in the trabecular meshwork.

Ocular: Myopia; glaucomatous field changes; ocular hypertension; iris translucency; abnormal number of iris processes; insertion of iris anterior to scleral spur; pigmentation of posterior trabecular meshwork, grades 3 to 4; Krukenberg spindles; presence of pigmentation on equatorial border of lens capsule; glaucomatous cupping and myopic optic nerve changes.

Becker B, et al. The pigment dispersion syndrome. *Am J Ophthalmol* 1977; 83:161.

Farrar SM, Shields MB. Current concepts in pigmentary glaucoma. *Surv Ophthalmol* 1993; 37:233-252.

Jabbur NS; Tuli S; Barequet IS, et al. Outsome of laser in situ keratomileusis in patients with pigment dispersion syndrome. *J Cataract Refract Surg* 2004; 30: 110-114.

Lehto I. Long-term prognosis of pigmentary glaucoma. *Acta Ophthalmol* 1991; 69:437-443.

Lunde MW. Argon laser trabeculoplasty in pigmentary dispersion syndrome with glaucoma. *Am J Ophthalmol* 1983; 96:721-725.

Richter CU, et al. Pigmentary dispersion syndrome and pigmentary glaucoma. *Arch Ophthalmol* 1986; 104:211-215.

Pillay Syndrome (Ophthalmomandibulomelic Dysplasia) 988

General: Autosomal dominant; both sexes affected.

Ocular: Corneal opacities.

Clinical: Temporomandibular fusion; obtuse mandibular angle; short forearms.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: Lippincott, 1981.

Pillay VK. Ophthalmomandibulomelic dysplasia. An hereditary syndrome. *J Bone Joint Surg* 1964; 46:858-862.

Pilodental Dysplasia with Refractive Errors (Euhidrotic Ectodermal Dysplasia; Trichodental Dysplasia with Hyperopia) 989

General: Autosomal recessive; damage to structures arising from the ectoderm.

Ocular: Hyperopia; astigmatism.

Clinical: Hypodontia; abnormally shaped teeth; scalp hypotrichosis; disorders of pigmentation of the hair; follicular hyperkeratosis on the trunk and limbs; intensified delineation and reticular hyperpigmentation of the skin of the nape; broadening of bridge and dorsum of the nose.

Kopysc Z, et al. A new syndrome in the group of euhidrotic ectodermal dysplasia: pilodental dysplasia with refractive errors. *Hum Genet* 1985; 70:376-378.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Pinta (Nonvenereal Treponematosis) 990

General: Caused by spirochete *Treponema carateum;* infectious; contagious; found in Mexico, Central America, West Indies, and the northern countries of South America; caused by an organism that is morphologically and antigenically identical to the causative agent of venereal syphilis.

Ocular: Hypopigmentation of eyelid.

Clinical: Cutaneous lesions with marked pigmentary changes; chronic relapsing course.

Domonkos AN, Arnold HL, eds. *Andrew's diseases of the skin,* 7th ed. Philadelphia: WB Saunders, 1982:473-477.

Koff AB, Rosen T. Nonvenereal treponematoses: yaws, endemic syphilis, and pinta. *J Am Acad Dermatol* 1993; 29: 519-535.

Roy FH. *Ocular Differential Diagnosis*, 7th ed. Philadelphia: Lippincott Williams & Wilkins, 2002.

PISK (Pressure Induced Intralamellar Stromal Keratitis) 991

General: Associated with LASIK post operative patients

Ocular: Elevated IOP; ocular discomfort; blurred vision; stromal keratitis

Belin MW, Hannush SB, Yau CW, et al. Elevated intraocular pressure-induced interlamellar stromal keratitis. *Ophthalmology*. 2002; 109: 1929-1933

Caceres V: Post-LASIK iop measurements may help dect PISK. *EyeWorld*. March 2005; 72-73.

Pituitary Dysfunction and Chorioretinopathy (CPD Syndrome; Chorioretinopathy and Pituitary Dysfunction) 992

General: Race, characterized by chorioretinopathy, trichosis, and pituitary dysfunction.

Ocular: Long lashes; bushy eyebrows; severe early-onset chorioretinopathy.

Clinical: Growth retardation; sexual infantilism; hypothyroidism; mental retardation; low birth weight; cerebellar ataxia.

Delleman JW, van Wallbeek K. The syndrome of trichomegaly, tapetoretinal degeneration and growth disturbances. *Ophthalmologica* 1975; 171:313-315.

Erdem E, et al. Cerebellar ataxia associated with hypogonadotropic hypogonadism and chorioretinopathy: a poorly recognized associated. *Clin Neural Neurosurg* 1994; 96:86-91.

Judisch GF, et al. Chorioretinopathy and pituitary dysfunction: the CPD syndrome. *Arch Ophthalmol* 1981; 99: 253-256.

Pituitary Gigantism Syndrome (Gigantism Syndrome; Launois Syndrome) 993

General: Increased production of growth hormone due to hyperplasia of the eosinophilic cells and chromophobe adenoma of the anterior pituitary gland; onset age 8 to 10 years; pituitary adenomas represent 10% to 15% of intracranial neoplasms; 40% of nonfunctioning tumors are prolactinomas; growth hormone-secreting tumors are the next most common type.

Ocular: Field defects according to extent and situation of the pituitary adenoma; optic nerve atrophy (partial).

Clinical: Gigantism; enlarged skull with prominent chin and forehead; retarded skeletal growth; delayed puberty; muscle weakness; headache; perspiration; joint pain; mental retardation; pallor; smooth skin; scanty facial and body hair; small penis and testes; high-pitched voice; large limbs, hands, and feet.

Abboud CF, Laws ER. Diagnosis of pituitary tumors. *Endocrinol Metab Clin North Am* 1988; 17:241.

Hoyt CS, Billson FA. Buphthalmos in neurofibromatosis: is it an expression of regional gigantism? *J Pediatr Ophthalmol* 1977; 14:228-234.

Milunsky A, et al. Cerebral gigantism in childhood: a report of two cases and a review of the literature. *Pediatrics* 1967; 40:395.

Prezio JA, et al. Acromegalic gigantism: the buffalo giant. *Am J Med* 1961; 31:966.

Pruitt A. Brain tumors. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994.

Rush S, et al. Neuroophthalmic assessment of radiotherapy alone for pituitary microadenomas. Identification of prognostic factors. *J Neurosurg* 1990; 72:594.

Pituitary Necrosis Syndrome (Postpartum Hypopituitarism Syndrome; Sheehan Syndrome; Simmonds-Sheehan Syndrome) 994

General: Etiology unknown; vascular occlusion of one of the vessels supplying the anterior lobe of the pituitary during childbirth; characterized by various degrees of anterior and/or posterior pituitary dysfunction due to pituitary necrosis after obstetric shock or hemorrhage.

Ocular: Hypotrichosis of eyebrows; loss of eyelashes; dry and scaly skin of the lids; visual loss due to vascular insufficiency; uveal depigmentation.

Clinical: Reduced sweating with dry skin; listlessness and lethargy; stupor; myxedema; premature aging; cutaneous hypopigmentation; reduced and sparse axillary and pubic hair; genital atrophy; menstrual irregularity; amenorrhea; thyroiditis.

Hoyt WF. Transient bilateral blurring of vision: considerations of episodic ischemic symptom of vertebral-basilar insufficiency. *Arch Ophthalmol* 1963; 70:746.

Sasaki H, et al. Simultaneous occurrence of postpartum hypopituitarism (Sheehan syndrome) and transient resolving thyrotoxicosis due to postpartum painless thyroiditis. *South Med J* 1992; 85:660-662.

Sheehan HL. Atypical hypopituitarism. *Proc R Soc Med* 1961; 54:43.

Simmonds M. Uber Hypophysisschwund mit Todlichem Ausgang. *Dtsch Med Wochenschr* 1914; 40:322.

Pityriasis Rubra Pilaris (Kaposi Disease [2]; Devergie Disease; Hebra Disease; Tarral-Besnier Disease; Lichen Ruber; Lichen Ruber Acuminatus; Pityriasis Pilaris) 995

General: Abnormal keratinization of unknown etiology; hereditary and acquired forms have been described in the literature; hereditary form tends to be less severe and more limited in extent.

Ocular: Papules on bulbar conjunctiva; keratitis; ectropion; pannus; corneal ulceration.

Clinical: Cutaneous manifestations; erythema; follicular papules.

Beamer JE, et al. Pityriasis rubra pilaris. *Cutis* 1972; 10:419-442.

Gilgor RS, et al. Evaluation of 13-cis retinoic acid in lamellar ichthyosis, pityriasis rubra pilaris and Darier's disease. *Cutis* 1980; 25:380-381,385.

Harley RD. ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Plague (Bubonic Plague; Pneumonic Plague) 996

General: Infectious disease of animals (principally wild animals and rodents) that is transmitted through the bite of infected ectoparasites; causative agent is *Yersinia pestis.*

Ocular: Inflammatory infiltrate of lids; subconjunctival hemorrhages; chemosis; staphyloma of sclera; keratitis; corneal abscess; iridocyclitis; choroiditis.

Clinical: Abrupt onset; chills; fever; rapid, thready pulse; painful, enlarged lymph nodes in bubonic plaque; headache and productive cough in pneumonic plaque; meningitis; occasional patients infected with Y. *pestis* become septic and die with bacteremia without developing any detectable lymphadenitis (septicemic plague).

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Butler T. Yersinia species (including plague). In: Mandell GL, Douglas RG Jr, Benett JE, eds. *Principles and Practice of infectious diseases*, 3rd ed. New York: Churchill Livingstone, 1990: 1748-1756.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hull HF, et al. Septicemia plague in New Mexico. *J Infect Dis* 1987; 155:113-118.

Plasma Lecithin Deficiency (Cholesterol Acyltransferase Deficiency) 997

General: Autosomal recessive.

Ocular: Corneal stromal opacities comprising small gray dots in central and peripheral areas; retinal hemorrhages; disk protrusion; dilated veins.

Clinical: Storage of lipid materials in various tissues.

Bethell W, et al. Lecithin cholesterol acyltransferase deficiency. Light and electron microscopic findings from two corneas. *Can J Ophthalmol* 1975; 10:494-501.

Horven I, et al. Ocular manifestations in familial LCAT deficiency. *Birth Defects* 1976; 12:271-278.

Plateau Iris Syndrome 998

General: Rare; occurs in younger age group; presumably due in part to an anterior insertion of the iris; pupillary block is not a significant part of the mechanism leading to angle closure.

Ocular: Spontaneous or mydriasis-induced angle closure despite a patent iridectomy; anterior chamber is of normal depth axially and the iris plane is flat, but a peripheral roll of iris can close the angle either when the pupil dilates spontaneously or after mydriatic drugs are administered.

Clinical: Nausea; vomiting.

Ritch R, Tham CC, Lam DS: Long-term success of argon laser peripheral iridoplasty in the management of plateau iris syndrome. *Ophthalmology* 2004; 111: 104-108

Shaffer RM. Gonioscopy, ophthalmoscopy and perimetry. *Trans Am Acad Ophthalmol Otolaryngol* 1960; 64: 112-127.

Shields BM. *Textbook of glaucoma,* 3rd ed. Baltimore: Williams & Wilkins, 1992.

Wand M, et al. Plateau iris syndrome. *Trans Ophthalmol Otolaryngol* 1977; 83:122-129.

Plummer-Vinson Syndrome (Sideropenic Dysphagia Syndrome; Paterson-Brown-Kelly Syndrome; Waldenstrom-Kjellberg Syndrome) 999

General: Deficiency of vitamin B complex and iron; female; onset middle age; pathogenic mechanism may be related to a frameshift mutation in the human apolipoprotein mutation in the human apolipoprotein A-I gene.

Ocular: Reduced tear formation; pale conjunctiva; dry eyes; retinal hemorrhages; papilledema.

Clinical: Dysphagia for solid food with main difficulties originating in the upper portion of the esophagus; glossitis and gastritis; anemia; atrophy of mucous membranes; dystrophy of the fingernails (koilonychia); fatigue.

Chen TS, Chen PS. Rise and fall of the Plummer-Vinson syndrome. *J Gastroenterol Hepatol* 1994; 9:654-658.

Godtfredsen E. Relations between Sjogren's disease, Plummer Vinson syndrome and ariboflavinosis. *Acta Ophthalmol* 1947; 25:95.

Plummer HS. Diffuse dilation of the esophagus without anatomic stenosis (cardiospasm). A report of ninety-one cases. *J Am Med Assoc* 1912; 58:2013.

Vinson PP. A case of cardiospasm with dilatation and angulation of the esophagus. *Med Clin North Am* 1919; 3:623.

Pneumococcal Infections *(Streptococcus Pneumoniae* Infections) 1000

General: Gram-positive diplococcus *Streptococcus pneumoniae;* some strains are encapsulated while others are not; ocular infections usually are caused by the encapsulated strains; conjunctivitis and corneal scarring produced in an animal model have been attributed to a hemolytic cytolytic exopeptidase.

Ocular: Hypopyon; conjunctivitis; keratitis; corneal ulcer; endophthalmitis; dacryocystitis; uveitis; orbital cellulitis; secondary glaucoma; ophthalmia neonatorum.

Clinical: Upper respiratory infection; chills; sharp pain in hemithorax; cough with sputum production; fever; headache; gastrointestinal symptoms.

Baum JL. Antibiotic use in *Ophthalmology*. In: Tasman W, Jaeger E, eds. *Duane's Clinical Ophthalmology, vol. 4.* Philadelphia: Lippincott, 1990: 1-26.

Burd EM. Bacterial keratitis and conjunctivitis. In: Smolin G, Thoft RA, eds. *The Cornea.* Boston: Little, Brown and Company, 1994.

Finland M. And the walls come tumbling down. More antibiotic resistance, and now the pneumococcus. *N Engl J Med* 1978; 299:770-771.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Johnson MK., Allen JH. The role of cytolysin in pneumococcal ocular infection. *Am J Ophthalmol* 1975; 80:518.

Jones DB. Early diagnosis and therapy of bacterial corneal ulcers. *Int Ophthalmol Clin* 1973; 13:1-29.

Okumoto M, Smolin G. Pneumococcal infections of the eye. *Am J Ophthalmol* 1974; 77:346.

Poison Ivy Dermatitis (Rhus Dermatitis; Poison Oak Dermatitis; Poison Sumac Dermatitis) 1001

General: Direct contact or airborne contact from burning of plant.

Ocular: Keratitis; chemosis; blepharospasm; pustules of eyelids.

Clinical: Dermatitis; linear lesions; erythema; pruritus.

Duke-Elder S, MacFaul PA, eds. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Grant WM. *Toxicology of the eye,* 2nd ed. Springfield, IL: Charles C. Thomas, 1974:845.

Rudner EJ, et al. The frequency of contact sensitivity in North America. *Contact Dermatitis* 1975; 1:277.

Poland-Möbius Syndrome 1002

General: Rare congenital disorder that is a combination of Möbius syndrome and Poland anomaly; has been proposed that Möbius syndrome, the Poland anomaly, and the Klippel-Feil defect all result from a transient interruption during the sixth week of gestation in the development of the subclavian artery and its branches.

Ocular: Bilateral sixth nerve paralysis; no movement of upper lid in horizontal gaze; no globe retraction during adduction; vertical nystagmus; diplopia; chronic keratitis; corneal ulcer.

Clinical: Paresis of the sixth and seventh cranial nerves; bilateral absence of pectoralis muscle and ipsilateral hand and digit anomalies; chronic drooling; speech difficulties; masklike facial expression.

Bavinck JN, Weaver DD. Subclavian artery disruption sequence: hypothesis of a vascular etiology from Poland, Klippel-Feil and Mobius anomalies. *Am J Med Genet* 1986; 23:903-918.

Gadoth N, et al. Mobius syndrome and limb abnormalities. *J Pediatr Ophthalmol* 1977; 14:134-138.

Hopper KD, et al. Poland-Mobius syndrome. *South Med J* 1985; 78:523-527.

Wishnick MM, et al. Mobius syndrome and limb abnormalities with dominant inheritance. *Ophthalmic Pediatr Gen* 1983; 2:277-281.

Poliomyelitis (Infantile Paralysis) 1003

General: Acute viral infection characterized by varying degrees of neuronal injury, with special localization in the anterior horns and motor nuclei of the brainstem.

Ocular: Diplopia; nystagmus; paralysis of third, fourth, and sixth nerves; paresis of seventh nerve; papilledema; visual agnosia; Homer syndrome; pupillary paralysis; optic neuritis; ophthalmoparesis; transient visual loss; internuclear ophthalmoplegia; papillary disturbances, spasm of near reflex.

Clinical: Flaccid paralysis of many muscle groups; death from asphyxia and involvement of vital centers in the brainstem.

Bhabha SK, Bharucha NE, Bharucha EP. Viral infections. In: Dradley WG, et al., eds. *Neurology in Clinical practice, vol. II,* 2nded. Boston: Butterworth-Heinemann, 1995:1272-1275.

Duke-Elder S. *System of Ophthalmology, vol. XII.* St. Louis: CV Mosby, 1976:720.

Harley RD, ed. *Pediatric Ophthalmology,* 4th ed. Philadelphia: WB Saunders, 1998.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 2, 4th ed. Baltimore: Williams & Wilkins, 1995.

Jubelt B, Miller JR. Viral infections. In: Rowland LP, ed. *Merritt's Textbook of Neurology,* 9th ed. Baltimore: Williams & Wilkins, 1995:145-148.

Polycythemia Vera (Erythema; Erythrocytosis Megalosplenica; Myelopathic Polycythemia; Vaquez Disease; Vaquez-Osler Syndrome; Cryptogenic Polycythemia; Polycythemia Rubra; Splenomegalic Polycythemia) 1004

General: Increased number of red blood cells; myeloproliferative disorder.

Ocular: Conjunctival vascular engorgement; dilated tortuous retinal veins; retinal hemorrhages; optic disk edema; central retinal vein occlusion; visual field defects; visual hallucinations; diplopia.

Clinical: Elevated red blood cells; systemic vascular congestion; leukocytosis; thrombocytosis; central nervous system involvement; splenomegaly; hepatomegaly; bleeding diathesis; gingival/mucosal bleeding; ecchymosis; epistaxis; neurologic abnormal dizziness; vertigo; ataxia.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Garfinkel D, et al. Oculomotor nerve paresis: presenting sign of acute myeloblastic leukemia in a patient with polycythemia vera. Haematologica 1980; 65:769-772.

Patterson WP, et al. Hyperviscosity syndrome and coagulopatbies. *Semin Oncol* 1990; 17:210-2l6.

Wells R. Syndromes of hyperviscosity. *N Engl J Med* 1970; 283:183-186.

Polymyalgia Rheumatica 1005

General: Affects older patients; usually Caucasian women; relationship between polymyalgia and temporal arteritis remains uncertain; in polymyalgia rheumatic a alone there is no arteritis per se, and this syndrome is not associated with blindness or other neurologic or cardiovascular sequelae; many patients with temporal arteritis have polymyalgia rheumatic a as part of their symptoms or as the presenting symptom.

Ocular: Amaurosis fugax; acute unilateral or bilateral visual loss; ischemic optic neuritis; optic atrophy; papilledema; retinal hemorrhages; cotton-wool spots; central retinal artery occlusion; palsy of extraocular muscles.

Clinical: Pain and stiffness in the neck, shoulders, and hips; rapid erythrocyte sedimentation rate; prompt response to corticosteroids; weakness.

Bengtsson BA, Malmvall BE. The epidemiology of giant cell arteritis including temporal arteritis and polymyalgia rheumatica: incidences of different *Clinical* presentations and eye complications. *Arthritis Rheum* 1981; 24: 899-904.

Chuang T, et al. Polymyalgia rheumatica. A l0-year epidemiological and *Clinical* study. *Ann Intern Med* 1982; 97:672.

Cohen MD, Ginsburg WW. Polymyalgia rheumatica. *Rheum Dis Clin North Am* 1990; 16:325.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Hart FD. Visual complications of polymyalgia rheumatic a (polymyalgia arteritica). *Practitioner* 1975; 215:763-766.

Jacobs DS, Foster SC. Temporal arteritis. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994.

Jones JG, Hazleman BL. Prognosis and management of polymyalgia rheumatica. *Ann Rheum Dis* 1981; 40:1-5.

Parsons C. Sudden blindness in polymyalgia arteritica. *Br Med J* 1977; 1:1536-1537.

Polymyositis-Dermatomyositis (Dermatomucomyositis; Neuromyositis; Polymyositis Gregarina; Wagner-Unverricht Syndrome) 1006

General: Autoimmune disease; etiology unknown; variable symptoms according to prevalence of skin (dermatomyositis) or muscular (polymyositis) involvement; association with neoplastic disease; prevalent in females; onset in childhood before age 10 years and in adults predominant in fourth to sixth decades; both children and adults are affected by this disease, but the prognosis is better for the childhood forms of disease.

Ocular: Violescent discoloration of eyelids; conjunctivitis; episcleritis; anterior uveitis; nystagmus; exophthalmos; cotton-wool spots; retinal edema; retinal hemorrhage; venous engorgement.

Clinical: Erythema involving face, forearms, and upper back; muscle weakness, especially shoulder and pelvic girdles; dysphagia; respiratory difficulty; malaise; fever; tachycardia.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. Clinical *Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Kissel JT, et al. Microvascular deposition of complement membrane attack complex in dermatomyositis. *N Engl J Med* 1986; 314:329.

Messner RP. Dermatomyositis and polymyositis. In: Wyngaarden JB, Smith LH, eds. *Cecil textbook of medicine*. 16th ed. Philadelphia: WB Saunders, 1982:1872-1873.

Opremcak EM. Collagen disorders: retinal manifestations of collagen vascular diseases. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology*, vol. V. Philadelphia: WB Saunders, 1994:989.

Pompe Disease (Generalized Glycogenosis) 1007

General: Absence of acid maltase; type II glycogen storage disease with decreased acid maltase resulting in elevated levels of lysosomal glycogen; inheritance is autosomal recessive.

Ocular: Cortical blindness.

Clinical: Anorexia; retardation of growth; cyanosis; dyspnea; convulsions; death; enlarged tongue; large heart; hypotonicity.

Goebel HH, et al. Ultrastructural observations on the retina in type II glycogenosis (Pompe's disease). *Ophthalmologica* 1978; 176:61-68.

Libert J, et al. Ocular ultrastructural study in a fetus with type II glycogenosis. *Br J Ophthalmol* I977; 61:476-482.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Pokorny KS, et al. Ultrastructure of the eye in fetal type II glycogenosis (Pompe's disease). *Invest Ophthalmol* 1982; 22:25-31.

Verity MA. Infantile Pompe's disease, lipid storage, and partial carnitine deficiency. *Muscle Nerve* 1991; 14:435-440.

Popliteal Pterygium Syndrome (PPS) 1008

General: Rare autosomal dominant disorder similar to Van der Woude syndrome (the two are possibly allelic).

Ocular: None.

Clinical: Popliteal webbing; cleft lip; cleft palate, lower lip pits; syndactyly; genital and nail abnormalities; severe flexure contract at the knee; equinus foot; velar pterygium.

Herold HZ, Shmueli G, Baruchin AM. Popliteal pterygium syndrome. *Clin Orthop Rel Res* 1986; 209:194-197.

Koch H, Grzonka M, Koch J. Popliteal pterygium syndrome with special consideration of the cleft malformation: case report. *Cleft Palate Craniofac J* 1992; 29:80-84.

Lees MM, et al. Popliteal pterygium syndrome: a clinical study of three families and report of linkage to the Van der Woude syndrome locus on Iq32. *J Med Genet* 1999; 36:888-892.

Porokeratosis 1009

General: Autosomal dominant; prominent in males; onset in early childhood; characterized by scaly papules that enlarge to form gyrate lesions.

Ocular: Conjunctivitis; lesions of cornea.

Clinical: Squamous cell carcinoma within lesions; lesions may occur on oral mucosa, palms, or soles.

Baden HP, et al. Treatment of ichthyosis with isotretinoin. *J Am Acad Dermatol* 1982; 6:716.

Goldsmith LA, et al. The structural proteins of harlequin fetus: stratum corneum. *J Invest Dermatol* 1973; 61:25.

Porphyria Cutanea Tarda 1010

General: Disorder of porphyria metabolism; highest incidence in Bantu population; both sexes affected; onset between ages 40 and 60 years; insidious onset; autosomal dominant; light-sensitive dermatitis in later adult life; associated with excretion of large amounts of uroporphyrin in urine.

Ocular: Synophrys; keratitis; palsies of third and seventh cranial nerves; scleromalacia perforans; optic atrophy; retinal hemorrhages and cotton-wool spots; macular edema; pinguecula; pterygium; brownish pigmentation in conjunctiva and lid margin.

Clinical: Cutaneous manifestations are solar hypersensitivity, vesiculobullous lesions, ulcerations, severe scarring, and hypertrichosis; erythrodontia.

Chumbley LC. Scleral involvement in symptomatic porphyria. *Am J Ophthalmol* 1977; 84:729.

Hammer H, Korom I. Photodamage of the conjunctiva in patients with porphyria cutanea tarda. *Br J Ophthalmol* 1992; 76:592-593.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Sober AJ, et al. Cicatricial ectropion and lacrimal obstruction associated with the sclerodermoid variant of porphyria cutanea tarda. *Am J Ophthalmol* 198l; 9l: 396-400.

Struck HG, et al. Klinische Untersuchungen zur Augenbeteiligung bei Porphyria cutanea tarda. *Klin Monatsbl Augenheilkd* 1991; 198:258-263.

Willerson D Jr, et al. Familial porphyria cutanea tarda in a patient with retinitis pigmentosa. *Ann Ophthalmol* 1979; 11:409-411.

Posner-Schlossman Syndrome (Glaucomatocyclitic Crisis) 1011

General: High intraocular tension lasting from hours to several weeks and recurring at varying frequencies; low-grade, intermittent, nongranulomatous inflammation; in one series of patients, HLA-BW54 was present in 41% of patients.

Ocular: Slight blurring of vision and colored halos during episodes of high intraocular tension; high intraocular pressure (unilateral); glaucomatocyclitic crisis (benign and usually unilateral); enlarged pupil; anisocoria; absence of ciliary or conjunctival injection; only trace of aqueous flare; no posterior synechiae; chamber angle open; heterochromia iridis; keratitic precipitates may be present.

Clinical: Allergy; associated with gastrointestinal disease (peptic ulcers).

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hirose S, et al. HLA-BW54 and glaucomatocyclitic crisis. *Arch Ophthalmol* 1985; 103:l837.

Posner A, Schlossman A. Syndrome of unilateral recurrent attacks of glaucoma with cyclitic symptoms. *Arch Ophthalmol 1948; 39:5l7.*

Theodore FH. Observations on glaucomatocyclitic crisis (Posner Schlossman syndrome). *Br J Ophthalmol* 1952; 36:207.

Posterior Iris Chafing Syndrome 1012

General: Complication following intraocular lens implantation attributed to sulcus-fixed posterior chamber lenses, decentration of the lens, traumatic insertion, movement of the lens during dilation and constriction of the pupil, or difficulty with positioning of the lens.

Ocular: Iris transillumination defects; recurrent microhyphemas; pigment dispersion glaucoma; pigment deposition in trabecular meshwork; iris pigment atrophy.

Clinical: None.

Jaffe NS. Current concepts in posterior chamber lens technology. *Am Intraocul Implant Soc J* 1985; 11:456-464.

Johnson SH, et al. Iris transillumination defect and microhyphema syndrome. *Am Intraocul Implant Soc J* 1984; 10: 425-428.

Smiddy WE, et al. Surgical management of dislocated intraocular lenses. *J Cataract Refract Surg* 1995; 21:64-69.

Smith SG, Lindstrom RL. Malpositioned posterior chamber lenses: etiology, prevention, and management. *Am Intraocul Implant Soc J* 1985; 11:584-591.

Woodhams JT, Lester JC. Pigmentary dispersion glaucoma secondary to posterior chamber intraocular lenses. *Ann Ophthalmol* 1984; 16:852-855.

Posterior Uveal Bleeding Syndrome 1013

General: Most commonly affects middle-aged black females; also observed in white or Asian people

Ocular: Miltiple recurrent pigment epithelial detachments; posterior uveal bleeding; polyps in the peripapillary macula and extramcular areas

Clinical: Hypertension; detected with indocyanine green angiograph

Ahuja RM, Stanga PE, VingerlingJR, Polypoidal choroidal vasculopathy in exudates and haemorrhagic pigment epithelial detachments. *Br J Ophthalmol* 2000; 84: 479-484.

Stanga PE, Lim, JI, Hamilton P: Indocyanine green angiography in chorioretinal diseases: indications and interpretation. *Ophthalmology* 2003; 110: 15-24

Posthypoxic Encephalopathy Syndrome (Posthypoxic Syndrome; Parietooccipital Syndrome) 1014

General: Hypoxia secondary to carbon monoxide poisoning, high altitude, complication during anesthesia, hypoglycemia, or cardiac failure, causing widespread demyelination in the parietal lobes, including the optic radiations.

Ocular: Nystagmus; nuclear ophthalmoplegia; visual hallucinations; partial cerebral blindness (predominant defects in the sphere of psychic elaboration rather than in primary visual perception); complete cortical blindness; central scotomata; pupillary paresis; retinal atrophy; optic atrophy.

Clinical: Confusion; irritability and agitation; alexia; disorientation (mainly spatial); muscle spasm.

Freedman A, Sevel D. The cerebro-ocular effects of carbon dioxide poisoning. *Arch Ophthalmol* 1966; 76:59.

Hoyt WF, Walsh FB. Cortical blindness with partial recovery following acute cerebral anoxia from cardiac arrest. *Arch Ophthalmol* 1958;60:1061.

Wechsler IS. Partial cortical blindness with preservation of color vision. Report of a case following asphyxia (carbon monoxide poisoning?): a consideration of the question of color vision and its cortical localization. *Arch Ophthalmol* 1933; 9:957.

Poststreptococal Uveitis Syndrome 1015

General: Associated with bilateral nongranulomatous ocular inflammation; seen in individuals over 40 years of age.

Ocular: Vitritis; focal retinitis; optic disk swelling; multifocal choroiditis

Cokington CD, Han DP: Bilateral nongranulomatous uveitis and a post streptoccal syndrome. *Am J Ophthalmol* 1991; 112: 595-596

Rehman SU, Anand S, Reddy A, et al. Poststreptococcal syndrome uveitis. *Ophthalmology* 2006; 113: 701-706

Postvaccinial Ocular Syndrome 1016

General: Immediate contamination and inoculation or as a delayed reaction; incubation time 3 days.

Ocular: Orbital cellulitis; pustules and vesicles on the eyelids (vesicles appear grayish with purulent discharge); lids swollen and red with swelling of preauricular and postauricular lymph nodes; ophthalmoplegia; mild anterior uveitis; conjunctival and corneal vesicles similar to the lid lesions; corneal marginal ulcers (eventually may lead to corneal perforation and loss of the eye); chorioretinitis (occasionally); central serous retinopathy; central retinal vein thrombosis; panophthalmitis; neuritis; ocular vaccinia may mimic signs of herpes simplex virus, varicella-zoster virus, and acanthamoeba keratitis.

Clinical: Postvaccinial encephalitis; severe local purulent skin reaction; severe headache; fever; malaise.

Jones BR, Al-Hussaini MK. Therapeutic considerations in ocular vaccinia. *Trans Ophthalmol Soc UK* 1963; 83:613.

Lee SF, et al. Vaccinia keratouveitis manifesting as a masquerade syndrome. *Am J Ophthalmol* 1994; 117:480-487.

Rosen E. A postvaccinial syndrome. *Am J Ophthalmol* 1948; 31:1443-1453.

Potter Syndrome (Renal Agenesis Syndrome; Renofacial Syndrome) 1017

General: Unknown etiology; may be severe form of the trisomy 18 syndrome; results from prolonged oligohydramnios of any cause.

Ocular: Hypertelorism; pronounced epicanthal folds extending down the cheeks; antimongoloid slant of palpebral fissure.

Clinical: Flat bridge of the nose; low-set ears; facial deformities; micrognathia; pulmonary hypoplasia; cystic dysplasia of kidney to agenesis; oligohydramnios; clubbing of hands and feet; spina bifida; prominent infracanthal folds; flattened beaked nose; creased skin; positional deformities of the limbs.

Franceschetti A. Craniofacial dysostosis. In: *Symposium on surgical and medical management of congenital anomalies of the eye*. St. Louis: CV Mosby, 1968:77.

Goldbloom RE, et al. Hereditary renal disease associated with nerve deafness and ocular lesions. *Pediatrics* 1957; 20:241

Kohart EC. End-stage renal disease. In: Oski, F, ed. *Principles and Practice of Pediatrics*, 2nd ed. Philadelphia: Lippincott, 1994:1782.

Passarge E, Southerland JM. Potter's syndrome: chromosome analysis of three cases with Potter's syndrome or related syndromes. *Am J Dis Child* 1965; 109:80.

Potter EL. Bilateral renal agenesis. *J Pediatr* 1946; 29:68.

Prader-Willi Syndrome (Prader-Labhart-Willi-Fanconi Syndrome; H_20 Syndrome; Hypogenital Dystrophy with Diabetic Tendency; Hypotonia-Hypomentia-Hypogonadism-Obesity [HHHO] Syndrome; Royer Syndrome) 1018

General: Etiology unknown; dominant inheritance is suspected; predominantly seen in males; Royer syndrome is Prader-Willi syndrome associated with diabetes mellitus; nongenetic condition characterized by infantile hypotonia, hypogonadism, and obesity.

Ocular: Strabismus; ocular hypertelorism; myopia; exotropia; glaucoma; cataracts; congenital ocular fibrosis syndrome; diabetic retinopathy.

Clinical: Mental retardation; short stature; muscular hypotonia; small hands and feet; obesity; cryptorchidism; hypogonadism; dental caries; clinodactyly; partial syndactyly of toes and fingers.

Libov AJ, Maino DM. Prader-Willi syndrome. *J Am Optom Assoc* 1994; 65:355-359.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Prader W, et al. Eli Syndrome von Adipositas, Kleinwuchs, Kryptorchismus and Oligophrenia nach myatonieartigem Zustand im Neuge-borenenalter. *Schweiz Med Wochenschr* 1956; 86:1260-1261.

Pregnancy 1019

General: Pregnancy results in hormonal changes that produce ocular effects; symptoms resolve at end of pregnancy term.

Ocular: Myopia; visual field defects; corneal edema; acute ischemic optic neuropathy; central serous retinopathy; glaucoma; ptosis; diabetic retinopathy; Krukenberg spindles; transient blindness; serous retinal detachment; retinal artery occlusion; retinal vein occlusion; disseminated intravascular coagulopathy; uveal melanoma.

Clinical: Nausea; headaches; hypertension; benign intracranial hypertension; preeclampsia; toxemia; fluid retention.

Ayaki M, et al. Postpartum central retinal artery occlusion simulating Purtscher's retinopathy. *Ophthalmologica* 1995; 209:37-39.

Carlson DC. Ocular manifestations of pregnancy. *J Am Optom Assoc* 1988; 59:49.

Sunness JS. Pregnancy and retinal disease. In: Ryan S, ed. *Retina.* St. Louis: Mosby 1994:1393.

Ward SC, et al. Pregnancy and acute optic disc edema of juvenile onset diabetes. *Obstet Gynecol* 1984; 64:816-818.

Primary Antiphospholipid Syndrome (PAS) 1020

General: Thrombophilic disorder characterized by the presence of autoantibodies and CNS involvement, venous thrombosis, and ocular manifestations.

Ocular: Visual disturbances (visual obscurations, amaurosis fugax, diplopia, homonymous field defects, and scintillating scotoma due to migraine); two forms of retinopathy: cotton-wool spots with or without hemorrhages and vasoocclusive ocular disease.

Clinical: Many patients will experience CNS involvement in the form of either a stroke or a transient ischemic attack; visual disturbances are largely due to CNS rather than ocular ischemia.

Bauer KA. The hypercoagulable state. In: Beutler E, Lichtman MA, Coller BS, et al., eds. *Williams' hematology*, 5th ed. New York: McGraw-Hill, 1995:1531-1550.

Brenner B, et al. Coexistence of familial antiphospholipid syndrome and factor V Leiden: impact on thrombotic diathesis. *Br J Haematol* 1996; 94:167.

Dahlback B. Resistance to activated protein C caused by the factor V R 506Q mutation is a common risk factor for venous thrombosis. *Thromb Haemost* 1997; 78:483-488.

Gelfand YA, Dori D. Visual disturbances and pathologic ocular findings in primary antiphospholipid syndrome. *Ophthalmology* 1999; 106:1537-1540.

Glacet-Bernard A, et al. Antiphospholipid antibodies in retinal vascular occlusions: a prospective study of 75 patients. *Arch Ophthalmol* 1994; 112:790-795.

Zoller B, et al. Resistance to activated protein C as an additional genetic risk factor in hereditary deficiency of protein S. *Blood* 1995; 85:3518-3523.

Progressive External Ophthalmoplegia and Scoliosis 1021

General: Rare; isolated muscle dystrophic involvement of extraocular muscles; onset in childhood or early adulthood; slowly progressive.

Ocular: Horizontal gaze paralysis; pendular nystagmus; ptosis; orbicularis oculi weakness.

Clinical: Scoliosis; facial myokymia; contracture of facial muscles.

Abyholm FE, Skolleborg KC. Aesthetic treatment of progressive hemifacial atrophy (Romberg's disease): use of a pedicled platysma muscle flap. *Plast Reconstr Surg* 1995; 96:71-77.

Anderl H. Free vascularized groin fat flap m hypoplasia and hemiatrophy of the face (a three year observation). *J Maxillofac Surg* 1979; 7:327-332.

Dehaene I, et al. Locked-in syndrome with bilateral horizontal pontine gaze paralysis and nuclear oculomotor nerve paralysis. *J Neurol* 1985; 232:366-367.

Dintiman BJ, et al. Parry-Romberg syndrome in association with contralateral Poland syndrome. *J Am Acad Dermatol* 1990; 22[2 Part 2]:371-373.

Ebersbach G, et al. Hemimasticatory spasm in hemifacial atrophy: diagnostic and therapeutic aspects in two patients. *Mov Disord* 1995; 10:504-507.

Jones NF. The contribution of microsurgical reconstruction to craniofacial surgery. *World J Surg* 1989; 13:454-464.

Longaker MT, Siebert JW. Microvascular free-flap correction of severe hemifacial atrophy. *Plast Reconstr Surg* 1995; 96:800-809.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Moore MH, et al. Progressive hemifacial atrophy (Romberg's disease): skeletal involvement and treatment. *Br J Plast Surg* 1993; 46:39-44.

Nerobyeyev AL, Brusova LA, Malakhovskava VI. Surgical treatment of progressive facial hemiatrophy. *Acta Chir Plast* 1990; 32:65-73.

Roddi R, et al. Clinical evaluation of techniques used in the surgical treatment of progressive hemifacial atrophy. *J Craniomaxillofac Surg* 1994; 22:23-32.

Progressive Foveal Dystrophy (Central Retinal Pigment Epithelial Dystrophy) 1022

General: Autosomal dominant; onset late in the first decade of life.

Ocular: Progressive foveal dystrophy; pigmentary changes and drusen of the macula; normal electroretinogram; subnormal electrooculogram.

Clinical: Generalized aminoaciduria; increased glycine levels.

Deutman AF. Macular dystrophies. In: Ryan S, ed. *Retina.* St. Louis: Mosby, 1994:214-1215.

Klein R, Bresnick G. An inherited central retinal pigment epithelial dystrophy. *Birth Defects* 1982; 189:281-296.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Progressive Inherited Retinal Arteriolar Tortuosity with Retinal Hemorrhages (Familial Retinal Arteriolar Tortuosity with Retinal Hemorrhages) 1023

General: Autosomal dominant; tortuosity increases most dramatically during adolescence and progresses throughout adulthood.

Ocular: Tortuosity of the retinal arterioles; spontaneous retinal hemorrhages.

Clinical: None.

Kayazawa F, Machida T. Retinal arteriolar tortuosity with macular hemorrhage. *Ann Ophthalmol* 1983; 15:42-43.

Wells CG, Kalina RE. Progressive inherited retinal arteriolar tortuosity with spontaneous retinal hemorrhages. *Ophthalmology* 1985; 92:1015-1024.

Progressive Intracranial Arterial Occlusion Syndrome (Taveras Syndrome) 1024

General: Children and young adults; caused by previous trauma and atheroma; occlusive endarteritis; predilection for the circle of Willis; occurs mostly in Japanese and Caucasians.

Ocular: Unilateral ptosis; defective optokinetic nystagmus; visual agnosia; amaurosis fugax; abnormally high ophthalmodynamometric readings may be the first correct diagnostic sign even before arteriography reveals the vascular pathologic condition.

Clinical: Progressive intracranial arterial occlusion with both internal carotid arteries involved; memory loss; muteness; localized numbness; crying spells; catatonic states and episodes, staring; seizures.

Taveras J. Multiple progressive intracranial arterial occlusions: a syndrome in children and young adults. *Am J Roentgenol* 1969; 106:235.

Zappia RJ, et al. Progressive intracranial arterial occlusion syndrome. *Arch Ophthalmol* 1971; 86:455.

Progressive Systemic Sclerosis (Scleroderma; Systemic Scleroderma) 1025

General: Chronic connective tissue disease of unknown etiology; chronic and usually progressive disorder; typical onset is in third to fifth decade; ratio of women to men is 4:l; primary sites of pathology are the arterioles and capillaries of affected organs.

Ocular: Marginal corneal ulcers; shortened fornices of the conjunctiva; ptosis; cotton-wool patches of retina; papilledema; retinal hemorrhages; cicatrization of conjunctiva and cornea; blepharitis; blepharospasm; thready, tenacious yellow-white conjunctival discharge; hypertrophy of lacrimal gland; episcleritis; ocular myositis; Sjögren syndrome; uveitis; vitreal haze; keratitis sicca; decreased corneal sensation; iritis; ischemic choroidopathy; iris sectorial atrophy; blepharophimosis; heterochromia; keratoconus; central retinal vein occlusion; branch retinal vein occlusion.

Clinical: Vascular insufficiency; Raynaud phenomenon; malaise; weight loss; stiffness; fever; polyarticular arthritis; diffuse edema of the hands; calcinosis; esophageal involvement; sclerodactyly; telangiectasis; esophageal stricture; renal failure; diffuse interstitial fibrosis.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

David R, Irvy M. Focal chorioretinitis and iridocyclitis associated with scleroderma. *Ann Ophthalmol* 1976; 8: 199-202.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Reddy CV, Foster CS. Scleroderma. In: Albert DM, Jakobiec FA, eds. Principles *and Practice* of *Ophthalmology, vol. V*. Philadelphia: WB Saunders, 1994:2919-2924.

Saari KM et al. Bilateral central retinal vein occlusion in a patient with scleroderma. *Ophthalmologica* 1981; 182: 7-12.

Protein C Deficiency 1026

General: Autosomal recessive; congenital; heterozygous individuals predisposed to recurrent venous thrombosis; homozygous individuals may develop widespread thrombotic complications in neonatal period; reduced clotting ability; neonatal protein C deficiency also may be acquired and transient in ill preterm babies.

Ocular: Vitreous opacities; vitreous hemorrhages; retinal detachment; cataract; shallow anterior chamber; leukocoria; prominent iris vessels; iris synechiae to lens; bilateral involvement; retinal hemorrhages; cavernous sinus thrombosis.

Clinical: Hematomas; epistaxis; prolonged bleeding; hydrocephalus; *Candida* sepsis; subarachnoid hemorrhage; pulmonary embolism; thrombotic hemorrhagic gastrointestinal and genitourinary mucosal infarcts.

Cassels-Brown A, et al. Ophthalmic manifestations of neonatal protein C deficiency. *Br J Ophthalmol* 1994; 78: 486-487.

Pulido JS, et al. Protein C deficiency associated with vitreous hemorrhage in a neonate. *Am J Ophthalmol* 1987; 104: 546-547.

Seligsohn U, et al. Homozygous protein C deficiency manifested by massive venous thrombosis in the newborn. *N Engl J Med* 1984; 310:599.

Proteus Infections 1027

General: Gram-negative bacilli found in water, soil, and decaying organic substances.

Ocular: Conjunctivitis; keratitis; corneal ulcers; endophthalmitis; panophthalmitis; dacryocystitis; gangrene of eyelid; uveitis; hypopyon; paralysis of seventh nerve.

Clinical: Cutaneous infection after surgery; usually occurs as a secondary infection of the skin, ears, mastoid sinuses, eyes, peritoneal cavity, bone, urinary tract, meninges, lung, or bloodstream; meningitis; intracranial subdural and epidural empyema; brain abscess; intracranial septic thrombophlebitis affecting cavernous/lateral sinuses.

Burke JP, et al. Proteus syndrome: ocular complications. *J Pediatr Ophthalmol Strabismus* 1988; 25:99.

Foulks GM. Bacterial infections of the conjunctiva and cornea. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology. vol. 1.* Philadelphia: WB Saunders, 1994: 167.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Kim SJ, et al. Ventriculoperitoneal shunt infections with gram-negative bacteria. *Neurosurgery* 1993; 33:858-862.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5. *part 1* 4th ed. Baltimore: Williams & Wilkins, 1995.

Okumoto M, et al. Proteus species isolated from human eyes. *Am J Ophthalmol* 1976; 81:495-501.

Smolin G. Proteus endophthalmitis. *Arch Ophthalmol* 1974; 91:419-420.

Proteus Syndrome 1028

General: A harmarteo neoplastic disorder with variable clinical manifestations

Ocular: Myopia; band keratopathy; cataract; vitreous hemorrhage; chorioretinal mass; serous retinal detachment

Clinical: Thickening of the bones of the external auditory meatus and cranial fossa; enlargement of the left internal auditory meatus; deformities of the feet and toes.

Burke JP, Bowell R, Doherty N: Proteus syndrome; ocular complications. *J Pediatr Ophthalmol Strabismus* 1988; 25: 99-102

Sheard R, Pope F, Snead M: A novel ophthalmic presentation of the proteus syndrome. *Ophthalmology* 2002; 109: 1192-1195

Proximal and Distal Click Syndrome of the Superior Oblique Tendon (Simulated Superior Oblique Tendon Syndrome) 1029

General: Produced by quick head movements; caused by adhesions (secondary to trauma and inflammation) or frontal sinus surgery; proximal click adhesions in front to trochlea; distal click adhesions behind trochlea; associated with Brown syndrome.

Ocular: Decreased elevation in adduction; downshoot of the affected eye on adduction; overaction of the tethered inferior oblique after cutting superior oblique tendon; widening of palpebral fissure on adduction; diplopia.

Clinical: Rheumatoid arthritis.

Brown HW. True and simulated superior oblique tendon sheath syndromes. *Doc Ophthalmol* 1973; 34:123-136.

Pittke EC. The proximal and distal click syndrome of the superior oblique tendon. *Graefes Arch Clin Exp Ophthalmol* 1987; 225:28-32.

Pruritus 1030

General: Unpleasant sensation perceived in the skin that elicits the response of scratching; causes may be physiologic or pathologic.

Ocular: Vesicles, urticaria, and eczematization of lid.

Clinical: Urticaria; hives; macular erythema; vesicles; eczematization of lid; skin excoriations; secondary infection; lichenification.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Sandvig K, Westerberg P. Ocular findings in psoriatics. *Acta Ophthalmol* 1955; 33:463-467.

Summerfield JA. Pain, itch and endorphins. *Br J Dermatol* 1981; 105: 725-726.

Pseudoexfoliation Syndrome 1031

General: Prevalent over age 70 years; rare before age 40 years; unilateral involvement in 40% to 50% of cases; asymmetry of severity in bilateral cases; most common in Caucasians, especially from Iceland and Scandinavian countries; pseudoexfoliation fibers were identified in autopsy tissue specimens of skin, heart, lungs, liver, and cerebral meninges; consistently associated with connective tissue components, i.e., fibroblasts, collagen and elastic fibers, myocardial tissue and heart muscle cell.

Ocular: Gray or white fluffy material deposited in particles, flakes, or sheets on anterior surface of iris, ciliary body, posterior surface of cornea, pupillary margin, lens, and trabecular meshwork; increased pigmentation of trabecular meshwork; zonular dialysis; displaced or dislocated lens; anterior chamber depth asymmetry; preoperative phacodonesis; glaucoma; cataract.

Clinical: None.

Mizuno K, Muroi S. Cycloscopy of pseudo exfoliation. *Am J Ophthalmol* 1979; 87:513-518.

Schlotzer-Schrehardt U, Maumann GO.A histopathologic study of zonular instability in pseudoexfoliation syndrome. *Am J Ophthalmol* 1994; 118:730-743.

Schlotzer-Schrehardt UM, et al. Pseudoexfoliation syndrome. Ocular manifestation of a systemic disorder? *Arch Ophthalmol* 1992; 110: 1752-1756.

Skuta GL, et al. Zonular dialysis during extracapsular cataract extraction in pseudoexfoliation syndrome. *Arch Ophthalmol* 1987; 105:632-634.

Suzuki R, Kurimoto S. Intraocular muscle function in pseudo exfoliation syndrome. *Ophthalmologica* 1992; 204: 192-198.

Pseudo-Foster Kennedy Syndrome 1032

General: Mimic disease of Foster Kennedy syndrome consisting of ischemic optic neuropathy and glioma.

Ocular: Optic disk pallor; visual field defects.

Clinical: Slow, progressive, decreased vision.

Lepore FE, Yarian DL. A mimic of "the exact diagnostic sign of Foster Kennedy." *Ann Ophthalmol* 1985; 17: 411-412.

Limaye SR, Adler J. Pseudo-Foster Kennedy syndrome in a patient with anterior ischemic optic neuropathy and a nonbasal glioma. *J Clin Neuro-Ophthalmol* 1990; 10:188-192.

Pseudo-Graefe Syndrome (Fuchs Sign) 1033

General: Misdirection of regenerating oculomotor nerve (cranial nerve III) fibers to other muscles after injury, aneurysm, tumor, exophthalmic goiter, tabes, anterior poliomyelitis, or vascular lesions of the brainstem.

Ocular: Elevation of the upper lid in downward gaze; lagging in upper lid movement on downward gaze (Graefe sign).

Clinical: None.

Bender MB. The nerve supply to the orbicularis muscle and the physiology of movements of the upper lid, with particular reference to the pseudo-Graefe phenomenon. *Arch Ophthalmol* 1936; 15:21.

Walsh FB, King AB. Ocular signs of intracranial saccular aneurysms. Experimental work on collateral circulation through the ophthalmic artery. *Arch Ophthalmol* 1942; 23:1169.

Pseudohypoparathyroidism Syndrome (Chronic Renal Tubular Insufficiency Syndrome; Seabright-Bantam Syndrome; Albright Hereditary Osteodystrophy) 1034

General: Etiology unknown; autosomal dominant; more common in females (2: 1); present from birth; kidney and skeleton fail to respond to parahormone; if patients receive parathyroid extract, their kidneys fail to respond with phosphate diuresis; genetic form of hypoparathyroidism resulting from end-organ resistance to parathyroid hormone; resulting hypocalcemia is responsible for many of the clinical features of this syndrome.

Ocular: Strabismus; blue sclera; punctate cataracts (white opacities and polychromatic cortex); papilledema; hypertelorism; keratitis; scleral and choroidal calcifications; blepharospasm; cataracts.

Clinical: Short stature; short metacarpals; short limbs; round face with short neck; decalcification of teeth; obesity; fat, stubby hands; tetany with positive Chvostek and Trousseau signs; atypical seizure disorder.

Albright F, et al. Pseudo-hypoparathyroidism: an example of "Seabright-Bantam syndrome." *Endocrinology 1942;* 30:922.

Blin O, et al. Blepharospasm associated with pseudohypoparathyroidism and bilateral basal ganglia calcifications. *Mov Disord* 1991; 6:379.

Faig JC, et al. Chronic atypical seizure disorder and cataracts due to diagnosis of pseudohypoparathyroidism. *West J Med* 1992; 157:64-65.

Wong S, et al. Scleral and choroidal calcifications in a patient with pseudohypoparathyroidism. *Br J Ophthalmol* 1979; 63:177-180.

Pseudo malignant Glaucoma Syndrome 1035

General: Related to obstruction of aqueous flow either by residual anterior hyaloid or by fibrin and other inflammatory debris at the level of the ciliary body/zonular apparatus following vitrectomy.

Ocular: Forward movement of lens-iris diaphragm; elevation of intraocular pressure; axial shallowing of anterior chamber; hydration of vitreous cavity.

Clinical: None.

Massicotte EC, Schuman JS. A malignant glaucoma-like syndrome following pars plana vitrectomy. *Ophthalmology* 1999; 106: 1375-1379.

Stem WH. Complications of vitrectomy. *Int Ophthalmol Clin* 1992; 32:205-212.

Pseudomonas Aeruginosa Infections 1036

General: Gram-negative rod with secondary contaminant of superficial wounds; *Pseudomonas* organisms produce a variety of enzymes that cause pathologic changes, including hemolysins and exotoxins as well as a glycocalyx that increases adherence.

Ocular: Hypopyon; conjunctivitis; keratitis; ulcerative abscess of cornea; endophthalmitis; panophthalmitis.

Clinical: Local tissue damage and diminished host resistance, which may occur in ear, lung, skin, and urinary tract.

Allen HF. Current status of prevention, diagnosis, and management of bacterial corneal ulcers. *Ann Ophthalmol* 1971; 3:235-246.

Cole GG, et al. Pseudomonas ophthalmia neonatorum: a cause of blindness. *Br Med J* 1980; 281:41-44.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Herbst RW. A guide to antibiotic therapy of ocular infections. *Ophthalmol Surg* 1972; 3:101-120.

Pseudoophthalmoplegia Syndrome (Roth-Bielschowsky Syndrome) 1037

General: Supranuclear lesion in the temporal lobe.

Ocular: Paralysis of lateral gaze in one direction; vestibular nystagmus in which the fast phase is absent on the ipsilateral side but the slow phase is present.

Clinical: Basal ganglia or tectum lesion.

Bielschowsky A. Das Klinische Bild der Assoziierten Blicklahmung und Seine Bedeutung fur die Topische Diagnostik. *Munchen Med Wochenschr* 1903; 40:1666.

Cogan DG, Adams RD. Type of paralysis of conjugate gaze (ocular motor apraxia). *Arch Ophthalmol* 1953; 50:434.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Roth WC. Demonstration von Kranken mit Ophthalmoplegie. *Neurol Zentbl* 1901; 20:921.

Pseudopapilledema (Optic Nerve Head Drusen) 1038

General: Autosomal dominant; incidence in males and females is approximately the same; two thirds of cases are bilateral; visual acuity usually unaffected; may cause slowly progressive visual field defect.

Ocular: Elevation of optic disk; drusen; injected conjunctiva; associated with retinitis pigmentosa, subretinal, subretinal pigment epithelium hemorrhages (rare).

Clinical: None.

Fite JD, Lewis AD. Family anomaly simulating papilledema: a case report. *J Pediatr* 1966; 68:927-931.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 1, 4th ed. Baltimore: Williams & Wilkins, 1982:358.

Pseudophakic Pigment Dispersion Syndrome 1039

General: Caused by rubbing of peripheral iris on lens zonules with iris plane and posterior chamber lenses.

Ocular: Transillumination of iris defects; pigment granules in aqueous humor; pigment dusting of anterior iris surface; band of pigment in filtration portion of trabecular meshwork; glaucoma.

Clinical: None.

Ballin N, Weiss DM. Pigment dispersion and intraocular pressure elevation in pseudophakia. *Ann Ophthalmol* 1982; 14:627-630.

Obstbaum SA. Management of glaucoma in the implanted patient. *J Am Intraocul Implant Soc* 1981; 7:252-259.

Pseudoprogeria Syndrome 1040

General: Rare; autosomal recessive; absent eyebrows and eyelashes with mental retardation.

Ocular: Glaucoma; absence of eyelashes and eyebrows.

Clinical: Progressive spastic quadriplegia; microcephaly; small, beaked nose; cervical spinal cyst; occipital cranium bifidum occultum; mental retardation.

Hall BD, et al. Pseudoprogeria-Hallerman-Streiff (PHS) syndrome. *Birth Defects* 1974; 10:137-146.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Pseudo-Pseudo-Foster Kennedy Syndrome 1041

General: Mimic disease of Pseudo-Foster Kennedy Syndrome consisting of ischemic optic neuropathy and meningioma.

Ocular: Optic disk pallor, visual field defects.

Clinical: Slow, progressive, decreased view.

Gelwan MJ, Seidman M, Kupersmith MJ. Pseudo-Pseudo-Foster Kennedy syndrome. *J Clin Neuro-Ophthalmol* 1988; 8:49-52.

Psittacosis (Ornithosis) 1042

General: Infectious disease transmitted from birds to man; causative agent is *Chlamydia psittaci;* poultry, pigeons, and parrots prominent carriers.

Ocular: Lid edema; corneal ulcers; scleritis; ophthalmoplegia; uveitis; acute focal retinitis; stellate retinopathy; cataract.

Clinical: Fever, malaise; headache; wheezing; intracranial hypertension.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Duke-Eider S, MacFaul PA, eds. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Prevett M, Harding AE. Intracranial hypertension following psittacosis. *J Neurol Neurosurg Psychiatry* 1993; 56:425.

Psoriasis (Psoriasis Vulgaris) 1043

General: Chronic skin disease of unknown etiology; both sexes affected; onset at any age; disease peaks at puberty; strong human leukocyte antigen (HLA) association resulting in heritable disease susceptibility.

Ocular: Desquamative psoriatic plaques of lids resulting in madarosis, trichiasis, or ectropion; corneal plaques; xerosis, symblepharon; keratitis; chronic corneal ulceration; phthisis bulbi; iritis.

Clinical: Thick, dry, elevated red patches of skin covered with coarse silvery scales that usually affect areas of skin not exposed to sun, such as scalp, sacrum, elbows, and knees; positive association with Sjögren syndrome and keratitis sicca.

Boss JM, et al. Peripheral corneal melting syndrome in association with psoriasis: a report of two cases. *Br Med J (Clin Res)* 1981; 282:609-610.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Collins P, et al. Psoriasis, psoriatic arthritis and the possible association with Sjogren's syndrome. *Br J Dermatol* 1992; 126:242-245.

Elder JT, et al. The genetics of psoriasis. *Arch Dermatol* 1994; 130:216-224.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Knox DL. Psoriasis and intraocular inflammation. *Trans Am Ophthalmol Soc* 1979; 77:210-224.

Meyer E, et al. Psoriasis associated with anterior uveitis. *J Pediatr Ophthalmol* 1977; 14:213-214.

Psoriatic Arthritis 1044

General: Chronic skin disease of unknown etiology; both sexes affected; onset at any age; disease peaks at puberty.

Ocular: Conjunctivitis; iritis; keratitis; uveitis.

Clinical: Rash; spondylitis; inflammatory bowel disease; diarrhea; degenerative disease of spine.

Lambert JR, Wright V. Eye inflammation in psoriatic arthritis. *Ann Rheum Dis* 1976; 35:354-356.

Pterygium of Conjunctiva and Cornea 1045

General: Autosomal dominant; more frequent in people who work outdoors; occurs late in life.

Ocular: Wing-shaped thickening in the conjunctiva, usually nasal, in the interpalpebral fissure area; alterations in corneal topography can cause a reduction in visual acuity.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Walland MJ, et al. The effect of recurrent pterygium on corneal topography. *Cornea* 1994; 13:463-464.

Puckering Syndrome 1046

General: Disturbance of vitreous with vitreous retraction.

Ocular: Epiretinal membrane formation following vitreous bleed, total retinal detachment, detachment of the macula, multiple retinal operations, multiple perforations, loss of formed vitreous at operation, or posterior vitreous separation from trauma to the eye or whiplash injury.

Clinical: Neck injury with whiplash.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Tannenbaum HL, et al. Macular pucker following retinal detachment surgery. *Arch Ophthalmol* 1970; 83:286-293.

Pugh Syndrome 1047

General: May be misdiagnosed as UGH (uveitis, glaucoma, hyphemas) syndrome; unrelated to the presence of an intraocular lens.

Ocular: Pseudouveitis; glaucoma; hyphemas as in UGH syndrome, but also neovascular membrane covering the iris; central retinal vein occlusion.

Epstein E. UGH syndrome and contact lenses. *J Cataract Refract Surg* 1987; 13:216-217.

Hagan JC. Complications while removing IOLAB IZ lens for the UGH-UGH+ syndrome. *J Am Intraocul Implant Soc* 1984; 10:209-213.

Pulmonary Insufficiency 1048

General: Elevated CO_2 levels.

Ocular: Tortuosity of conjunctival and retinal vasculature; retinal hemorrhages; papilledema; retinal edema.

Clinical: Vascular decompensation; elevation of cerebrospinal fluid pressure; polycythemia; chronic respiratory disease.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Mason, 1982.

Pupil, Egg-Shaped 1049

General: Autosomal dominant; rare.

Ocular: Oval pupils; enlarged pupils; pupils react poorly to constricting stimuli.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

White BV, et al. A rare pupillary defect inherited by identical twins. *J Hered* 1937; 28: 177-179.

Pupillary Membrane, Persistent 1050

General: Autosomal dominant.

Ocular: Remnants of pupillary membrane persist as strands and other irregular tissue in pupil; congenital cataract; corneal edema; Rieger syndrome; keratoconus.

Clinical: None.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Purtscher Syndrome (Fat Embolism Syndrome; Traumatic Retinal Angiopathy; Traumatic Liporrhagia; Valsalva Retinopathy of Duane; Duane Retinopathy) 1051

General: Most frequently seen in accidents associated with sudden rise in blood pressure and congestion in the head and chest; presence of fat embolism may be the causative factor; neurovascular changes in retina referred to as traumatic retinal angiopathy; several mechanisms have been proposed, including compressive trauma and posttraumatic fat embolism; most likely mechanism appears to be leukocyte aggregation by activated complement factor 5 (C5A), which can occur in diverse conditions such as trauma, acute pancreatitis, and connective tissue disease.

Ocular: Retinal and preretinal hemorrhages over entire fundus; cotton-wool exudates, mainly posterior aspect; retinal edema; posterior and macular serous detachment; venous congestion and engorgement; papilledema; usually bilateral, although unilateral causes have been reported.

Clinical: Multiple fractures (mainly extensive crushing); lung congestion; dyspnea; lymphorrhagia; pancreatitis; scleroderma; dermatomyositis; lupus erythematosus; childbirth.

Burton TC. Unilateral Purtscher's retinopathy. *Ophthalmology* 1980; 87:1096-1105.

Hoare GW. Traumatic retinal angiopathy resulting from chest compression by safety belt. *Br J Ophthalmol* 1970; 54:667.

Inkeles DM, et al. Purtscher's retinopathy in acute pancreatitis. *Am J Med Sci* 1976; 2772:335-338.

Kelley JS, Dhaliwal RS. Traumatic chorioretinopathies. In: Ryan S, ed. *Retina. vol. II*, 2nd ed. St. Louis: Mosby, 1994.

Purtscher O. Angiopathia Retinae Traumatica. Lymphorrhagien des Augengrundes. *Graefes Arch Clin Exp Ophthalmol* 1912; 82:341.

Pyoderma Vegetans (Dermatitis Vegetans) 1052

General: Occurs in both sexes and all ages; frequently seen with preexisting eczema or infective dermatitis; nonspecific skin reaction; chronic; malnutrition and alcoholism are contributing factors.

Ocular: Lid lesions; keratitis.

Clinical: Chronic granulomatous growth with epithelial hyperplasia often with pustules, ulcers, multiple abscesses, and fistulae; lesions of mouth and nose.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Rook A, et al. *Textbook of dermatology*, 2nd ed. Philadelphia: FA Davis, 1972.

Pyostomatitis 1053

General: Reaction of chronic inflamed skin due to secondary infection; body resistance leads to folding of mucous membrane and development of a verrucous surface.

Ocular: Conjunctivitis; blepharitis.

Clinical: Early lesions are small pustules that can develop to large, weeping inflammatory plaques with foul odors; massive crust may form granulations of buccal mucosa, hand, soft palate, lips, and gingiva.

Buxman MM, et al. Harlequin ichthyosis with epidermal lipid abnormality. *Arch Dermatol* 1979; 115:189.

Erickson L, Kahn G. The granular layer thickness in atrophy and ichthyosis vulgaris. *J Invest Dermatol* 1970; 54: 11.

Q

Q Fever 1054

General: Acute rickettsial infection caused by *Coxiella burnetii;* at least 11 serotypes of this organism are capable of causing human infection; elevated inflammatory response results in granulomatous formation.

Ocular: Conjunctivitis; gangrene of eyelids; retinal hemorrhages; perivasculitis; episc1eritis; optic neuritis; uveitis; papilledema; nystagmus; ocular motor nerve pareses; Miller-Fisher syndrome.

Clinical: Fever; severe headache; tissue necrosis; pneumonia; self-limited fever; endocarditis; hepatitis.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:240.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Garibaldi RA. Q fever. In: Conn HF, ed. *Current therapy.* Philadelphia: WB Saunders, 1977:60-61.

Ley HL Jr. Q fever. In: Beeson PB, McDermott W, Wyngaarden JB, eds. *Textbook of medicine.* 15th ed. Philadelphia: WB Saunders, 1979:329-331.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part 1,* 4th ed. Baltimore: Williams & Wilkins, 1994:3623-3633.

Stein A, Raoult D. Phenotypic and genotypic heterogeneity of 8 new human Coxiella burnetii isolates. *Acta Virol* 1992; 36:7-12.

R

Radiation Injury (Gamma Rays; Infrared Rays; Microwaves; Radio Waves; X-Rays) 1055

General: Electromagnetic radiation can cause ionization in biologic tissues.

Ocular: Hyperemia of conjunctiva; corneal ulcer; punctate keratitis; keratoconjunctivitis sicca; blepharitis; ectropion; entropion; madarosis; poliosis; depigmentation of eyelids; uveitis; atrophy of lacrimal gland; cataracts; true exfoliation of lens capsule; orbital necrosis; retinal hemorrhage; macular degeneration; macular holes; neovascularization of retina; glaucoma; macular edema; retinal microvascular changes; optic nerve edema and hemorrhage; atrophy.

Clinical: Thermal bums of any part of the body; necrosis; carcinomas; edema.

Bagan SM, Hollenhorst RW. Radiation retinopathy after irradiation of intracranial lesions. *Am J Ophthalmol* 1979; 88:694-697.

Kinyoun C, et al. Radiation retinopathy. *Am J Ophthalmol* 1988; 105:471-478.

Mukai S, Guyer DR, Gragoudas ES. Radiation retinopathy. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology. vol. II.* Philadelphia: WB Saunders, 1994:1038-1041.

Raeder Syndrome (Paratrigeminal Paralysis; Horton Headache; Histamine Cephalalgia; Ciliary Neuralgia; Cluster Headache; Periodic Migrainous Neuralgia) 1056

General: Interruption of sympathetic fibers about the carotid artery and involvement of the fifth nerve; meningioma and aneurysm of the internal carotid artery most frequent causes; prominent in males; possible pathogenetic mechanism of this condition is an ischemic injury of the gasserian ganglion.

Ocular: Mild enophthalmos; mild ptosis (unilateral); epiphora; scotoma possible; hypotonia; unilateral miosis; increased tear secretion; periocular pain; Homer syndrome.

Clinical: Facial pain; occasionally weakness of the jaw muscles; headaches (V-region); hypertension; associated inflammatory processes are not infrequent.

Boniuk M, Schlezinger NS. Raeder's paratrigeminal syndrome. *Am J Ophthalmol* 1962; 54: 1074.

Coppeto JR. Migraine and other head pains. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV* Philadelphia: WB Saunders, 1994:2694.

Desai BT, et al. Raeder's syndrome. *Ann Ophthalmol* 1975; 7:1082-1084.

Fraunfelder FT, Roy FH. *Current Ocular Therapy.* 5th ed. Philadelphia: WB Saunders, 2000.

Grimson BS, Thompson S. Raeder's syndrome: a *Clinical* review. *Surv Ophthalmol* 1980; 24:199-210.

Lederman RJ. Fibromuscular dysplasia of the internal carotid artery. A cause of Raeder's paratrigeminal syndrome. *Neurology* 1976; 26:353.

Raeder JG. "Paratrigeminal" paralysis of oculo-pupillary sympathetic. *Brain* 1924; 47:149.

Riley FC Jr, Moyer NJ. Oculosympathetic paresis associated with cluster headaches. *Am J Ophthalmol* 1971; 72: 763-768.

Raymond Syndrome (Raymond-Cestan Syndrome; Cestan [2] Syndrome; Pontine Syndrome; Disassociation of Lateral Gaze Syndrome) 1057

General: Lesion involving the pyramidal tracts as they traverse the pons; posterior longitudinal bundle and medial lemniscus may be involved; tumor and vascular thromboses are common causes; can be caused iatrogenically after neurosurgical procedures.

Ocular: Ipsilateral abducens palsy; paralysis of lateral conjugate gaze.

Clinical: Contralateral hemiplegia; anesthesia of the face, limbs, and trunk.

Isobe I, et al. Rare case of Raymond-Cestan syndrome. *Naika* 1970; 26:388.

Raymond F, Cestan R. Trois Observations de Paralysie des Mouvements Associes des Globes Oculaires. *Rev Neural (Paris)* 1901; 9:70-77.

Seyer H, et al. Raymond's syndrome following petrosal sinus sampling. *Acta Neurachir* 1994; 131:157-159.

Raynaud Disease (Symmetrical Gangrene; Symmetrical Asphyxia) 1058

General: Primary, or idiopathic, form of paroxysmal digital cyanosis; possible abnormality of sympathetic nervous system; occurs in females between ages 15 and 40 years.

Ocular: Spasm of retinal arteries; papillitis; retrobulbar neuritis; amaurosis fugax; cotton-wool spots; retinal hemorrhages; transient corneal opacification.

Clinical: Intermittent attacks of pallor or cyanosis in fingers, precipitated by cold or occasionally by emotional upsets; atrophy of terminal fat pads and digital skin; gangrenous ulcers.

Chatton MJ, Krupp MA. *Current medical diagnosis and treatment.* Los Altos, CA: Lange Medical Publishers, 1981:269-271.

Harley RD, ed. *Pediatric Ophthalmology,* 4th ed. Philadelphia: WB Saunders, 1998.

McWhae JA, Andrews DM. Transient corneal opacification induced by cold in Raynaud's disease. *Ophthalmology* 1991; 98:666-669.

Salmenson BD, et al. Macular capillary hemodynamic changes associated with Raynaud's phenomenon. *Ophthalmology* 1992; 99:914-919.

Storimans CW, et al. A new autosomal dominant vascular retinopathy syndrome. *Eur J Ophthalmol* 1991; 1:73.

Rebeitz-Kolodny-Richardson Syndrome 1059

General: Etiology unknown; possibly metabolic failure at cellular level; occurs in late middle age; neural achromasia with corticodentatonigral degeneration.

Ocular: Paralysis of ocular muscles.

Clinical: Clumsiness, slowness of movement of left limbs; severe impairment in control of muscular movements; postural abnormalities; tremor; severe contractures; dysphagia; speech impairment; Babinski sign.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Rebeitz JJ, Kolodny H, Richardson EP. Corticodentatonigral degeneration with achromasia. *Arch Neural* 1968; 18:20-33.

Refsum Syndrome (Heredopathia Atactica Polyneuritiformis Syndrome; Phytanic Acid Oxidase Deficiency; Phytanic Acid Storage Disease; Refsum-Thiebaut Syndrome) 1060

General: Autosomal recessive; disorder of lipid metabolism; interstitial hypertrophic polyneuropathy; delamination of myelin sheaths; onset usually between ages 4 and 7 years; caused by deficiency of phytanic acid hydroxylase.

Ocular: Progressive external ophthalmoplegia; night blindness; visual field constriction; pupillary abnormalities; corneal opacities; retinal degeneration beginning in macula; retinitis pigmentosa; cataracts.

Clinical: Spinocerebellar ataxia; deafness (progressive); polyneuritis-like effect on limbs; central nervous system degeneration; ichthyosis; sensory changes; wasting of extremities; complete heart block; relapses and remissions in adolescence; normal intelligence.

Collins JF. *Handbook of Clinical Ophthalmology.* New York: Masson, 1982, 249.

Gibberd FB, et al. Heredopathia atactica polyneuritiformis (Refsum's disease) treated by diet and plasma exchange. *Lancet* 1979; 1:575-578.

Hansen E, et al. Refsum's disease. Eye manifestations in a patient treated with a low phytol, low phytanic acid diet. *Acta Ophthalmol* 1979; 57:899.

Refsum S. Heredo-ataxia hemeralopica polyneuritiformis familial syndrome. *Acta Psychiatr Scand (Suppl)* 1946; 38:1.

Smith LH. Inherited metabolic disease with Pediatric Ocular Manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology. vol. V.* Philadelphia: WB Saunders, 1994:2783.

Toussaint D, Danis P. An ocular pathologic study of Refsum's syndrome. *Am J Ophthalmol* 1971; 72:342.

Weleber RG, et al. Ophthalmic manifestations of infantile phytanic acid storage disease. *Arch Ophthalmol* 1984; 102:1317-1321.

Reimann Syndrome (Hyperviscosity Syndrome) 1061

General: Frequently found in association with Waldenström syndrome, other hyperglob-ulinemias, and occasionally with myeloma, reticulum sarcoma, and other tumors; increase in y-globulin or other dysproteinemias that affect blood viscosity (see Bing-Neel Syndrome; Waldenström Syndrome); leukemia.

Ocular: Nystagmus; tortuosity of conjunctival vessels; sludging phenomenon with aggregation of intravascular erythrocytes; retinal vascular tortuosity and sludging; retinal hemorrhages and exudates; micro aneurysms of various degrees and sizes; central retinal vein occlusion.

Clinical: Mucous membrane; hemorrhages; headaches; paraesthesia; ataxia; heart failure and low pulse pressure; partial loss of hearing; anorexia; vertigo; dyspnea; syncope; convulsions; peripheral edema; sausage-shaped veins.

Luxenberg MN, Mausolf FA. Retinal circulation in the hyperviscosity syndrome. *Am J Ophthalmol* 1970; 70:588.

Reimann HA. Hyperproteinemia as cause of autoagglutination: observations in case of myeloma. *J Am Med Assoc 1932;* 99:1411.

Ryan S, ed. *Retina,* vol. I, 2nd ed. St. Louis: Mosby, 1994.

Reiter Syndrome (Fiessinger-Leroy Syndrome; Conjunctivo-Urethro-Synovial Syndrome; Idiopathic Blennorrheal Arthritis Syndrome; Polyarthritis Enterica) 1062

General: Etiology unknown; males; onset ages 16 to 42 years; probably a combined infectious/autoimmune pathogenetic mechanism; reactive arthritis probably associated with infection with many different species of microorganisms; HLA-B27 confers disease susceptibility to infection.

Ocular: Sterile mucopurulent conjunctivitis, usually bilateral; photophobia; epiphora; iritis; keratitis; uveitis; paralysis of extraocular muscles; optic neuritis; secondary glaucoma; hypopyon; hyphema.

Clinical: Skin erythema; genital ulcerations; urethritis with discharge; cystitis with dysuria, abacterial pyuria, and hematuria; arthritis with pain, swelling, heat, and effusion; fever; weight loss; fatigue; malaise; fever; diarrhea; oral mucosal lesions; arthralgia.

Fong DS, Raizman MB. Spontaneous hyphema associated with anterior uveitis. *Br J Ophthalmol* 1993; 77:635-638.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hughes RA, Keat AC. Reiter's syndrome and reactive arthritis: a current view. *Semin Arthritis Rheum 1994;* 24:190-210.

Kiss S, Letko E, Qamruddin S, et al. Long-term progression, prognosis, and treatment of patients with recurrent ocular manifestations of Reiter's syndrome. *Ophthalmology* 2003; 110: 1764-1769.

Lee DA, et al. The *Clinical* diagnosis of Reiter's syndrome. *Ophthalmology* 1986; 93:350-356.

Ostler HB, et al. Reiter's syndrome. *Am J Ophthalmol* 1971; 71:986.

Vergnani RJ, Smith RS. Reiter's syndrome in a child. *Arch Ophthalmol* 1974; 91:165.

Relapsing Fever (Recurrent Fever) 1063

General: Acute infectious disease caused by *Borrelia* transmitted by lice; characterized by recurrent bouts of fever separated by relatively asymptomatic periods; there is an endemic form of rheumatic fever transmitted by tick vectors and spirochetes of the genus *Borrelia.*

Ocular: Extraocular muscle paralysis; uveitis; interstitial keratitis; hypopyon; conjunctivitis; optic nerve atrophy; subconjunctival and retinal hemorrhages; ptosis; mydriasis; retinal venous occlusion.

Clinical: Toxemia and febrile paroxysms separated by afebrile periods.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Whitcup SM, Raizman MB. Spirochetal infections and the eye. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3078-3092.

Relapsing Polychondritis (Jaksch Wartenhost Syndrome; Meyenburg-Altherz-Vehlinger Syndrome; Von Meyenberg II Syndrome) 1064

General: Episodic, yet generally progressive; onset usually in middle life; possibly caused by lysosomal labilizing factor of endogenous or exogenous toxic nature or immunologic reactions; possible association with Reiter syndrome.

Ocular: Conjunctivitis; corneal ulcer; exophthalmos; panophthalmitis; phthisis bulbi; proptosis; optic neuritis; papilledema; retinal detachment; blue sclera; episcleritis; scleromalacia; vitreous opacity; cataracts; nystagmus; retinal artery thrombosis; keratoconjunctivitis sicca; secondary glaucoma; scotoma; uveitis; paresis of third or sixth nerve; conjunctival mass (salmon patch); chorioretinitis.

Clinical: Destruction of cartilage and eventual replacement with connective tissue; polyarthritis; chondritis; tracheal collapse; bronchial collapse; anemia; liver dysfunction; death; malaise; fever; dyspnea; changes in pitch of voice; hearing impairment; vertigo; deformed ears; aortic valve insufficiency.

Arkin CR, Masi AT. Relapsing polychondritis: review of current status and case report. *Semin Arthritis Rheum* 1975; 5:41-62.

Brink H, et al. Ocular manifestations of relapsing polychondritis. Three case histories. *Doc Ophthalmol* 1994; 87:159-166.

Crovato F, et al. Exophthalmos in relapsing polychondritis. *Arch Dermatol* 1980; 116:383-384.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Isaak BL, et al. Ocular and systemic findings in relapsing polychondritis. *Ophthalmology* 1986; 93:681-689.

McKay DAR, et al. Relapsing polychondritis and eye disease. *Br J Ophthalmol* 1974; 58:600-605.

Silva J, et al. Relapsing polychondritis and Reiter's syndrome. *J Rheumatol* 1991; 18:908-910.

Tucker SM, et al. Relapsing polychondritis, another cause for a "salmon patch." *Ann Ophthalmol* 1993; 25:389-391.

Renal Failure 1065

General: Absence of renal function.

Ocular: Cotton-wool spots; retinal edema; optic disk edema; conjunctival calcium deposits; band keratopathy; cortical blindness; severe retinopathy is more likely to be found in patients with renal insufficiency; patients undergoing hemodialysis are at increased risk of elevated intraocular pressure, particularly in eyes that have undergone vitrectomy.

Clinical: Hypertension; azotemia; hypervolemia; metabolic disturbances; hyponatremia; hypercalcemia.

Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine*. 11th ed. New York: McGraw-Hill, 1987.

Collins JF. *Handbook of Clinical Ophthalmology* New York: Masson, 1982.

Miller JW, D'Amico DJ. Proliferative diabetic retinopathy. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:760-782.

Renal Transplantation 1066

General: Ocular complication due to systemic immunosuppressive drugs used to prevent rejection of the transplanted kidney.

Ocular: Cataracts; steroid-induced glaucoma; cytomegalovirus retinitis; fungal endoph-thalmitis; conjunctivitis; diffuse anterior scleritis with possible association with OKT3 monoclonal antibody therapy for renal transplant rejection; typical herpetic dendritic keratitis.

Clinical: Renal failure; kidney transplant.

Astle JN, Ellis PP. Ocular complications in renal transplant patients. *Ann Ophthalmol* 1974; 6:1269-1274.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:258.

Matsunami C, et al. Ocular complications in renal transplant patients. *Aust N Z J Ophthalmol* 1994; 22:53-57.

McCarthy JM, et al. Diffuse anterior scleritis during OKT3 monoclonal antibody therapy for renal transplant rejection. *Can J Ophthalmol* 1992; 27:22-24.

Rendu-Osler Syndrome (Rendu-Osler- Weber Syndrome; Hereditary Hemorrhagic Telangiectasis; Babington Disease; Goldstein Hematemesis; Osler Syndrome [2]) 1067

General: Etiology unknown; autosomal dominant in Jews; repeated epistaxis begins in childhood; gastrointestinal hemorrhages with melena and hematemesis manifest in middle and later life.

Ocular: Star-shaped angiomas of the palpebral conjunctiva; intermittent filamentary keratitis; small retinal angiomas and occasionally retinal hemorrhages; subconjunctival hemorrhages; small retinal arteriovenous malformations; bloody tears; conjunctival telangiectasias.

Clinical: Epistaxis; hematuria; melena; angiomata of the pharynx and oral and nasal mucosa; angiomas on lips, face, and upper extremities; cyanosis; polycythemia.

Brant AM, et al. Ocular manifestations III hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber disease). *Am J Ophthalmol* 1989; 107:642.

Ebert EM, Boger WP III, Albert DM. Phakomatoses. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3323.

Osler W. On a familial form of recurring epistaxis associated with multiple telangiectases of the skin and mucous membrane. *Bull Johns Hopkins Hosp* 1901; 12:333.

Rendu M. Epistaxis Repetees Chez un Sujet Porteur de Petits Angiomes Cutanes et Muqueux. *Bull Mem Soc Med Hop* 1896; 13:731.

Vase I, Vase P. Ocular lesions in hereditary haemorrhagic telangiectasia. *Acta Ophthalmol* 1979; 57:1084-1090.

Wolper J, Laibson PR. Hereditary hemorrhagic telangiectasis (Rendu-Osler-Weber disease) with filamentary keratitis. *Arch Ophthalmol* 1969; 81:272.

Reticular Degeneration of Pigment Epithelium (RDPE) 1068

General: Etiology unknown; associated with age-related macular degeneration.

Ocular: Hypopigmentation of retinal pigment epithelium interspersed with hyperpigmented lines forming a coarse netlike pattern of irregular polygons; multiple drusen of peripheral fundus; choroidal nevus; central retinal vein occlusion.

El Baba F, et al. Clinicopathologic correlation of lipdation and detachment of the retinal pigment epithelium. *Am J Ophthalmol* 1986; 101:576-583.

Gass JDM, et al. Drusen and disciform macular detachment and degeneration. *Arch Ophthalmol* 1973; 90:206-217.

Lewis H, et al. Reticular degeneration of the pigment epithelium. *Ophthalmology* 1985; 92:1485-1495.

Reticular Pigmentary Retinal Dystrophy of Posterior Pole (Sjögren Disease) 1069

General: Autosomal recessive; characterized by peculiar network of black-pigmented lines in posterior pole of retina.

Ocular: Fishnet-like knots on posterior pole of retina; drusen.

Clinical: None.

Deutman AF, et al. Reticular dystrophy of retinal pigment epithelium. Dystrophia reticularis laminae pigmentosae retinal of Sjogren. *Arch Ophthalmol* 1969; 82:4-9.

Deutman AF. Macular dystrophies. In: Ryan SJ, ed. *Retina, vol. II,* 2nd ed. St. Louis: Mosby, 1994:1216-1217.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Wocheslander E, et al. Die retikulare Pigment dystrophie nach Sjogren. *Klin Monatsbl Augenheilkd* 1980; 176:684.

Reticulum Cell Sarcoma (Non-Hodgkin Lymphoma) 1070

General: Autosomal recessive; large-cell lymphoma with chronic inflammation with a predominance of cells in vitreous cavity; average age at time of diagnosis is 60 years; female to male ratio is approximately 2: 1; 80% bilateral (frequently asymmetrical).

Ocular: Chronic uveitis; chorioretinal lesions; mycosis fungoides; necrosis of orbital tissues; phthisis bulbi; endophthalmos; exophthalmos; exudative retinal detachment; iris neovascularization; glaucoma; branch retinal vein occlusion; macular edema; optic neuropathy; vitreous hemorrhage; partial cranial nerve III palsy; multiple retinal pigment epithelium masses.

Clinical: Lymphocytic hyperplasia; fever; anemia; thrombocytopenia; liver and spleen enlargement; associated with immune dysfunction states, such as acquired immunodeficiency syndrome, or following transplantation.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Lewis H, Schachat AP. Non-Hodgkin's ("reticulum cell") lymphoma. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:893.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology*, 4th ed. Baltimore: Williams & Wilkins, 1987.

Retinal Arteries, Tortuosity 1071

General: Autosomal dominant; retinal vascular tortuosity is the isolated physical finding but may be associated with a variety of ocular and systemic anomalies; tortuosity predominantly found in the arteriolar tree.

Ocular: Retinal vascular tortuosity; foveal hemorrhage; macular and paramacular hemorrhages; retinal hemorrhages; peripapillary retinal hemorrhage; juxtapapillary retinal hemorrhage; conjunctival hemorrhage; retinal telangiectasia; asteroid hyalitis; amblyopia; myopia; hyperopia; glaucoma; Coats disease; von Hippel-Lindau syndrome; racemose aneurysms of the retina; arteriolar ectasia.

Clinical: Polycythemia; splenomegaly; recurrent nosebleeds; nasal telangiectasia; pulmonary emphysema; systemic hypertension; aortic coarctation; hereditary hemorrhagic telangiectasia; leukemia; macroglobulinemia; cryoglobulinemia; sickle cell disease; familial dysautonomia; Maroteaux-Lamy syndrome; Fabry disease; hypogammaglobulinemia; intracranial or facial aneurysmal abnormalities; migraine headaches; psoriasis; colitis; diabetes mellitus; hypertension.

Goldberg MF, et al. Familial retinal arteriolar tortuosity with retinal hemorrhage. *Am J Ophthalmol* 1972; 73: 183-191.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Retinal Cone Degeneration 1072

General: Autosomal dominant; diffuse cone degeneration; progressive loss of visual acuity; macular lesion has a bull's-eye appearance produced by a central area of uninvolved epithelium.

Ocular: Photophobia; defective color vision; loss of side vision; night blindness; macular lesion; poor central acuity and visual field scotomata closer to fixation compared to patients with retinal rod degeneration.

Clinical: None.

Heckenlively JR, et al. Telangiectasia and optic atrophy in cone-rod degenerations. *Arch Ophthalmol* 1981; 99: 1983-1991.

Krill AE, et al. The cone degenerations. *Doc Ophthalmol* 1973; 35:1-80.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995.

Rabb MF, et al. Cone-rod dystrophy: a *Clinical* and histopathologic report. *Ophthalmology* 1986; 93:1443-1450.

Weleber RG. Retinitis pigmentosa and allied disorders. In: Ryan SJ, ed. *Retina, vol. L* 2nd ed. St. Louis: Mosby, 1994:359-360.

Retinal Detachment 1073

General: Autosomal dominant; multilayered retinal tissue separates along an interface formed in course of embryonic development.

Ocular: Retinal tear or hole; retinal detachment.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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McNeil NA, McPherson A. The inheritance of detached retina in a texas family. *J Hered* 1971; 62:73-76.

Retinal Disinsertion Syndrome 1074

General: None.

Ocular: Subluxation of the lens; microphthalmos; bilateral keratoconus; retinal detachment.

Clinical: None.

Ryan SJ, ed. *Retina, vol. III,* 2nd ed. St. Louis: Mosby, 1994.

Havland KP, et al. Developmental giant retinal tears associated with lens coloboma. *Arch Ophthalmol* 1968; 80:325-331.

Shammas HJF, McGaughey AS. Retinal disinsertion syndrome: report of a case. *J Pediatr Ophthalmol Strabismus* 1979; 16:284-286.

Retinal Ischemic Syndrome 1075

Ocular: Bilateral conjunctival injection; iris rubeosis; posterior synechia; retinal vascular tortuosity; retinal detachment; Axenfeld's syndrome; central retinal artery occlusion; glaucoma; corneal anesthesia; uveitis; ischemic retinopathy

Clinical: Associated with partial occlusion of carotid artery**;** diabetes, hypercholesterolemia; tetralogy of fallot

Brear DR, Insler MS: Axenfeld's syndrome associated with systemic abnormalities. *Ann Ophthalmol* 1985; 17: 291-294

Wu WC, Lai CC, Su WJ, et al. Ischemic retinopathy and uveitis in a patient with tetralogy of fallot. *Ophthalmology* 2005; 112: 1936-1940

Retinal Vascular Hypoplasia with Persistence of Primary Vitreous 1076

General: Bilateral congenital retinopathy characterized by retinal vascular hypoplasia and persistence of primary vitreous; etiology unknown.

Ocular: Buphthalmos; microphthalmia; fixed and dilated pupils; neovascularization of iris; glaucoma; cataract; white opaque fibrovascular retrolental membrane; retinal detachment; vitreous hemorrhage; retinal vascular hypoplasia.

Clinical: None.

Ryan SJ, ed. *Retina, vol. II*, 2nd ed. St. Louis: Mosby, 1994.

Pollard JF. Treatment of persistent primary hypoplastic primary vitreous. *J Pediatr Ophthalmol Strabismus* 1985; 22:180-183.

Pruett RC. The pleomorphism and complications of posterior hyperplastic primary vitreous. *Am J Ophthalmol* 1975; 80:625-629.

Sneed PJ, et al. Bilateral retinal vascular hypoplasia associated with persistence of the primary vitreous: a new *Clinical* entity? *J Pediatr Ophthalmol Strabismus* 1988; 25:77-85.

Retinitis Pigmentosa, Deafness, Mental Retardation, and Hypogonadism 1077

General: Autosomal recessive; similar to Laurence-Moon-Biedl-Bardet, Alstrom, and Usher syndromes, with the difference being the absence of polydactyly.

Ocular: Nystagmus; hyperplasia and thickening of prickle cell layer to lids or comer of eyes; myopia; retinitis pigmentosa; keratoconus.

Clinical: Acanthosis nigricans; multiple keloids; gynecomastia, small testes in males; oligomenorrhea; mental retardation; deafness; hypogonadism; glucose intolerance; hyperinsulinism.

Edward JA, et al. A new familial syndrome characterized by pigmentary retinopathy, hypogonadism, mental retardation, nerve deafness and glucose intolerance. *Am J Med* 1976; 60:23-32.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Retinoblastoma 1078

General: Malignant tumor arising in one or both retinas of young children, usually under the age of 2 years; usually unilateral; autosomal dominant; most common intraocular malignancy of childhood; incidence is one in 20,000 live births; origin is questionably neuroectodermal cells capable of multipotentiality; one third of patients have heritable (bilateral or have a positive family history) autosomal dominant and two thirds are sporadic; genetic transmission obeys two-mutation hypothesis of Knudson; trilateral retinoblastoma is bilateral retinoblastoma plus midline central nervous system tumor (most commonly pinealoma); most common second tumor is an osteogenic sarcoma (begins in second decade).

Ocular: Hyphema; hypopyon; corneal tumor; lid edema; endophthalmitis; exophthalmos; intraocular calcification of globe; heterochromia; neovascularization of iris or retina; papilledema; panophthalmitis; retinoblastoma extension into orbit and choroid; cat's-eye reflex; leukocoria; mydriasis; vitreous hemorrhage tumor seeding; esotropia; exotropia; glaucoma; visual loss.

Clinical: Metastasis into the lymph system, bone marrow, and subarachnoid space; basal meningitis; death.

Abramson DH, Ellsworth RM. The surgical management of retinoblastoma. *Ophthalmic Surg* 1980; 11:596-598.

Abramson DH, et al. The management of unilateral retinoblastoma without primary enucleation. *Arch Ophthalmol* 1982; 100:1249-1252.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Lueder GT, Smith ME. Retinoblastoma. *Semin Diagn Pathol* 1994; 11: 104-106.

Ohira A, et al. Glial-neuronal-and photoreceptor-specific cell markers in rosettes of retinoblastoma and retinal dysplasia. *Curr Eye Res* 1994; 13:799-804.

Retinohypophysary Syndrome (Benign Retinohypophysary Syndrome; Lijo Pavia-Lis Syndrome) 1079

General: Alterations of the bony structure of the sella turcica with decalcifications and osteolysis of the posterior clinoid process; all ages; more frequent in women.

Ocular: Superior nasal field contraction; narrowing of retinal vessels; macular edema; optic neuritis; optic atrophy; visual field defects.

Clinical: Glycosuria; headache; vertigo; psychic disturbances.

Geeraets WJ *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Lijo Pavia J Retino-hypophysary syndrome: treatment with gonadotropin. Four new cases. *Rev Otoneurooftal* 1947; 22:5; *Am J Ophthalmol* 1948; 31:382(abst).

Retinopathy, Pigmentary, and Mental Retardation (Mirhosseini-Holmes-Walton Syndrome) 1080

General: Autosomal recessive; this disorder may be the same as (or alleli) to Cohen syndrome.

Ocular: Pigmentary retinal degeneration; cataract; keratoconus.

Clinical: Microcephaly; severe mental retardation; hyperextensible joints; scoliosis; arachnodactyly; hypogonadism.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Mendez HM, et al. The syndrome of retinal pigmentary degeneration, microcephaly and severe mental retardation (Mirhosseini-Holmes-Walton syndrome): report of two patients. *Am J Med Genet* 1985; 22:223-228.

Steinlein O, et al. Tapetoretinal degeneration in brothers with apparent Cohen syndrome: nosology with Mirhosseini-Holmes-Walton syndrome. *Am J Med Genet* 1991; 41:196-200.

Retinoschisis (RS) 1081

General: Sex-linked; may not manifest until middle life.

Ocular: Intraretinal splitting due to degeneration or detachment of retina; retinal atrophy with sclerosis of the choroid; cystic maculopathy.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Wieacker P, et al. Linkage relationships between retinoschisis Xg and a cloned DNA sequence from the distal short arm of X chromosome. *Hum Genet* 1983; 64:143-145.

Retinoschisis, Acquired (Acquired Retinoschisis [RS]) 1082

General: Present in 4% to 22% of normal population over age 40 years; splitting in the outer plexiform layer; occasionally in the inner nuclear layer; generally asymptomatic.

Ocular: Slowly progressive; may cause retinal detachment when there are breaks in both outer and inner layers of retinoschisis.

Clinical: None.

Hirose T. Retinoschisis. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:1071-1084.

Schepens CL. *Retinal detachment and allied diseases, Vol.* 2. Philadelphia: WB Saunders, 1983.

Retinoschisis, Autosomal Dominant 1083

General: Autosomal dominant; degenerative abnormal splitting of retinal sensory layers.

Ocular: Loss of retinal function; retinal degeneration; macular degeneration.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

YassurY, et al. Autosomal dominant inheritance of retinoschisis. *Am J Ophthalmol* 1982; 94:338-343.

Retinoschisis, Congenital 1084

General: X-linked recessive; nearly always found in males.

Ocular: Retinal splitting usually occurs in the nerve fiber layer; slow progression; frequently affects the macula; associated with vitreous hemorrhage; strabismus; nystagmus; retinal folds radiating from the fovea; macular pigmentary mottling; retinal detachment (rare complication).

Clinical: None.

Condon GP, et al. Congenital hereditary (juvenile X-linked) retinoschisis. Histopathologic and ultrastructural findings in 3 eyes. *Arch Ophthalmol* 1986; 104:576.

Hirose T. Retinoschisis. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:1071-1084.

Regillo CD, et al. Surgical management of complications associated with X-linked retinoschisis. *Arch Ophthalmol* 1993; 111:1080-1086.

Retinoschisis of Fovea 1085

General: Autosomal recessive.

Ocular: Foveal dystrophy; rod-cone dystrophy; nyctalopia; hyperopia; paramacular tapetal sheen reflex.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Noble KG, et al. Familial foveal retinoschisis associated with rod-cone dystrophy. *Am J Ophthalmol* 1978; 85: 551-557.

Retrolental Fibroplasia (RLF; Retinopathy of Prematurity) 1086

General: Bilateral disease seen primarily in premature infants with immature retinal vessels; excessive use of oxygen responsible for the majority of cases, but disease is seen despite oxygen restrictions or even when no oxygen supplementation is used; known factors that correlate with degrees of retinopathy of prematurity are low birth weight, short gestational age, length of time with supplemental oxygen, length of time on a mechanical ventilator; role of excessive light in newborn nurseries also has been proposed.

Ocular: Anterior or posterior synechiae; neovascularization of iris; pallor of optic disk; dragged disk; attenuated vessels; retinal detachment; dilation of veins; retinal folds; retinal hemorrhage; retrolental mass; vascular tortuosity; vasoconstriction of retina; retinal pigmentary changes; vitreous haze; vitreous traction; vitreous hemorrhages; cataract; glaucoma; leukocoria; myopia; shallow anterior chamber; opaque retrolental membrane; ciliary body drawn anteriorly; ciliary process around dilated pupil; absent pupillary reflexes; keratoconus; associated strabismus; amblyopia.

Clinical: Low birth weight; prematurity.

Mosley MJ, et al. Light toxicity and the neonatal eye. *Clin Vis Sci* 1988; 3:75-82.

Patz A. Retrolental fibroplasia. In: Ryan SJ Jr, Smith RE, eds. *Selected Topics on The Eye in Systemic Disease.* New York: Grune & Stratton, 1974:15-43.

Payne JW, Patz A. Fluorescein angiography in retrolental fibroplasia. *Int Ophthalmol Clin* 1975; 17:121-135.

Petersen RA, Hunter DG, Mukai S. Retinopathy of prematurity. In: Albert DM. Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. II.* Philadelphia: WB Saunders, 1994:2799-2812.

Retroparotid Space Syndrome (Villaret Syndrome; Posterior Retroparotid Space Syndrome) 1087

General: Lesions (traumatic, inflammatory, tumors) involving cranial nerves IX to XII and the cervical sympathetic (see Jugular Foramen Syndrome [Vernet Syndrome]).

Ocular: Enophthalmos; ptosis; lagophthalmos; epiphora; miosis; may produce sympathetic overactivity resulting in increased sympathetic outflow (i.e., pupillary dilation, widened palpebral fissure, and facial sweating).

Clinical: Homolateral paralysis cranial nerves IX to XII, with dysphagia and loss of taste in posterior third of the tongue; dysphonia; paralysis of sternocleidomastoid and trapezium muscles; paralysis cranial nerve VII occasionally.

Garrett D Jr, et al. Villaret's syndrome: a report of two cases. *Surg Neurol* 1993; 39:282-285.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Villaret M. Le Syndrome Nerveux de l'Espace Retro-parotidien Posterieur. *Rev Neurol* 1916; 23:188.

Reye Syndrome (Acute Encephalopathy Syndrome) 1088

General: Etiology unknown, although some relation to ingestion of aspirin with febrile illnesses, especially varicella and influenza, have been reported; both sexes; onset 6 months to 10 years; acute metabolic encephalopathy largely affecting children and adolescents; pathogenesis is controversial, although there is new evidence for a generalized defect in intramitochondrial enzyme processing resulting in lowered ratio of adenosine triphosphate to adenosine diphosphate.

Ocular: Cortical blindness; dilated pupils with absent or sluggish reaction to light; papilledema.

Clinical: Respiratory infections with recovery between 3 and 21 days; vomiting after recovery from infection; dyspnea; hypotonia; coma; convulsions; fever; flexion of elbows and hands.

Brown AK, et al. Aspirin and Reye syndrome. *J Pediatr* 1983; 102:157-158.

Glasgow JF, Moore R. Current concepts in Reye's syndrome. *Br J Hosp Med* 1993; 50:599-604.

Hoekelman RA. Take two aspirin and call me in the morning: salicylate use and Reye's syndrome. *Am J Dis Child* 1982; 136:973-974.

Massey JY, et al. Ocular manifestations of Reye syndrome. *Arch Ophthalmol* 1974; 91:441.

Reye RDK, Morgan G. Baral J. Encephalopathy and fatty degeneration of the viscera: a disease entity in childhood. *Lancet* 1963; 2:749.

van Coster RN, et al. Adult Reye's syndrome: a review with new evidence for a generalized defect in intramitochondrial enzyme processing. *Neurology* 1991; 41:1815-1821.

Rhabdomyosarcoma 1089

General: Most common malignant orbital neoplasm of childhood; usually occurs before age 10 years; more commonly seen in males; rarely may develop in adults; shows evidence of striated muscle differentiation; has been divided into three histopathologic types: embryonal, alveolar, and pleomorphic.

Ocular: Choroidal folds; corneal edema; exposure keratitis; rhabdomyosarcoma of orbit or extraocular muscles; decreased motility; proptosis; papilledema; orbital edema; enlarged optic foramen; erosion of bony walls of orbit; pupil irregularity; epiphora; glaucoma; visual loss; nasolacrimal duct obstruction; conjunctival mass.

Clinical: Metastasis to the lymph system, bone marrow, and lungs; headaches.

Abramson DH, et al. The treatment of orbital rhabdomyosarcoma with irradiation and chemotherapy. *Ophthalmology* 1979; 86:1330-1335.

Baron EM, et al. Rhabdomyosarcoma manifesting as acquired nasolacrimal duct obstruction. *Am J Ophthalmol* 1993; 115:239-242.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 4th ed. Philadelphia: WB Saunders, 2000.

Knowles DM II, et al. Ophthalmic striated muscle neoplasms. *Surv Ophthalmol* 1976; 21:219-261.

RHE Syndrome (Retino-Hepato-Endocrinologic Syndrome) 1090

General: More common in females.

Ocular: Total color blindness; thinning of retinal vessels with atrophy; optic disk pallor; poor photopic vision; progressive cone dystrophy.

Clinical: Degenerative liver disease; endocrine dysfunction; hypothyroidism; diabetes mellitus; repeated abortions or infertility; elevated blood creatine phosphokinase.

Berg K, et al. Familial syndrome of progressive cone dystrophy, degenerative liver disease and endocrine dysfunction. III. Genetic studies. *Clin Genet* 1978; 13:190-200.

Hansen E, et al. Familial syndrome of progressive cone dystrophy, degenerative liver disease and endocrine dysfunction. *Clin Genet* 1978; 13:119.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Rheumatic Fever 1091

General: Streptococcal infection; mechanisms may involve immune cross-reactivity between bacterial heat shock proteins and similar proteins in normal human tissues.

Ocular: Lid edema; characteristic arborizations of conjunctiva; subconjunctival hemorrhages; episcleritis; scleritis; tenonitis; uveitis; retinal detachment; central retinal artery occlusion; optic neuritis.

Clinical: Hematuria; proteinuria; proliferative glomerulonephritis; microembolization; calcific emboli; sore throat; fever; chorea; erythema marginatum; disseminated encephalomyelitis.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Munn R, et al. Acute encephalomyelitis: extending the neurological manifestations of acute rheumatic fever? *Neuro-pediatrics* 1992; 23:196-198.

Pantel GA, et al. Monocular blindness secondary to calcific embolization: an unusual presentation of rheumatic mitral valvular disease. *Chest* 1976; 69:555-556.

Smiley JD, Hoffman WL. The role of infections in the rheumatic diseases: molecular mimicry between bacterial and human stress proteins? *Am J Med Sci* 1991; 301:138-149.

Rheumatoid Arthritis (Adult) 1092

General: Systemic disease of unknown cause; more common in women (3:1); thought to have a strong autoimmune pathogenesis with positive immunoglobulins M, G, and A directed against the Fc portion of immunoglobulin G.

Ocular: Sjögren syndrome; episcleritis; scleritis; keratitis; corneal ulcers; corneal perforation; uveitis; motility disorders; dry eyes; posterior scleritis (rare).

Clinical: Synovitis; stiffness; swelling; cartilaginous hypertrophy; joint pain; fibrous ankylosis; malaise; weight loss; vasomotor disturbance.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Sainz de la Maza M, et al. Scleritis associated with systemic vasculitic diseases. *Ophthalmology* 1995; 102:687-692.

Rhinoscleroma (Klebsiella Rhinoscleromatis) 1093

General: Chronic granulomatous disease; Gram-negative bacillus; cicatricial deformities; chronic progressive granulomatous infection of the upper airways caused by the bacterium *Klebsiella rhinoscleromatis.*

Ocular: Conjunctivitis; chronic dacryocystitis; lid inflammation.

Clinical: Granulomas affecting nose and upper respiratory tract causing sclerosis and deformities; airway obstruction; leprosy; paracoccidioidomycosis; sarcoidosis; basal cell carcinoma; Wegener granulomatosis; also may occur in immunocompromised human immunodeficiency virus patients.

Andraca R, et al. Rhinoscleroma: a growing concern in the United States? Mayo Clinic experience. *Mayo Clin Proc* 1993; 68:1151-1157.

Bordley JE, et al. *Ear nose and throat disorders in children.* New York: Raven Press, 1986.

Duke-Elder S, MacFaul PA, eds. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Paul C, et al. Infection due to Klebsiella rhinoscleromatis III two patients with human immunodeficiency virus. *Clin Infect* Vis 1993; 16:441-442.

Rieger Syndrome (Axenfeld-Rieger Syndrome; Dysgenesis Mesodermalis Corneae et Irides; Dysgenesis Mesostromalis; Axenfeld Posterior Embryotoxon-Juvenile Glaucoma) 1094

General: Autosomal dominant; neural crest abnormality; 50% of patients develop glaucoma.

Ocular: Microphthalmia; congenital glaucoma; iris hypoplasia; deformed and acentric pupil; anterior synechiae; aniridia; microcornea; corneal opacities in Descemet membrane parallel to the limbus; dislocated lens; optic atrophy; cataract; strabismus; ptosis; hypertelorism; keratoconus; posterior embryotoxon; broad iris processes to embryotoxon; iris stromal hypoplasia; corectopia; polycoria; secondary glaucoma.

Clinical: Face wide; hypodontia; underdeveloped maxilla; teeth deformities; myotonic dystrophy; facial anomalies: maxillary hypoplasia, protrusion of the lower lip, broad, flat nose; dental anomalies include absent teeth, piglike incisors, and decreased crown size; hypospadias.

Eagle RC. Congenital, developmental and degenerative disorders of the iris and ciliary body. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. I.* Philadelphia: WB Saunders, 1994:367-387.

Montes JG, Montes JCG. Syndrome de Rieger, Anomalie de Axenfeld con Glaucoma Juvenil Familiar. *Arch Soc Ophth Hisp Am* 1967; 27:93.

Rieger H. Beitrage zur Kenntnis seltener Missbildungen der Iris. *Graefes Arch Clin Esp Ophthalmol* 1935; 133:602.

Wesley RK, et al. Rieger's syndrome: oligodontia and primary mesodermal dysgenesis of the iris - *Clinical* features and report of an isolated case. *J Pediatr Ophthalmol Strabismus* 1978; 15:67-70.

Rift Valley Fever 1095

General: Acute viral infection transmitted by mosquito that occurs in regions of Africa.

Ocular: Central serous retinitis; central scotoma; macular scarring; retinal vascular occlusion; uveitis; retinitis; choroiditis.

Clinical: Fever; chills; severe headache; muscle and joint pain; leukopenia; bradycardia.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Schirire L. Macular changes in Rift Valley fever. *S Afr Med J* 1951; 25:926-930.

Weiss KE. Rift Valley fever: a review. *Bull Epizoot Dis Afr* 1957; 5:431-458.

Wilson ML. Rift Valley fever ecology and the epidemiology of disease emergence. *Ann NY Acad* Sci 1994; 740: 169-180.

Yoser SL, et al. Systemic viral infections and their retinal and choroidal manifestations. *Surv Ophthalmol* 1993; 37:313-352.

Riley-Day Syndrome (Congenital Familial Dysautonomia) 1096

General: Autosomal recessive; occurs in Ashkenazi Jewish population; impaired catechol metabolism; manifested in first few days of life; characterized by developmental loss of neurons from the sensory and autonomic nervous systems.

Ocular: Congenital failure of tear production; corneal anesthesia; neuroparalytic keratitis; keratitis sicca; corneal ulcers; optic atrophy.

Clinical: Excessive salivation; failure to thrive; recurrent respiratory infections; diarrhea; insensitivity to pain; spontaneous fractures; pandysautonomia; orthostatic hypotension; gastrointestinal paresis; decreased fungiform papillae on the tongue.

Blumenfeld A, et al. Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. *Nat Genet* 1993; 4:160-164.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Howard RO. Familial dysautonomia (Riley-Day syndrome). *Am J Ophthalmol* 1967; 64:392.

Riley CM, et al. Central autonomic dysfunction with defective lacrimation: report of 6 cases. *Pediatrics* 1949; 3:468.

Rizzo JF, et al. Optic atrophy in familial dysautonomia. *Am J Ophthalmol* 1986; 102:463-467.

Thomas PK. Autonomic involvement in inherited neuropathies. *Clin Auton Res* 1992; 2:51-56.

Riley-Smith Syndrome (Bannayan-Riley-Ruvalcaba Syndrome) 1097

General: Etiology unknown; possibly heterozygous condition (single autosomal gene); macrocephaly-pseudopapilledema-multiple hemangiomas; noted at birth; has been proposed that there is overlap with the Bannayan-Zonana syndrome and the Ruvalcaba-Myhre syndrome; autosomal dominant inherited condition.

Ocular: Pseudopapilledema; prominent Schwalbe line; visible corneal nerves; ocular hypertelorism; strabismus; amblyopia.

Clinical: Macrocephaly; subcutaneous hemangiomas; pulmonary infections; mild-to-severe mental retardation; skin hemangiomas; hypotonus; gastrointestinal polyps; pigmented maculae on the skin.

Gorlin RJ, et al. Bannayan-Riley-Ruvalcaba syndrome. *Am J Med Genet* 1992; 44:307-314.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Riley HD, Smith WR. Macrocephaly, pseudopapilledema and multiple hemangiomata. *Pediatrics* 1960; 26:293-300.

Ring Chromosome 6 (Aniridia, Congenital Glaucoma, and Hydrocephalus) 1098

General: Rare disorder associated with various congenital anomalies; autosomal dominant with recessive sporadically reported.

Ocular: Microphthalmia; aniridia; congenital uveal ectropion; Rieger anomaly; congenital glaucoma; corneal clouding; prominent Schwalbe line with attached iris strands; hypopigmented fundi; hypoplasia of iris stroma; strabismus; ptosis; nystagmus; megalocornea; iris coloboma; optic atrophy; hypertelorism; antimongoloid slant of palpebral fissures; ectopic pupils; angle anomalies; posterior embryotoxon; microcornea; colobomatous.

Clinical: Hydrocephalus; agenesis of corpus callosum; congenital heart defects; mental retardation; low-set malformed ears; broad nasal bridge; micrognathia; short neck; hand anomalies; high-arched palate; widely spaced nipples; deformity of feet; respiratory distress syndrome; hyperbilirubinemia; hypocalcemia; anemia; seizure; bulging anterior fontanelle.

Bateman JB. Chromosomal anomalies and the eye. In: Wright KW, ed. *Pediatric Ophthalmology and Strabismus*. St. Louis: Mosby, 1995:595.

Chitayat D, et al. Ring chromosome 6: report of a patient and literature review. *Am J Med Genet* 1987; 26:145-151.

Deluise UP, Anderson DR. Primary infantile glaucoma. *Surv Ophthalmol* 1983; 28:1-19.

Levin H, et al. Aniridia, congenital glaucoma and hydrocephalus in a male infant with ring chromosome 6. *Am J Med Genet* 1986; 25:281-287.

Ring D Chromosome 1099

General: Variant of the chromosome 13 deletion syndrome; ring chromosomes.

Ocular: Hypertelorism; epicanthal folds; ptosis; microphthalmos; uveal colobomas; abnormal palpebral fissure; strabismus; retinoblastoma.

Clinical: Aplasia of thumbs; mental and physical retardation; trigonocephaly; malrotation of ear; micrognathia; hypoplastic nipple; widely spaced first and second toes.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981:711.

Ring Dermoid Syndrome 1100

General: Autosomal dominant; usually bilateral.

Ocular: Dermoid choristoma; conjunctival plaques of keratinization; corneal lipid deposition; irregular corneal astigmatism; amblyopia; concomitant strabismus.

Henkind P, et al. Bilateral corneal dermoids. *Am J Ophthalmol* 1973; 76:972-977.

Mattos J, et al. Ring dermoid syndrome. *Arch Ophthalmol* 1980; 98: 1059-1 061.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Oakman JH Jr, et al. Corneal dermoid: case report and review of classification. *J Pediatr Ophthalmol Strabismus* 1993; 30:388-391.

Rinx Disease (VSX1 Mutation) 1101

General: Mutation of the homeobox transcription factor gene VSX1 (RINX**)** Wide interpupillary distance; hypertelorism; anomalies of corneal endothelium; abnormal cone bipolar cells

Clinical: Empty sella turcica; posterior fossa cyst; anterior encephalocele; hypertelorism; hydrocephalus

Mintz-Hittner HA, Semina EV, Frishman LJ, et al. VSX1 (RINX) mutation with craniofacial anomalies, empty sella, corneal endothelial changes and abnormal retinal and auditory bipolar cells. *Ophthalmology* 2004; 111: 828-836.

Roberts Pseudothalidomide Syndrome 1102

General: Rare autosomal recessive disorder characterized by prenatal and postnatal growth retardation, limb defects, and craniofacial anomalies.

Ocular: Cataracts; glaucoma; microcornea; corneal clouding.

Clinical: Patients usually do not survive past I month; patients often are mentally retarded.

Holden KR, Jabs EW, Sponseller PD. Roberts/pseudothalidomide syndrome and normal intelligence: approaches to diagnosis and management. *Dev Med Child Neural* 1992; 34:534-539.

Mandal AK, Singh AP. Roberts pseudothalidomide syndrome. *Arch Ophthalmol* 2000; 118: 1462-1463.

Otano L, Matayoshi T, Gadow EC. Roberts syndrome: first-trimester prenatal diagnosis. *Prenat Diagn* 1996; 16: 770-771.

Van Den Berg DJ, Francke U. Roberts syndrome: a review of 100 cases and a new rating system for severity. *Am J Med Genet* 1993; 47:1104-1123.

Robinow-Silverman-Smith Syndrome (Achondroplastic Dwarfism; Mesomelic Dwarfism; Robinow Dwarfism) 1103

General: Autosomal dominant; both sexes affected; present from birth.

Ocular: Hypertelorism; epicanthal folds.

Clinical: Dwarfism; shortened forearms; bulging forehead; depressed nasal bridge; hypoplastic mandible; small, upturned nose; micrognathia; crowded teeth; penile hypoplasia.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Robinow M. et al. A newly recognized dwarfing syndrome. *Am J Dis Child* 1969; 117:649-650.

Smith DW Recognizable patterns of human malformation. Philadelphia: WB Saunders, 1976.

Rochon-Duvigneaud Syndrome (Superior Orbital Fissure Syndrome) 1104

General: Inflammatory, traumatic, tumor, or vascular lesions such as meningioma of the sphenoid, carotid aneurysm, and arachnoiditis; infections originating in the maxillary sinus.

Ocular: Mild exophthalmos; lid edema; partial or complete ophthalmoplegia (III, IV, and VI); decreased corneal sensitivity; papilledema; optic atrophy.

Clinical: Decreased sensitivity in area of nasociliary, lacrimal, frontal, and ophthalmic nerve distribution; may result from a metastatic tumor.

Falcone F Jr, et al. Superior orbital fissure syndrome. Secondary to infected dentigerous cyst of the maxillary sinus. *NY State Dental J* 1994; 60:62-64.

Hedstrom J. et al. Superior orbital fissure syndrome: report of a case. *J Oral Surg* 1974; 32:198.

Phanthumchinda K, et al. Superior orbital fissure syndrome as a presenting symptom in hepatocellular carcinoma. *J Med Assoc Thailand* 1992; 75:62-65.

Rochon-Duvigneaud A. Quelques Cas de Paralysie de Tous les Nerfs Orbitaires (Ophtalmoplegie Totale avec Amaurose et Anesthesie dans le Domaine de l'Ophtalmique d'Origine Syphilitique). *Arch Ophthalmol* 1896; 16:746.

Rocky Mountain Spotted Fever 1105

General: Acute systemic disease caused by *Rickettsia rickettsii* transmitted by a wood tick or dog tick.

Ocular: Conjunctivitis; optic atrophy; cotton-wool spots; scotoma; uveitis; optic neuritis; paralysis of accommodation; paralysis of extraocular muscles; retinal vascular occlusion; vitreal opacity; hypopyon; anterior uveitis with fibrin clots.

Clinical: Fever; chills; headache; muscle aches; rash.

Burton TC, Smith TW. The retinal manifestations of Rocky Mountain spotted fever. *Am J Ophthalmol* 1977; 84:259.

Duffey RJ, Hammer ME. Ocular manifestations of Rocky Mountain spotted fever. *Ann Ophthalmol* 1987; 19: 301-306.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

O'Brien JM, Albert DM, Foster SC. Anterior uveitis. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. I.* Philadelphia: WB Saunders, 1994:419.

Presley GD. Fundus changes in Rocky Mountain spotted fever. *Am J Ophthalmol* 1969; 67:263-267.

Rollet Syndrome (Orbital Apex-Sphenoidal Syndrome) 1106

General: Lesion in the apex of the orbit (neoplastic, hemorrhagic, or inflammatory) involving the third, fourth, and sixth cranial nerves, the ophthalmic branch of the fifth sympathetic fibers when they pass through the sphenoidal fissure, and the optic nerve; manifestations vary greatly with extent of lesion; pain is frequent early sign; orbital fissure syndrome and sphenocavernous syndrome are similar; sudden onset.

Ocular: Exophthalmos; ptosis; hyperesthesia or anesthesia of the upper lid; ophthalmoplegia (partial or complete); wide pupil with loss of reaction on accommodation; neuralgic pain in the region of the ophthalmic branch of cranial nerve V; anesthesia of the cornea; papilledema; optic neuritis; optic atrophy; diplopia; herpes zoster ophthalmicus.

Clinical: Hyperesthesia or anesthesia of the forehead; inflammation of cavernous sinuses; meningoencephalitis.

Bourke RD, Pyle J. Herpes zoster ophthalmicus and the orbital apex syndrome. *Aust N Z J Ophthalmol* 1994; 22:77-80.

Goldberg RA, et al. Microanatomy of the orbital apex. Computed tomography and microcryoplaning of soft and hard tissue. *Ophthalmology* 1992; 99:1447-1452.

Kander PL, Leaver PK. Orbital apex syndrome complicating frontal pyocele. *J Laryngol Otol* 1974; 88:551.

Kattaah JC, Kennerdell JS. Orbital apex syndrome secondary to herpes zoster ophthalmicus. *Am J Ophthalmol* 1978; 85:378-382.

Rollet J. Dict Traite des Maladies Veneriennes, Vol. 15. Paris: Masson, 1865:993.

Romberg Syndrome (Parry-Romberg Syndrome; Progressive Hemifacial Atrophy; Progressive Facial Hemiatrophy; Facial Hemiatrophy) 1107

General: Autosomal dominant; irritation in the peripheral trophic sympathetic system; onset in the second decade; both sexes affected.

Ocular: Enophthalmos; outer canthus lowered; absence of nasal portion of eyebrow; ptosis; paresis of ocular muscles; miosis; iritis; iridocyclitis; heterochromia iridis; keratitis; neuroparalytic keratitis; cataracts; choroiditis; Fuchs heterochromic cyclitis; retinal vascular abnormalities; association with Coats syndrome and exudative stellate neuroretinopathy; scleral melting.

Clinical: Atrophy of soft tissue on one side of the face, including tongue; trigeminal neuralgia and/or paresthesia; alopecia and poliosis not uncommon.

Aracena T, et al. Progressive hemifacial atrophy (Parry-Romberg syndrome): report of two cases. *Ann Ophthalmol* 1979; 11:953-958.

Gass JD, et al. Exudative stellate neuroretinopathy and Coats' syndrome in patients with progressive hemifacial atrophy. *Eur J Ophthalmol* 1991; 1:2-10.

Hoang-Xuan T, et al. Romberg's progressive hemifacial atrophy: an association with scleral melting. *Cornea* 1991; 10:361-366.

La Hey E, Baarsma GS. Fuchs' heterochromic cyclitis and retinal vascular abnormalities in progressive hemifacial atrophy. *Eye* 1993; 7[Part 3]:426-428.

Parry CH. *Collections from unpublished papers, vol. I.* London: Underwood, 1825.

Romberg MH. *Trophoneurosen, Klin. Ergebn.* Berlin: A. Forster, 1846.

Rosenberg-Chutorian Syndrome 1108

General: Recessive inheritance; inheritance is considered X-linked semidominant.

Ocular: Optic atrophy.

Clinical: Polyneuropathy; neural hearing loss.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Konigsmark BW, et al. Dominant congenital deafness and progressive optic nerve atrophy. *Arch Ophthalmol* 1974; 91:99-103.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Rosenberg RN, Chutorian A. Familial opticoacoustic nerve degeneration and polyneuropathy. *Neurology* 1967; 17: 827-832.

Rosenthal-Kloepfer Syndrome 1109

General: Autosomal dominant; unilateral or bilateral; onset in early childhood; hyperplasia and furrowing of skin, especially of face and scalp, beginning about the fourth decade of life; etiology unknown; affects both sexes.

Ocular: Prominent lateral aspect of the supraorbital arch of the frontal bone; corneal leukomas (initially superficial stromal infiltrations that develop to thick and dense corneal opacities).

Clinical: Acromegaly; thickening of the bony skull; prominent lower jaw; skin changes (with verticis gyrata); abnormal ridges and creases of the palms.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Rosenthal JW, Kloepfer HW An acromegaloid, cutis verticis gyrata, corneal leukoma syndrome. A new medical entity. *Arch Ophthalmol* 1962; 68:722.

Rothmund Syndrome (Rothmund-Thomson Syndrome; Telangiectasia-Pigmentation-Cataract Syndrome; Ectodermal Syndrome; Congenital Poikiloderma with Juvenile Cataract) 1110

General: Autosomal recessive; more common in females (2:1); Werner syndrome in adults has certain similarities to this syndrome; inflammatory phase progresses to atrophy and telangiectasia; onset at age 3 to 6 months.

Ocular: Eyebrows may be sparse or absent; hypertelorism; cilia sometimes are diminished or absent; trichiasis; epiphora; cataracts (anterior subcapsular, posterior stellate, or perinuclear type); corneal lesions; retinal hyperpigmentation; keratoconus; strabismus; epibulbar dermoids.

Clinical: Poikiloderma; hypogonadism; hypomenorrhea; head deformity (enlarged with depressed nasal bridge as well as microcephaly); small stature, with short or malformed distal phalanges; aplasia cutis congenita (congenital absence of skin in one or more areas); alopecia.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Evers ME, et al. Oculo-ectodermal syndrome: a new case. *Am J Med Genet* 1994; 53:378-379.

Gardner J, Viljoen D. Aplasia cutis congenita with epibulbar dermoids: further evidence for syndromic identity of the ocular ectodermal syndrome. *Am J Med Genet* 1994; 53:317-320.

Rothmund A. Uber Katarakte in Verbindung mit einer eigentumlichen Hautdegeneration. *Graefes Arch Clin Exp Ophthalmol* 1868; 14:159.

Sri-Skandu-Rajah-Sivayoham I, Ratnaike VT. Rothmund-Thomson syndrome in an oriental patient. *Ann Ophthalmol* 1975; 7:417.

Thomson MS. A hitherto undescribed familial disease. *Br J Dermatol* 1923; 35:455.

Roussy-Cornil Syndrome 1111

General: Etiology unknown; sporadic hypertrophic neuropathy; onset in second to third decade of life or later; progressive; occasionally, remissions and exacerbations.

Ocular: Sluggish pupils; disturbed vision.

Clinical: Nerves palpable and tender; peripheral atrophy; tendon reflexes diminished or abolished; scoliosis; muscle fasciculations; ataxia; lancinating pains.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Roussy G, Cornil L. Nevrite Hypertrophique Progressive Non-Familiale de l'Adulte. *Ann Med* 1919; 6:296-305.

Suarez GA, et al. Localized hypertrophic neuropathy. *Mayo Clin Proc 1994;69:747-748.*

Roy Syndrome 1112

General: Long history of smoking tobacco.

Ocular: Cataract, usually unilateral, posterior subcapsular, and cortical.

Clinical: Emphysema; cardiac disorder; cancer.

Roy FH. Cigarette smoking and the risk of cataract [Letter]. *JAMA* 1993; 269:748.

Roy Syndrome II 1113

General: Long history of smoking tobacco.

Ocular: Cataract; nuclear cataract may be cortical or posterior subcapsular cataract.

Clinical: Emphysema; cardiac disorder; cancer.

Klein RE, et al. Cigarette smoking and lens opacities. The Beaver Dam Eye Study. *Am J Prev Med* 1993; 9:27-30.

Roy FH. Cigarette smoking and the risk of cataract [Letter]. *JAMA* 1993; 269:748.

Rubella Syndrome (Congenital Rubella Syndrome; German Measles; Gregg Syndrome) 1114

General: Rubella infection of the mother during first trimester of pregnancy; ocular disease is the most commonly found abnormality in patients with congenital rubella syndrome (75%), multiorgan disease is common (greater than 75%); no significant association has been found between gestational age and time of maternal infection and incidence of individual ocular conditions.

Ocular: Nystagmus; glaucoma; corneal haziness; cataracts; retinal pigmentary changes; appearance and central distribution of lesions are quite distinguishable from retinitis pigmentosa; retinopathy is not progressive and has little, if any, effect on vision; waxy atrophy of optic disk; conjunctivitis; megalocornea or microcornea; buphthalmos; microphthalmos; uveitis; iris atrophy; spherophakia; strabismus.

Clinical: Low birth weight; diarrhea; pneumonia; urinary infection; hearing loss; heart disease; hepatosplenomegaly; mental retardation; inguinal hernias; ataxia; cardiac abnormalities.

Boger WP III, Petersen RA. Congenital rubella syndrome. In: Pavan-Langston D, ed. *Manual of ocular diagnosis and therapy*, 4th ed. New York: Little, Brown and Company, 1995:266-267,299.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Givens KT, et al. Congenital rubella syndrome: ophthalmic manifestations and associated systemic disorders. *Br J Ophthalmol* 1993; 77:358-363.

Roy FH, et al. Congenital rubella syndrome. *Am J Ophthalmol* 1966; 62:222.

Wolff SM. The ocular manifestations of congenital rubella. *Trans Am Ophthalmol Soc* 1972; 70:577-614.

Rubinstein-Taybi Syndrome 1115

General: Inheritance polygenic or multifactorial; rare.

Ocular: Antimongoloid slant of lid fissure; epicanthus; long eyelashes and highly arched brows; strabismus; myopia; hyperopia; iris coloboma; cataract; optic atrophy; ptosis; retinal detachment.

Clinical: Motor and mental retardation; broad thumbs and toes; highly arched palate; allergies; heart murmurs; anomalies of size, shape, and position of ears; dwarfism; cryptorchidism.

Critchley LA, Gin T, Stuart JC. Anaesthesia in an infant with Rubinstein-Taybi syndrome. *Anaesthesia* 1995; 50:37-38.

Larned DC, et al. Association of congenital ptosis and congenital heart disease. *Ophthalmology* 1986; 93:492-494.

Marcus-Harel T, et al. Retinal detachment with high myopia in the Rubinstein-Taybi syndrome. *Metab Pediatr Syst Ophthalmol* 1991; 14:53-56.

Maurer I, Regensburger D, Bernhard A. Aortic valve reconstruction in Rubinstein- Taybi syndrome: the valuable aid of transesophageal echocardiography. *J Cardiovasc Surg (Torino)* 1991; 32:327-329.

Roy FH, et al. Ocular manifestations of the Rubinstein-Taybi syndrome. *Arch Ophthalmol* 1968; 79:272.

Rubinstein JH, Taybi H. Broad thumbs and toes and facial abnormalities. *Am J Dis Child* 1963; 105:588.

Rud Syndrome 1116

General: Etiology unknown; immature nerve cells and decreased number of cells; Betz cells in the motor cortex show chronic chromatolysis; excess of oligodendroglia in the frontal cortex; relationship to tuberous sclerosis and neurofibromatosis; X-linked inheritance has been reported.

Ocular: Retinal pigmentary degeneration.

Clinical: Epilepsy; infantilism; idiocy; congenital ichthyosis; muscular atrophy; male hypogonadism.

Duke-Elder S, ed. *System of Ophthalmology, vol. III. part* 2. St. Louis: CV Mosby, 1976:1130.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Rud E. Et Tilfaelde af Infantilsms med Tetani, Epilepsy, Polyneuritis, Ichthiosis og Anaemi of Pernicios Type. *Hospitalstidende* 1927; 70:525.

Wisniewski K, et al. X-linked inheritance of the Rud syndrome. *Am J Hum Genet* 1985; 37:A83.

Russell Syndrome 1117

General: Onset between 3 months and 2 years; caused by tumors of the anterior portion of the thalamus (usually astrocytoma), optic chiasm, midcerebellar region, and midline ependymoma; erosion under the anterior clinoid processes that causes a characteristic J-shaped sella in lateral skull films.

Ocular: Lid retraction; nystagmus (horizontal, vertical, or rotatory); homonymous hemianopsia; optic nerve atrophy.

Clinical: Extreme emaciation; euphoria; pale skin.

Ciccarelli EC, Huttenlocher PR. Diencephalic tumor: a cause of infantile nystagmus and cachexia. *Arch Ophthalmol* 1967; 78:350.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Russell A. A diencephalic syndrome: emaciation in infancy and childhood. *Arch Dis Child* 1951; 26:274.

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Sabin-Feldman Syndrome 1118

General: Etiology unknown; similar to toxoplasmosis; results of toxoplasma dye and complement fixation tests are negative; onset in early infancy.

Ocular: Microphthalmia; strabismus; fixed pupils; posterior lenticonus; microcornea; chorioretinitis or atrophic degenerative chorioretinal changes; optic atrophy.

Clinical: Cerebral calcifications (infrequent); convulsions (frequent); microcephaly; hydrocephalus.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Sabin AB, Feldman HA. Chorioretinopathy associated with other evidence of cerebral damage in childhood: a syndrome of unknown etiology separable from congenital toxoplasmosis. *J Pediatr* 1949; 35:296.

Saint Anthony Fire (Erysipelas) 1119

General: Acute localized inflammation of the skin and subcutaneous tissue; erysipelas is a febrile infection of the skin and subcutaneous tissue, most commonly caused by *Streptococcus,* characterized by the acute onset of a red, indurated expanding plaque that nearly disappears with the use of antibiotics; sometimes caused by *Staphylococcus.*

Ocular: Conjunctivitis; blepharitis; elephantiasis and gangrene of lid; ptosis; dacryocystitis; cellulitis of orbit; keratitis; panophthalmitis; uveitis; eyelid involvement.

Clinical: Edema; fever; rigor; vesicles; tenderness; headache; vomiting; localized pain.

Binford RT Jr, Lindo SD. Dermatologlc conditions affecting the eye. In: Dunlap EA, ed. *Gordon's medical management of ocular disease,* 2nd ed. New York: Harper & Row, 1976:91-110.

Bratton RL, Nesse RE. St. Anthony's fire: diagnosis and management of erysipelas. *Am Fam Physician* 1995; 51: 401-404.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

McHugh D, Fison PN. Ocular erysipelas. *Arch Ophthalmol* 1992; 110: 1315.

Saldino-Mainzer Syndrome 1120

General: Autosomal recessive; Leber congenital amaurosis associated with familial juvenile nephronophthisis and cone-shaped epiphysis of the hands; similar to Senior-Loken syndrome, with the difference being cone-shaped epiphysis.

Ocular: Tapetoretinal degeneration; retinal atrophy; Leber congenital amaurosis; retinitis pigmentosa.

Clinical: Nephronophthisis; cone-shaped epiphyses of hands and feet; flared ribs; hypoplastic pelvis; brachydactyly; hyperparathyroidism; osteomalacia; osteopetrosis; renal failure.

Ellis DS, et al. Leber's congenital amaurosis associated with familial juvenile nephronophthisis and cone-shaped epiphysis of the hands. *Am J Ophthalmol* 1984; 97:233-239.

Foxman SG, et al. Classification of congenital and early onset retinitis pigmentosa. *Arch Ophthalmol* 1985; 103: 1502-1507.

Sandhoff Disease (Gangliosidosis Type 2 [G_{M2}]) 1121

General: Hereditary cerebromacular degeneration-sphingolipidoses; onset by age 6 months; autosomal recessive inheritance; enzyme defect; caused by deficiency of hexosaminidases A and B; defect localized to chromosome 5 (5q13).

Ocular: Cherry-red spot of macula; visual loss; clinically identical to Tay-Sachs disease.

Clinical: Motor retardation; doll-like facies; dementia; hyperacusis; frequent respiratory infections; variable hepatosplenomegaly.

Brownstein S, et al. Sandhoff's disease (GM2 gangliosidosis type 2). Histopathology and ultrastructure of the eye. *Arch Ophthalmol* 1980; 98:1089-1097.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Messer G, et al. Ultrastructure of the conjunctiva, skin and gingiva: a case of Sandhoff's disease in a Jewish patient. *Arch Pathol Lab Med* 1980; 104:123-129.

Nrby S, et al. Retinal and cerebellar changes in early fetal Sandhoff disease (GM2-gangliosidosis type 2). *Metab Pediatr Ophthalmol* 1980; 4:115-119.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV*. Philadelphia: WB Saunders, 1994:3102-3120.

Sandifer Syndrome (Hiatal Hernia-Torticollis Syndrome) 1122

General: Inheritance not known; males affected; hiatal hernia.

Ocular: Strabismus (not related to existing torticollis).

Clinical: Rotation of the head to one shoulder with stretching of the neck (more pronounced during eating and reading); epigastric pain associated with vomiting, primarily in infancy; malnutrition; hiatal hernia; asthenia.

Kinsbourne M. Hiatus hernia with contortions of the neck. *Lancet* 1964; 1:1058.

O'Donnell JJ, Howard RO. Torticollis associated with hiatus hernia (Sandifer's syndrome). *Am J Ophthalmol* 1971; 71:1134.

Sands of the Sahara Syndrome (Diffuse Lamellar Keratitis) 1123

General: Interface inflammation after laser *in situ* keratomileusis (LASIK).

Ocular: Interface inflammation after LASIK is a rare but potential sight-threatening complication; syndrome presents 1 to 5 days after LASIK; affected patients often complain of decreased or cloudy vision, foreign body sensation, and photophobia; symptoms may be mild or severe; cause of the interface debris is unknown, but microkeratome material is implicated.

Castillo A, et al. Peripheral melt of flap after laser in situ keratomileusis. *J Cataract Refract Surg* 1998; 14:61-63.

Holland SP, et al. Diffuse lamellar keratitis related to endotoxins released from sterilizer reservoir biofilms. *Ophthalmology* 2000; 107:1227-1234.

Kaufman SC, et al. Interface inflammation after laser in situ keratomileusis-sands of the Sahara syndrome. *J Cataract Refract Surg* 1998; 24:1589-1593.

Smith RJ, Maloney RK. Diffuse lamellar keratitis. *Ophthalmology* 1998; 105:1721-1726.

Sanfilippo-Good Syndrome (Heparitinuria; Mucopolysaccharidosis III; MPS III) 1124

General: Autosomal recessive; excess urinary excretion of heparitin sulfate (see Hunter Syndrome; Hurler Syndrome; Maroteaux-Lamy Syndrome; Morquio Syndrome; Scheie Syndrome). Lack of a β-galactosaminidase-like enzyme causing accumulation of glycolipids, acid mucopolysaccharides, and their precursors; both sexes affected; death occurs by second decade in the majority of cases; autosomal recessive; divided into type A (with decreased levels of heparan sulfatase) and type B (with decreased levels of N-acetyl-α-D-glucosaminidase).

Ocular: Night blindness; slight narrowing of retinal vessels; pigment deposits in the fundi; bushy eyebrows; coarse eyelashes; acid mucopolysaccharide deposits in cornea, iris, lens, and sclera; retinal degeneration; optic nerve atrophy.

Clinical: Mental deficiency progressing to severe degrees within a few years; seizures; gargoyle features very mild; dwarfism; stiff joints; hepatosplenomegaly; hirsutism; mitral valve insufficiency.

Collins JF. *Handbook of Clinical Ophthalmology* New York: Masson, 1982.

Francois J. Ocular manifestations of the mucopolysaccharidoses. *Ophthalmologica* 1974; 169:345-361.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gills JP, et al. Electroretinography and fundus oculi findings in Hurler's disease and allied mucopolysaccharidoses. *Arch Ophthalmol* 1965; 74:596.

Muenzer J, et al. Severe mitral insufficiency in mucopolysaccharidosis type III-B (Sanfilippo syndrome). *Pediatr Cardiol* 1993; 14:130-132.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:3102-3120.

Savin Syndrome 1125

General: Congenital ichthyosis combined with urticarial manifestations.

Ocular: Nodular thickening in parenchyma of cornea.

Clinical: Dry, scaly skin; atopic dermatitis; pruritus.

Korting GW, et al. *The skin and the eye: a dermatologic correlation of diseases of the periorbital region*. Philadelphia: WB Saunders, 1973:72.

Savin LH. Corneal dystrophy associated with congenital ichthyosis and allergic manifestations in male members of a family. *Br J Ophthalmol* 1956; 40:82-89.

Scalded Skin Syndrome (Toxic Epidermal Necrolysis; Ritter Disease; Toxic Epidermal Necrolysis of Lyell; Staphylococcal Scalded Skin Syndrome; Lyell Syndrome; Epidermolysis Acuta Toxica; Toxic Epidermal Necrolysis) 1126

General: Generalized exfoliative dermatitis frequently affecting neonates and resulting from an initial focal staphylococcal infection (i.e., staphylococcal ophthalmia neonatorum); toxic epidermal necrolysis usually refers to manifestation in the adult secondary to a drug reaction but affects all ages; immunopathogenetic mechanisms probably initiated with drug-skin binding with aberrant immune responses, including complement and immunoglobulin G deposition with the epidermis and mucosa; recent reports suggest that patients with the acquired immunodeficiency syndrome (AIDS) are at higher risk for developing mucocutaneous reactions, such as toxic epidermal necrolysis; mortality rate approximately 30%.

Ocular: Necrotic areas of lids, conjunctiva, and cornea; symblepharon; loss of corneal epithelium; corneal ulcer; leukoma; perforation of globe; abolition of lacrimal secretion; conjunctival chemosis; blepharitis; entropion; periorbital swelling; trichiasis; distichiasis; fornix shortening.

Clinical: Widespread reddening and tenderness of the skin followed by the exfoliation of large areas of skin; in children, erythema starts usually around the mouth and spreads over the entire body within hours, followed by blisters and large exudative lesions; fever; shock.

Belfort R Jr, et al. Ocular complications of Stevens-Johnson syndrome and toxic epidermal necrolysis in patients with AIDS. *Cornea* 1991; 10:536-538.

Foster CS. Immunologic disorders of the conjunctiva, cornea, and sclera. In: Albert D, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. I.* Philadelphia: WB Saunders, 1994:190.

Kobayashi A, Yoshita T, Sugiyama K, et al. Amniotic membrane transplantation in acute phase of toxic epidermal necrolysis with severe corneal involvement. *Ophthalmology* 2006; 113: 126-132

Power WJ, et al. Analysis of the acute ophthalmic manifestations of the erythema multiforme/Stevens-Johnson syndrome/toxic epidermal necrolysis disease spectrum. *Ophthalmology* 1995:102:1669-1676.

Scaphocephaly Syndrome 1127

General: Craniofacial dysostoses with failure in the development of the primitive mesoderm; facial features result from premature fusion of the sagittal cranial suture; males more commonly affected (4:1).

Ocular: Shallow orbits; proptosis; nystagmus; exotropia; aniridia; cataract; papilledema; optic atrophy; aniridia; dislocated lens.

Clinical: Long anteroposterior head diameter; short transverse diameter of the head; increased intracranial pressure; flat forehead with absent superciliary arches; prominent nose; mental retardation.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Mangelschots K, et al. Reciprocal translocation between the proximal regions of the long arms of chromosomes 13 and 15 resulting in unbalanced offspring: characterization by fluorescence in situ hybridization and DNA analysis. *Hum Genet* 1992; 89:407-413.

Mohan H, Sen DK, Gupta DK. Scaphocephaly, bilateral aniridia, and bilateral anterior polar cataract with posterior dislocation of the lens. *Br J Ophthalmol* 1971; 55:642.

Schafer Syndrome (Tyrosine Transaminase Deficiency; Richer-Hanhart Syndrome; Keratosis Palmoplantaris Syndrome) 1128

General: Etiology unknown; dominant form manifested as an ectodermal dysplasia with disseminated follicular keratosis and leukokeratosis of the oral mucosa; recessive form also involves cornea; oculocutaneous syndrome; autosomal recessive phenotype associated with tyrosine transaminase deficiency.

Ocular: Dustlike, randomly distributed corneal lesions in the lower portion of the cornea; herpetoid corneal lesions; cataract; herpetiform corneal ulcers.

Clinical: Keratosis of palms and soles; pachyonychia; alopecia; microcephaly; dwarfism; oligophrenia; painful punctate keratoses of digits, palms and soles.

Beinfang DC, et al. The Richer-Hanhart syndrome: report of a case with associated tyrosinemia. *Arch Ophthalmol* 1976; 94: 1133-1137.

Grayson M. Corneal manifestations of keratosis plantaris and palmaris. *Am J Ophthalmol* 1965; 59:483.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Schafer E. Zur Lehre von den Congenitalen Dyskeratosen. *Arch Dermatol Syphil (Germ)* 1925; 148:425.

Schaumann Syndrome (Besnier-Boeck-Schaumann Syndrome; Boeck Sarcoid; Sarcoidosis) 1129

General: Etiology unknown; theories include tuberculosis, hypersensitivity to pine pollen, virus infection; affects blacks most often; chronic course with spontaneous re- missions (see Heerfordt Syndrome); hilar or paratracheal nodes with erythema nodosum known as Lofgren syndrome; onset most often in middle and old age; ocular involvement in 20% to 25% of all cases.

Ocular: Orbital granulomatous mass; bony defects; cutaneous and subcutaneous nodules; myogenic palsy; lacrimal gland adenopathy; decreased tear formation; secondary glaucoma; granulomatous uveitis with iris nodules, cells, and flare; mutton fat keratitic precipitates; keratitis sicca; vitreous floaters; band-shaped keratitis; complicated cataract; inflammatory retinal exudates; "candle wax drippings"; optic nerve atrophy; neuritis; eyelid nodules; ocular nerve enlargement (granuloma).

Clinical: Lymphadenopathy; hilar nodes; fatigue; cystic, punched-out or reticulated changes in small bones (mainly hands and feet); muscle wasting; contractures; weakness in legs and arms.

Beck AD, et al. Optic nerve enlargement and chronic visual loss. *Surv Ophthalmol* 1994; 38:555-566.

Fulton A, et al. Gastrointestinal sarcoidosis diagnosed by conjunctival biopsy. *Am J Ophthalmol* 1976; 82: 102-104.

Hall JG, Cohen KL. Sarcoidosis of the eyelid skin. *Am J Ophthalmol* 1995; 119:100-101.

Karma A. Diagnostic aspects of ocular sarcoidosis. *Sarcoidosis* 1994; 11:58-60.

Kelley JS, Green WR. Sarcoidosis involving the optic nerve head. *Arch Ophthalmol* 1973; 89:489.

Khalatbari D, Stinnett S, McCallum RM, et al. Demographic-related variations in posterior segment ocular sarcoidosis. *Ophthalmology* 2004; 111:357-362.

Mizuno K, Takahashi J. Sarcoid cyclitis. *Ophthalmology* 1986; 93:511-517.

Obenaut CD, et al. Sarcoidosis and its ophthalmic manifestations. *Am J Ophthalmol* 1978; 86:648-655.

Schaumann J. Benign lymphogranuloma and its cutaneous manifestations. *Br J Dermatol* 1924; 36:615.

Scheie Syndrome (Mucopolysaccharidosis IS; MPS IS; MPS V; Mucopolysaccharidosis V) 1130

General: Autosomal recessive; chondroitin sulfate B excreted in excess in the urine; formerly MPS V (see Hurler Syndrome; Hunter Syndrome; Sanfilippo-Good Syndrome; Morquio Syndrome; Maroteaux-Lamy Syndrome). Both sexes affected; deficiency of α-L-iduronidase; increased urinary dermatan and heparan sulfate; fibrous long-spacing collagen on histopathologic examination; least severe form of mucopolysaccharidosis.

Ocular: Night blindness; fields may show general constriction; ring scotomata; diffuse corneal haze to marked corneal clouding (progressive); bushy eyebrows; coarse eyelashes; optic atrophy; anisocoria; cataracts; proptosis; acid mucopolysaccharide deposits in the iris and sclera; tapetoretinal degeneration; glaucoma.

Clinical: Normal intelligence; broad facies; thickened joints; aortic valvular disease; psychosis; claw hand; carpal tunnel syndrome; excessive body hair; progressive juxtaarticular cystic lesions.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hamilton E, Pitt P. Articular manifestations of Scheie's syndrome. *Ann Rheum Dis* 1992; 51:542-543.

Quigley HA, Goldberg ME Scheie's Syndrome and macular corneal dystrophy: an ultrastructural comparison of conjunctiva and skin. *Arch Ophthalmol* 1971; 85:553.

Rummelt V, et al. Light and electron microscopy of the cornea in systemic mucopolysaccharidosis type I-S (Scheie's syndrome). *Cornea* 1992; 11:86-92.

Scheie HG, et al. A newly recognized form fruste of Hurler's disease (gargoylism). *Am J Ophthalmol* 1962; 53:753.

Schilder Disease (Encephalitis Periaxialis Diffusa) 1131

General: Lesions situated in the subcortical white matter (area 17); occurs in males; any age; etiology unknown; possibly toxic, infectious, or abiotrophic neural defects.

Ocular: Nystagmus; extraocular muscle palsy, either nuclear or supranuclear; hemianopsia (in occipital lobe involvement); optic nerve, chiasm, and tract involvement can lead to blindness; papilledema; optic neuritis; optic atrophy; central scotomata.

Clinical: Progressive spastic paralysis; progressive mental deterioration; irritability and peevishness; deafness if the temporal lobe becomes involved; tremor; dullness; characteristically bilateral lesions in the brain.

Francois J. Ocular manifestations in demyelinating disease. *Adv Ophthalmol* 1979; 39:1-36.

Martin JJ, Guazzi GC. Schilder's diffuse sclerosis. *Dev Neurosci* 1991; 13:267-273.

Powers JM, Schaumburg HH. Adrenoleukodystrophy (sex-linked Schilder's disease): a pathogenetic hypophysis based on ultrastructural changes in adrenal cortex, peripheral nerve and testis. *Am J Pathol* 1974; 76:481.

Schilder P. Zur Kenntnis der Sogenannten Diffusen Sklerose (Ueber Encephalitis Periaxialis Diffusa). *Z Ges Neurol Psychiatr* 1912; 10:1.

Schistosomiasis (Bilharziasis) 1132

General: Parasitic infection by *Schistosoma mansoni.*

Ocular: Hyphema; parasite in anterior chamber; granuloma of conjunctiva, lid, lacrimal gland, and orbit; uveitis; keratitis; iritis; retinitis; dacryoadenitis; cataract; posterior uveitis; choroiditis; blepharitis; optic atrophy.

Clinical: Dermatitis; fever; allergic reaction; short-lasting hepatosplenomegaly, inflammation, granuloma formation, and fibrosis; meningitis; cor pulmonale.

Conkle DM, et al. A surgical approach in the treatment of Schistosoma mansoni. *South Med J* 1976; 69:747-748.

Creed TD. Unilateral optic atrophy presence secondary to schistosomiasis of the optic nerve. *J Am Optom Assoc* 1993; 64:440-445.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Schizophrenia 1133

General: Organic brain syndrome due to degeneration or toxic, infectious, or metabolic conditions; acute or insidious onset; disturbances of thinking, mood, and behavior; etiology unknown.

Ocular: Miosis; ocular hallucinations; associated with retinitis pigmentosa in one pedigree; abnormal smooth-pursuit eye movement; visual perceptual dysfunction: low amplitude of accommodation, esophoria or exophoria, and vergence duction suppression.

Clinical: Paranoia; depression; fear; anxiety; catatonia; delusions; hallucinations; misinterpretation of reality; lack of will or enthusiasm; hypochondriasis; preoccupation with own thoughts; meaningless repetitive speech; meaningless repetitive motions; sensorineural deafness; mental retardation associated in one pedigree.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Flach F, et al. Visual perceptual dysfunction in patients with schizophrenic and affective disorders versus control subjects. *J Neuropsychiatry Clin Neurosci* 1992; 4:422-427.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology,* 4th ed. Baltimore: Williams & Wilkins, 1987.

Schlenker R, et al. Smooth-pursuit eye movement dysfunction in schizophrenia: the role of attention and general psychomotor dysfunctions. *Eur Arch Psychiatry Clin Neurosci* 1994; 244:153-160.

Sharp CW, et al. Schizophrenia and mental retardation associated in a pedigree with retinitis pigmentosa and sensorineural deafness. *Am J Med Genet* 1994; 54:354-360.

Schomberg Disease 1134

General: Blood dyscrasias; associated with thrombocytopenia.

Ocular: Subconjunctival hemorrhage.

Clinical: Petechiae may occur in any tissue; bleeding from any orifice.

Duke-Elder S, ed. *System of Ophthalmology, vol. VIII, part 1.* St. Louis: CV Mosby, 1976:33-39.

Givner I. Noninfectious conjunctival congestion. In: *Infectious Diseases of the Conjunctiva and Cornea. Symposium of the New Orleans Academy of Ophthalmology*. St. Louis: CV Mosby, 1963:48.

Schonenberg Syndrome (Dwarf-Cardiopathy Syndrome) 1135

General: Consanguinity and familial occurrence; etiology obscure.

Ocular: Blepharophimosis; epicanthal folds; pseudoptosis.

Clinical: Dwarfism (proportionate); congenital heart disease.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Schonenberg H. Uber ein Neues Kombinationsbild Multipler Abartungen. (Minderwuchs, Vitium Cordis, Beiderseitige Congenitale Ptose.) *Ann Pediatr* 1954; 182:229.

Schwartz Syndrome 1136

General: Glaucoma associated with retinal detachment; caused by inflammation of trabecula or pigment granules obstructing outflow; photoreceptor outer segments identified in the aqueous humor of patients with this syndrome are thought to playa role in the elevation of intraocular pressure.

Ocular: Secondary open-angle glaucoma; retinal detachment; uveitis; myopia; blepharophimosis; long eyelashes; microcornea.

Clinical: Small stature; myotonia; expressionless facies; joint limitation in hips; dystrophy of epiphyseal cartilage, vertical shortness of vertebrae, short neck; low hairline.

Baruch E, et al. Glaucoma due to rhegmatogenous retinal detachment: Schwartz syndrome. *Glaucoma* 1981; 3:229-230.

Harley RD, ed. *Pediatric Ophthalmology,* 4th ed. Philadelphia: WB Saunders, 1998.

Matsuo N, et al. Photoreceptor outer segments in the aqueous humor in rhegmatogenous retinal detachment. *Am J Ophthalmol* 1986; 101:673-679.

Matsuo T. Photoreceptor outer segments in aqueous humor: key to understanding a new syndrome. *Surv Ophthalmol* 1994; 39:169-210.

Netland PA, et al. Elevated intraocular pressure secondary to rhegmatogenous retinal detachment. *Surv Ophthalmol* 1994; 39:234-240.

Schwartz A. Chronic open-angle glaucoma secondary to rhegmatogenous retinal detachment. *Am J Ophthalmol* 1973; 75:205.

Sclerocornea 1137

General: Autosomal dominant; feature of cornea plana.

Ocular: Malformation of cornea; indistinct limits of cornea and sclera.

Clinical: Found in a patient with monosomy 21; may be found in association with hypertelorism, syndactyly, ambiguous genitalia, and epidermolysis bullosa dystrophica.

Bloch N. Les Differents Types de Sclerocornea, Lewis Modes d'Heredite et les Malformations Congenitales Concornitantes. *J Genet Hum* 1965; 14:133-172.

Doane JF, et al. Bilateral penetrating keratoplasty for sclerocornea in an infant with monosomy 21. Case report and review of the literature. *Cornea* 1994; 13:454-458.

Martinez-Frias ML, et al. Sclerocornea, hypertelorism, syndactyly, and ambiguous genitalia. *Am J Med Genet 1994;* 49:195-197.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Sharkey JA, et al. Cornea plana and sclerocornea in association with recessive epidermolysis bullosa dystrophica. Case report. *Cornea* 1992; 11:83-85.

Sebaceous Gland Carcinoma 1138

General: Ocular adnexa contains various sebaceous glands from which carcinomas may arise; predilection for the upper lids but may involve both lids; usually in older age groups; slight female preponderance.

Ocular: Blepharitis; madarosis; meibomianitis; sebaceous carcinoma of lids or orbit; orbital edema; proptosis; conjunctivitis; superficial keratitis; lacrimal gland tumor.

Clinical: Metastasis to preauricular or cervical lymph nodes, or submandibular area.

Boniuk M, Zimmerman LE. Sebaceous carcinoma of the eye lid, eyebrow, caruncle, and orbit. *Int Ophthalmol Clin* 1972; 12:225-257.

Harvey PA, et al. Primary sebaceous carcinoma of the lacrimal gland: a previously unreported primary neoplasm. *Eye* 1994; 8[Part 5]:592-595.

Rao NA, et al. Sebaceous carcinomas of the ocular adnexa: a clinicopathologic study of 104 cases, with five-year followup data. *Hum Pathol* 1982; 13:113-122.

Seborrheic Dermatitis 1139

General: Genetically determined structural and functional abnormality of the skin that produces an inflammatory reaction and dryness.

Ocular: Scaling and pruritus of lids and lateral angles of eyes; chronic blepharitis; conjunctivitis; keratitis; iritis; keratomalacia in severe forms; scaling of the eyebrows and glabella; honey-colored crusting along rim of the eyelids; keratinaceous debris around eyelashes.

Clinical: Eruption consists of an erythematous scaling dermatitis with a distribution predisposed to involve the forehead at the hairline.

Harley RD, ed. *Pediatric Ophthalmology*. 4th ed. Philadelphia: WB Saunders, 1998.

McCulley JP, Dougherty JM. Blepharitis associated with acne rosacea and seborrheic dermatitis. *Int Ophthalmol Clin* 1985; 25:159.

Messaritakis J, et al. Generalized seborrhoeic dermatitis: *Clinical* and therapeutic data of 25 patients. *Arch Dis Child* 1975; 50:871-874.

Seckel Syndrome (Bird-Headed Dwarf Syndrome; Virchow-Seckel Dwarfism; Ateliosis; Intrauterine Growth Retardation; Low-Birth-Weight Dwarfism; Nanocephalic Dwarfism; Primordial Dwarfism) 1140

General: Autosomal recessive; syndrome shows variations in phenotypic appearance; both sexes affected; present at birth.

Ocular: Widely spaced eyes; incomplete eyebrow; strabismus; horizontal nystagmus; bilateral macular coloboma with pigmentation and umbilicated appearance; disk hypoplasia.

Clinical: Dwarfism; cranial deformity (bird head); developmental anomalies with short arms and clawlike hands; skeletal anomalies with narrow chest and beading of ribs; facial deformities with hypoplasia of maxilla and mandible; beaklike protrusion of central face; sparse hair; absent thumb; malformation of genitourinary tract and rectum; birdlike malformation of the face and further abnormalities; cardiac anomalies; hypophyseal hypoplasia; association with Legg-Calve Perthes disease.

Di Blasi S, et al. Seckel's syndrome: a case report. *J Med* 1993; 24:75-96.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976, p.389.

Rappen U, von Brenndorff AL. Kardiale Symptomatik kei 2 Patienten mit Seckel-Syndrom. *Monatschr Kinderheilkd* 1993; 141:584-586.

Seckel HPC. *Bird-headed dwarfs.* Springfield, IL: Charles C. Thomas, 1960.

Second Eye Syndrome 1141

General: Associatedwith the second cataract surgery within one month of the first.

Ocular: Increased pain during the second surgery

Matthew MRK, Webb LA, Hill R: Surgeon experience and patient comfort during clear cornea phacoemulsificiation under topical local anesthesia. *J Cataract Refract Surg* 2002; 28: 1977-1981.

O'Brian PD, Fulcher T, Wallace D, et al. Patient pain during different stages of phacoemulsification using topical anesthesia. *J Cataract Refract Surg* 2001; 27: 880-883.

Senior Syndrome (Senior-Loken Syndrome; Tubulointerstitial Nephropathy Syndrome; Loken-Senior Syndrome; Renal-Retinal Syndrome; Renal Dysplasia and Retinal Aplasia) 1142

General: Autosomal recessive trait; pleiotropic gene with variable expression; death before adulthood by renal failure with uremia; both sexes affected; onset in early childhood.

Ocular: Visual loss progressing to complete blindness; progressive tapetoretinal degeneration that becomes manifest in early infancy; retinitis pigmentosa; rubeosis iridis; bilateral or unilateral cataracts; spherophakia; anterior lenticonus; hydrophthalmos; corneal opacities; pallor of optic disk; narrowed arterioles; bone spicule formation.

Clinical: Nephropathy (tubulointerstitial type); growth and mental retardation; nocturia; thirst; polyuria; short stature; hearing loss; brain maldevelopment; convulsions; osteomalacia; deafness; nephronophthisis; osteomalacia; arterial hypertension; aortic insufficiency.

Fillastre JP, et al. Familial chronic interstitial nephropathy and retinal dystrophy (Senior-Loken syndrome). *Nouv Presse Med* 1974; 3:309.

Olmer M, et al. Letterer-Senior-Loken's syndromes. *Nouv Presse Med* 1974; 3:1032.

Rosenthal G, et al. Senior's syndrome (hereditary renal retinal dysplasia) associated with cataracts. *Ann Ophthalmol* 1992; 24:104-105.

Schuman JS, et al. Senior-Loken syndrome and Coats' disease. *Am J Ophthalmol* 1985; 100:822-827.

Senior B, et al. Juvenile familial nephropathy with tapetoretinal degeneration: a new oculo-renal dystrophy. *Am J Ophthalmol* 1961; 52:625.

Senter Syndrome (Keratitis-Ichthyosis-Deafness Syndrome; KID Syndrome; Ichthyosiform Erythroderma, Corneal Involvement, and Deafness 1143

General: Autosomal recessive.

Ocular: Corneal involvement.

Clinical: Ichthyosiform erythroderma; deafness; hepatomegaly; hepatic cirrhosis; glycogen storage; short stature; mental retardation; hepatitis.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Senter TP, et al. Atypical ichthyosiform erythroderma and congenital neurosensory deafness-a distinct syndrome. *J Pediatr* 1978; 92:68-72.

Wilson GN, et al. Keratitis, hepatitis, ichthyosis, and deafness: report and review of KID syndrome. *Am J Med Genet* 1991; 40:255-259.

Shaken Baby Syndrome (Battered-Baby Syndrome; Battered-Child Syndrome; Child Abuse Syndrome; Silverman Syndrome) 1144

General: Associated with parental abuse or accidents.

Ocular: Exophthalmos with orbital hemorrhages; lid hematoma; lid edema; secondary glaucoma; hyphema; vitreous hemorrhages; retinal exudates and hemorrhages (Berlin edema); choroidal atrophy; retinal detachment; papilledema; optic nerve sheath hemorrhage; preretinal, intraretinal, and subretinal hemorrhages; optic disk edema; choroidal hemorrhage.

Clinical: Soft tissue bruises; multiple fractures of long bones, ribs, and skull; pharyngeal bruising, subdural hematoma; seizures; failure to thrive; vomiting associated with lethargy or drowsiness; respiratory irregularities; coma or death; intracranial hemorrhage.

Budenz DL. Ocular and optic nerve hemorrhages in abused infants with intracranial injuries. *Ophthalmology* 1994; 101:559-565.

Coody D, et al. Shaken baby syndrome: identification and prevention for nurse practitioners. *J Pediatr Health Care* 1994; 8:50-56.

Lambert SR, et al. Optic nerve sheath and retinal hemorrhages associated with the shaken baby syndrome. *Arch Ophthalmol* 1986; 104:1509-1512.

Munger CE, et al. Ocular and associated neuropathologic observations in suspected whiplash shaken infant syndrome. A retrospective study of 12 cases. *Am J Forensic Med Pathol* 1993; 14:193-200.

Shamberg Disease 1145

General: Common benign skin disorder; self-limiting cutaneous vasculitis.

Ocular: Discrete foci of retinal periphlebitis associated with localized intraretinal hemorrhages and exudates; retinal and cutaneous vasculitides wax and wane concurrently; retinal vasculitis is self-limiting cause of visual disturbance and requires no therapy.

Clinical: Purpuric, erythematous patches appear on trunk and extremities; mononuclear perivascular infiltrate of involved skin.

Bedrick JJ, et al. Retinal vasculitis in Shamberg's disease. *Ophthalmology* 1982; 89:188.

Shigellosis (Bacillary Dysentery) 1146

General: Caused by *Shigella;* frequently passed through food and via food handlers; more commonly seen in countries with poor sanitation; evidence suggests that the ability of shigellae to invade and multiply within the corneal epithelium is similar to the invasion in the intestinal epithelium.

Ocular: Scleroconjunctivitis; severe uveitis; conjunctival xerosis.

Clinical: Fever; abdominal pain; diarrhea; intestinal perforation; toxic megacolon; dehydration; there has been one case reported of an association with the Klüver-Bucy syndrome.

Bennish ML. Potentially lethal complications of shigellosis. *Rev Infect Dis* 1991; 13[Suppl 4]:S319-S324.

Kaslow RA, et al. Search for Reiter's Syndrome after an outbreak of Shigella sonnei dysentery. *J Rheumatol* 1979; 6:562-566.

Overton ME, et al. Conjunctivitis and ethmoiditis due to Shigella flexneri in an infant. *Clin Pediatr* 1981; 20:231.

Ronen S, et al. Orbital inflammation: an unusual extraintestinal complication of shigellosis. *Ophthalmologica* 1980; 180:46-50.

Yoshikawa M, Sasakawa C. Molecular pathogenesis of shigellosis: a review. *Microbiol Immunol* 1991; 35:809-824.

Short Syndrome 1147

General: Autosomal recessive; short stature; hyperextensibility; hernia; ocular depression; Rieger anomaly; teething delay.

Ocular: Sunken eyes; ocular depression; Rieger syndrome; glaucoma.

Clinical: Short stature; hyperextensibility of joints; hernia; low birth weight; teething delay; delayed speech development; deafness; diabetes mellitus.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Schwingshandl J, et al. SHORT syndrome and insulin resistance. *Am J Med Genet* 1993; 47:907-909.

Sensenbrenner JA, et al. A low birthweight syndrome? Rieger syndrome. *Birth Defects* 1975; 11:423-426.

Stratton RF, et al. Sibs with growth deficiency delayed bone age, congenital hip dislocation and iridocorneal abnormalities with glaucoma. *Am J Med Genet* 1989; 32:330-332.

Shy-Drager Syndrome (Orthostatic Hypotension Syndrome; Shy-MeGee-Drager Syndrome) 1148

General: Etiology unknown; gradual onset; adults; progressive degeneration of the nervous system.

Ocular: External ophthalmoplegia; iris atrophy; ocular sympathetic and parasympathetic insufficiency (alternating Homer syndrome, cholinergic supersensitivity, decreased lacrimation, and corneal hypesthesia).

Clinical: Orthostatic hypotension; rigidity; tremor; adiadochokinesia; wasting of muscles; mental retardation; impotence; dysphagia; bilateral vocal cord paralysis; neurogenic bladder; anhydrosis; extremity weakness and paresthesias; dizziness; abnormal postural balance.

Bawa R, et al. Bilateral vocal cord paralysis with Shy-Drager syndrome. *Otolaryngol Head Neck Surg* 1993; 109: 911-914.

Ohashi N, et al. Otoneurological manifestations of the Shy-Drager syndrome. *Eur Arch Otorhinolaryngol* 1991; 248: 150-152.

Rosen J, Brown SI. New ocular signs in Shy-Drager syndrome. *Am J Ophthalmol* 1974; 78: 1032.

Shy GM, Drager GA. A neurological syndrome associated with orthostatic hypotension. *Arch Neurol* 1960; 2:511.

Verinder S, et al. Ocular manifestations of Shy-Drager syndrome. *Ann Ophthalmol* 1982; 14:635-638.

Shy-Gonatas Syndrome 1149

General: Unknown etiology; similar to Hunter and Refsum syndromes; accumulation of lipids in muscles simulates gargoylism; present from birth.

Ocular: Mild proptosis; hypertelorism; ptosis; external ophthalmoplegia (progressive); concentric visual field constriction; keratopathy with possible corneal ulcer; lattice-like white opacities in the area of Bowman membrane; retinal pigmentary degeneration (atypical retinitis pigmentosa) with difficulties with night vision.

Clinical: Weakness of extremities (proximal); myopathy and neuropathy; cerebellar ataxia.

Geeraets WJ *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gonatas NK. A generalized disorder of nervous system, skeletal muscle and heart resembling Refsum's disease and Hurler's syndrome. II. Ultrastructure. *Am J Med* 1967; 42: 169.

Shy GM, et al. A generalized disorder of nervous system, skeletal muscle and heart resembling Refsum's disease and Hurler's syndrome. I. Clinical, pathologic and biochemical characteristics. *Am J Med* 1967; 42:163.

Siegrist Syndrome (Pigmented Choroidal Vessels) 1150

General: Rare; more common in females (2:1); malignant hypertension; onset in advanced age.

Ocular: Exophthalmos; granular pigmented spots in the choroid, fairly uniform, following the course of larger choroidal vessels with extension radially toward the periphery; changes related to arteriosclerotic choroidal changes and seen following chorioretinitis of pregnancy and albuminuric choroiditis; Elschnig spots.

Clinical: Hypertension; albuminuria.

Archer D, et al. Fluorescein studies of choroidal sclerosis. *Am J Ophthalmol Suppl* 1971; 71:266-285.

Deutman AF, et al. Acute posterior multi focal placoid pigment epitheliopathy. Pigment epitheliopathy of choriocapillaris. *Br J Ophthalmol* 1972; 56:863.

Duke-Elder S, ed. *Textbook of Ophthalmology, vol. III.* St. Louis: CV Mosby, 1941.

Schmidt D, Loffer KU. Elschnig's spots as a sign of severe hypertension. *Ophthalmologica* 1993; 206:24-28.

Siegrist A. *Report at the 9th International Congress, Utrecht*. 1899:36.

Silent Sinus Syndrome 1151

General: Spontaneous enophthalmos and hypoglobus associated with a small, ipsilateral maxillary sinus.

Ocular: Enophthalmos; hypoglobus.

Clinical: Patients usually undergo painless progressive sinking of the eye.

Davidson JK, et al. Negative sinus pressure and normal predisease imaging in silent sinus syndrome. *Arch Ophthalmol* 1999; 117:1653-1654.

Eto RT, House JM. Enophthalmos: a sequela of maxillary sinusitis. *AJNR Am J Neuroradiol* 1995; 16:939-941.

Kass ES, Salman S, Montgomery WW. Manometric study of complete ostial occlusion in chronic maxillary atelectasis. *Laryngoscope* 1996; 106: 1255-1258.

Rose, GE, Sandy C, Hallberg L, et al. Clinical and radiologic characteristics of the imploding antrum or silent sinus syndrome. *Ophthalmology* 2003; 110: 811-818.

Scharf KE, et al. Pressure measurements in the normal and occluded rabbit maxillary sinus. *Laryngoscope* 1995; 105: 570-574.

Soparker CNS, et al. The silent sinus syndrome. *Ophthalmology* 1994; 101:772-778.

Silver Syndrome (Congenital Hemihypertrophy) 1152

General: Muscular hypertrophy of one side of face; etiology unknown; reported association of this condition with a small deletion in chromosome 13.

Ocular: Café-au-lait spots of the lid.

Clinical: Broad forehead; small triangular face; inverted V-shaped mouth; genitourinary abnormalities; precocious puberty; medullary sponge kidney; Wilms tumor.

Aita JA. *Congenital facial anomalies with neurologic defects*. Springfield, IL: Charles C. Thomas, 1969.

Beetz R, et al. Medullary sponge kidneys and unilateral Wilms tumor in a child with Beckwith-Wiedemann syndrome. *Eur J Pediatr* 1991; 150:489-492.

Wahlstrom J, et al. Silver-like syndrome and a small deletion on chromosome 13. *Acta Paediatr* 1993; 82:993-996.

Simmonds Syndrome (Hypopituitarism Syndrome) 1153

General: Anterior pituitary gland destroyed by various causes, such as hemorrhage, infarction, injuries, or postparturition infections; females; late form of Simmonds syndrome is Snapper-Witts with achlorhydria and subacute combined degeneration, and hypochromic or hyperchromic anemia; onset during postpubertal period.

Ocular: Loss of eyebrow; loss of eyelashes; central scotomata; diabetic retinopathy tends to improve after development of this syndrome; optic nerve atrophy (descending type).

Clinical: Weight loss and generalized weakness (progressive); anorexia; amenorrhea; dry skin and brittle nails; hypotension with bradycardia; anemia; psychosis; loss of libido.

Blondell RD. Hypopituitarism. *Am Fam Physician* 1991; 43:2029-2936.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Simmonds M. Uber Hypophysisschwund mit Todlichem Ausgang. *Dtsch Med Wochenschr* 1914; 40:322.

Sipple Syndrome (Multiple Endocrine Neoplasia 2 or 2A; Multiple Endocrine Adenomatosis 2 or 2A; Familial Chromaffinomatosis; Multiple Neuroma; Pheochromocytoma-Thyroid Medullary Carcinoma; PCT; MEN2 or MEN2A; MEA2 or MEA2A) 1154

General: Autosomal dominant; sporadic types have been described; both sexes affected; genetic mapping has assigned the genes responsible for these tumors to the pericentromeric region of chromosome 10.

Ocular: Prominent corneal nerves (rare).

Clinical: Association of medullary thyroid carcinoma and pheochromocytoma; parathyroid tumors; neurofibromas; diabetes mellitus; diarrhea.

Ledger GA, et al. Genetic testing in the diagnosis and management of multiple endocrine neoplasia type II. *Ann Intern Med* 1995; 122:118-124.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Sipple JH. The association of pheochromocytoma with carcinoma of the thyroid gland. *Am J Med* 1961; 31: 163-166.

Sjögren Syndrome (Gougerot-Sjögren Syndrome; Secretoinhibitor Syndrome; Sicca Syndrome) 1155

General: Etiology unknown; autosomal recessive; occurs in women over age 40 years; failure of the lacrimal and conjunctival glands to maintain adequate secretion; similarities exist with Mikulicz syndrome (see Mikulicz-Radecki Syndrome); insidious onset; associated with collagen disorders; Epstein-Barr virus infection.

Ocular: Blepharoconjunctivitis; tears show no lysozyme; keratoconjunctivitis sicca; superficial corneal ulcers; thready, tenacious, yellow-white discharge of the conjunctiva; hypertrophy of lacrimal gland; decreased tear secretion with cellular and mucous debris in tear film; cicatrization of cornea and conjunctiva.

Clinical: Dryness of mouth and other mucous membranes; enlarged salivary glands; dysphagia; painless swelling of joints; polyarthritis; dental cavities; vaginitis; laryngitis; rhinitis sicca; hepatomegaly; focal myositis; alopecia; splenomegaly.

Fischbach M, et al. Immune complexes in Sjogren's syndrome. *Arthritis Rheum* 1980; 23:791-795.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Friedalaender MH. Ocular manifestations of Sjogren's syndrome: keratoconjunctivitis sicca. *Rheum Dis Clin North Am* 1992; 18:591-608.

Gudas PP, et al. Corneal perforations in Sjogren's syndrome. *Arch Ophthalmol* 1973; 90:470.

Miyasaka N, et al. Possible involvement of Epstein-Barr virus in the pathogenesis of Sjogren's syndrome. *Clin Immunol Immunopathol* 1994; 72:166-170.

Sjogren H. Zur Kenntnis der Keratoconjunctivitis Sicca. *Acta Ophthalmol* 1933[Suppl 2].

Williamson J, et al. Management of the dry eye in Sjogren's syndrome. *Br J Ophthalmol* 1974; 58:798-805.

Wu AJ, Fox PC. Sjogren's syndrome. *Semin Dermatol* 1994; 13:138-143.

Sjögren-Larsson Syndrome (Oligophrenia Ichthyosis Spastic Diplegia Syndrome) 1156

General: Rare; autosomal recessive; consanguinity; loss of neurons and gliosis throughout gray matter; autosomal recessively inherited disorder characterized by the triad of congenital ichthyosis, spastic diplegia or tetraplegia, and mental retardation.

Ocular: Hypertelorism; ichthyosis of lid; chorioretinitis with macular and perimacular pigment degeneration or bright, glistening intraretinal dots; atypical retinitis pigmentosa; blepharitis; conjunctivitis; keratitis; tan/white areas of retinal pigment epithelium loss; maculopathy.

Clinical: Oligophrenia idiocy; ichthyosis (congenital); spastic disorders; epilepsy; speech defect.

Barnard NA, et al. Sjogren-Larsson syndrome: case reports of two brothers. *Ophthalmic Physiol Optics* 1991; 11: 180-183.

Ghadially R, Chong LP. Ichthyoses and hyperkeratotic disorders. *Dermatol Clin* 1992; 10:597-607.

Ionasesco V, et al. Amino acid abnormality in Sjogren-Larsson syndrome. *Arch Neurol* 1973; 28:197.

Sjogren T, Larsson T. Oligophrenia in combination with congenital ichthyosis and spastic disorders: a *Clinical* and genetic study. *Acta Psychiatr Neurol Scand* 1957[Suppl 113].

Skew Deviation Syndrome (Hertwig-Magendie Syndrome) 1157

General: Vascular deficiency involving the brainstem and middle cerebral peduncle; observed after encephalitis, tumors of the cerebellum, and lesions of the labyrinth; this is a vertical strabismus due to a supranuclear lesion; bilateral and alternating skew deviation suggests midbrain or caudal medullar lesion.

Ocular: Alternating paresis of elevators of one eye combined with depressor paresis of the other eye or alternating hyperphorias; in skew deviation the eyes point in diagonally opposite directions; this effect is more pronounced in the eye contralateral to the lesion.

Clinical: Cerebellar ataxia.

Halmagyi GM. Central eye movement disorders. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV*. Philadelphia: WB Saunders, 1994:2411.

Keane JR. Alternating skew deviation: 47 patients. *Neurology* 1985; 35:725.

Miller NR. *Walsh and Hoyt's Clinical Neuro-Ophthalmology*, 4th ed. Baltimore: Williams & Wilkins, 1987.

Moster ML, et al. Alternating skew deviation on lateral gaze (bilateral abducting hypertropic). *Ann Neurol* 1988; 23:190.

Silfverskiold BP. Skew deviation in Wallersberg's syndrome. *Acta Neurol Scand* 1966; 41:381.

Sleep Apnea (Obstructive Sleep Apnea) 1158

General: Interruption of normal breathing during sleep secondary to airway obstruction; life threatening.

Ocular: Papilledema; optic disk edema; floppy eyelid syndrome.

Clinical: Obesity; hypertrophic tonsils and adenoids; excessive daytime sleepiness; snoring with periods of silence; memory loss; headache; increased intracranial pressure; personality changes; oscillopsia; pickwickian syndrome; chronic disequilibrium; increased intracranial pressure.

De Voll JR, Sacks EJ. Evaluation of an aviator with chronic disequilibrium, oscillopsia, and central sleep apnea. *Aviat Space Environ Med* 1994; 65:263-266.

Doyle KJ, Tami TA. Increased intracranial pressure and blindness associated with obstructive sleep apnea. *Otolaryngal Head Neck Surg* 1991; 105:613-616.

Netland PA, et al. Histopathologic features of the floppy eyelid syndrome. Involvement of tarsal elastin. *Ophthalmology* 1994; 101:174-181.

Sugita Y, et al. Marked episodic elevation of cerebrospinal fluid pressure during nocturnal sleep in patients with sleep apnea hypersomnia syndrome. *Electroencephalogr Clin Neurophysiol* 1985; 60:214.

Sullivan CE, et al. Reversal of obstructive sleep apnea by continuous positive airway pressure applied through nose. *Lancet* 1981; 1:862.

Wolin MJ, Brannon WL. Disk edema in an overweight woman. *Surv Ophthalmol* 1995; 39:307-314.

Slit Ventricle Syndrome 1159

General: Self limited episodes of shunt malfunction associated with a small or unchanged ventricular system; rare.

Ocular: Esotropia; nystagmus; optic atrophy

Clinical: Hydroencephalus; headache; nausea and vomiting; altered levels of consciousness

Nguyen TN, Polomeno RC, Farmer JP, et al. Ophthalmic complications of slit-bentricle syndrome in children. *Ophthalmology* 2002; 109: 520-525

Sluder Syndrome (Sphenopalatine Ganglion Neuralgia Syndrome; Lower Facial Neuralgia Syndrome) 1160

General: Irritation of the sphenopalatine ganglion; attacks of pain last from minutes to days (see Charlin Syndrome).

Ocular: Severe orbital pain; increased lacrimation during episodes of pain.

Clinical: Unilateral facial pain, mainly root of nose, orbit, and mastoid area; episodes of headaches; nasal congestion.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol. II*, 4th ed. Baltimore: Williams & Wilkins, 1982.

Seltzer AP. Facial pain. *J Natl Med Assoc* 1971; 63:354.

Sluder G. The role of sphenopalatine (Meckel's) ganglion in nasal headaches. *NY Med J* 1908; 87:989.

Smallpox (Variola) 1161

General: Highly contagious cutaneous disease caused by viral infection.

Ocular: Conjunctivitis; keratitis; corneal ulcer; hypopyon; endophthalmitis; congenital corneal clouding; albinotic spots on iris; choroiditis; vitreous opacities; papillitis; extraocular muscle palsies; entropion; dacryocystitis; chorioretinitis; optic neuritis; and vesicles of the eyelid; preauricular adenopathy; eyelid ulcerating pustules; several conditions predispose to the spread of vaccinia, including eczema, hypogammaglobulinemia, steroid therapy, and AIDS.

Clinical: Fever, headache, and vomiting prior to appearance of the rash on the face, upper trunk, and down to the extremities.

Fillmore GL, Ward TP, Bower KS, et.al: Ocular complications in the department of defense smallpox vaccination program. *Ophthalmology* 2004; 111: 2086-2093.

Harley RD, ed. *Pediatric Ophthalmology*. 4th ed. Philadelphia: WB Saunders, 1998.

Ruben FL, Lane JM. Ocular vaccinia: an epidemiologic analysis of 348 cases. *Arch Ophthalmol* 1970; 84:45-48.

Wright DC, et al. Disseminated vaccinia in a military recruit with human immunodeficiency virus (HIV) disease. *N Engl J Med* 1987; 316:673-676.

Smith Syndrome (Facio-Skeleto-Genital Dysplasia) 1162

General: Autosomal recessive; more common in males.

Ocular: Ptosis; antimongoloid slant; epicanthus.

Clinical: Microcephaly; high-arched palate; large, low-set ears; mental retardation; broad nose; hypoplastic mandible; pedal syndactyly.

Aita JA. *Congenital facial anomalies with neurologic defects.* Springfield, IL: Charles C. Thomas, 1969:246.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Smith-Lemli-Opitz Syndrome (Cerebrohepatorenal Syndrome) 1163

General: Autosomal recessive; similarities with trisomy 18 syndrome; prognosis poor, with death in early infancy (see Zellweger Syndrome); onset in fetal life; prevalent in males; reduced myelination in the cerebral hemispheres, cranial nerves, and peripheral nerves secondary to a defective cholesterol biosynthesis.

Ocular: Joining of the eyebrows (synophrys); ptosis (bilateral); pronounced epicanthal folds; strabismus; nystagmus; cataract; optic nerve demyelinization.

Clinical: Mental retardation; microcephaly; hypertonia; low-set ears; high-arched palate; failure to thrive; vomiting; hypospadias; cryptorchidism; metatarsus adductus.

Harbin RL, et al. Sclerocornea associated with the Smith-Lemli Opitz syndrome. *Am J Ophthalmol* 1977; 84: 72-73.

Kretzer FL, et al. Ocular manifestation of the Smith-Lemli-Opitz syndrome. *Arch Ophthalmol* 1981; 99:2000-2006.

Nevo S. "Smith-Lemli-Opitz syndrome" in an inbred family. *Am J Dis Child* 1972; 124:431.

Setchell KD, et al. Oral bile acid treatment and the patient with Zellweger syndrome. *Hepatology* 1992; 15:198-207.

Smith DW, et al. Newly recognized syndrome of multiple anomalies. *J Pediatr* 1964; 64:210.

Tint GS, et al. Defective cholesterol biosynthesis associated with the Smith-Lemli-Opitz syndrome. *N Engl J Med* 1994; 330:107-113.

Smith-Magenis Syndrome (SMS) 1164

General: Mental retardation, physical dysmorphia, and behavior abnormalities due to a deletion at chromosome l7pl1.2.

Ocular: High myopia; retinal detachment; iris anomalies (absent collarette, nasal corectopia, stromal dysplasia); microcornea; strabismus; iris nodules called Wolfflin-Kruckmann spots.

Clinical: Wolfflin-Kruckmann spots may be confused with Brushfield spots, which are seen only in Down syndrome patients.

Barnicoat AJ, et al. An unusual presentation of Smith-Magenis syndrome with iris dysgenesis. *Clin Dysmorphol* 1996; 5:153-158.

DeRijk-van Andel JR, et al. Clinical and chromosome studies of three patients with Smith-Magenis syndrome. *Dev Med Child Neurol* 1991; 33:343-347.

Finecane BM, et al. Eye abnormalities in the Smith-Magenis contiguous gene deletion syndrome. *Am J Med Genet* 1993; 45:443-446.

Sneddon Disease 1165

General: Livedo reticularis; neurologic abnormalities; labile hypertension; apparently autosomal dominant inheritance characterized by a rare potentially severe, arterioocclusive disorder; probably an immunologically mediated disorder leading to the migration and proliferation of smooth cells of small arteries, resulting in partial or complete narrowing of the vessel lumen.

Ocular: Central retinal artery occlusion; visual loss; optic atrophy; visual field defect; cherry-red macula.

Clinical: Diffuse headaches; hemihypesthesia; transient aphasic attack; hemianopsia; reticular blue discoloration of skin; hypertension; transient global amnesia; livedo reticularis; progressive neurologic deterioration; multiple ischemic cerebrovascular episodes; renal cell carcinoma.

Carella F, et al. Sneddon's syndrome and renal carcinoma. Case report. *Funct Neurol* 1992; 7:395-400.

Green KM, et al. Livedo reticularis with ulcers and circulating immune complexes. *Cutis* 1983; 31:312.

Jones J, et al. Central retinal artery occlusion in Sneddon's disease associated with antiphospholipid antibodies. *Am J Ophthalmol* 1986; 102:37-40.

Lossos A, et al. Familial Sneddon's syndrome. *J Neurol* 1995; 242:164-168.

Sepp N. et al. Sneddon's syndrome-an inflammatory disorder of small arteries followed by smooth muscle proliferation. Immunohistochemical and ultrastructural evidence. *Am J Pathol* 1995; 19:448-453.

Sneddon IB. Cerebrovascular lesions and livedo reticularis. *Br J Dermatol* 1965; 77: 180.

Snowflake Vitreoretinal Degeneration 1 1166

General: Autosomal dominant; very small yellow-white dots on the retina.

Ocular: Fibrillar vitreous degeneration; thickening of cortical vitreous; optically empty vitreous cavity; vitreous hemorrhage; posterior vitreous detachment with collapse; retinal detachment; retinal hemorrhage; retinal holes; marked retinal pigmentation; obliterated retinal vessels; sheathed retinal vessels; preretinal retraction; chorioretinal atrophy; corneal opacities; myopia; hyperopia; astigmatism; amblyopia; cataract; glaucoma.

Clinical: None.

Gheiler M, et al. Hereditary snowflake vitreoretinal degeneration. *Birth Defects* 1982; 18:577-580.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Snuff-Out Syndrome (Snuff Syndrome) 1167

General: Rare; sudden vision loss following ocular or laser surgery in patients with advanced glaucoma; etiology unknown but probably involves several factors, including unrecognized increased intraocular pressure, sudden hypotony, nerve injury, or retrobulbar anesthesia.

Ocular: Loss of central fixation; reduction in visual acuity; reduction in visual field; cataract; glaucoma.

Clinical: None.

Harrington DO, ed. *The visual fields: a textbook and atlas of Clinical perimetry*. 5th ed. St. Louis: CV Mosby, 1981.

Kolker AE. Visual prognosis in advanced glaucoma: a comparison of medical surgical therapy for retention of vision in 101 eyes with advanced glaucoma. *Trans Am Ophthalmol Soc* 1977; 75:539-555.

Lichter PR, Ravin JG. Risks of sudden visual loss of glaucoma surgery. *Am J Ophthalmol* 1974; 78:1009.

Sharma N, Ooi JL, Francis IC, et al. Sudden visual loss after uneventful cataract surgery: Snuff syndrome. *J Cataract Refract Surg* 2004; 30: 2435-2437.

Sorsby I Syndrome (Hereditary Macular Coloboma Syndrome) 1168

General: Autosomal dominant; related to Laurence-Moon-Bardet-Biedl and Biemond syndromes; apical dystrophy of the extremities and bilateral macular colobomata; both sexes affected; onset from birth (see Laurence-Moon-Bardet-Biedl Syndrome; Biemond Syndrome).

Ocular: Hypermetropia; nystagmus; bilateral macular colobomata with various degrees of pigmentation but sharply lined borders.

Clinical: Distal dystrophy of the hands and feet; rudimentary or absent index fingernails; absence of big toe; cleft palate.

Francois J. *Heredity in Ophthalmology.* St. Louis: CV Mosby, 1961:694.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Sorsby A. Congenital coloboma of the macula: together with an account of the familial occurrence of bilateral macular coloboma in association with apical dystrophy of hands and feet. *Br J Ophthalmol* 1935; 19:65.

Sorsby II Syndrome (Sorsby Macular Dystrophy) 1169

General: Both sexes affected; onset in third and fourth decades of life.

Ocular: Retinal hemorrhages; retinal exudates; chorioretinitis; macular dystrophy.

Clinical: None.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Sorsby A. The dystrophies of the macula. *Br J Ophthalmol* 1940; 24:469-529.

Sorsby III Syndrome (Sorsby Fundus Dystrophy) 1170

General: Both sexes affected; onset in fifth decade of life; autosomal dominant; mutations in the tissue inhibitor of metalloproteinases-3 have been associated with this condition; condition has been genetically linked to chromosome 22q 13-qter.

Ocular: Retinal hemorrhages; retinal exudates; retinal pigmentary deposits; choroid atrophy; choroidal neovascularization; abnormal color vision; generalized fine granularity of the retinal pigment epithelium and peripheral iris transillumination.

Clinical: None.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Sorsby A, et al. A fundus dystrophy with unusual features (late onset and dominant inheritance of central retinal lesion showing edema, hemorrhage and exudates developing into generalized choroidal atrophy with massive pigment proliferation). *Br J Ophthalmol* 1949; 33:67-100.

Weber BH, et al. Mutations in the tissue inhibitor of metalloproteinases-3 (TIMP3) in patients with Sorsby's fundus dystrophy. *Nat Genet* 1994; 8:352-356.

Wu G, et al. Hereditary hemorrhagic macular dystrophy. *Am J Ophthalmol* 1991; 111:294-301.

Sotos Syndrome (Cerebral Gigantism) 1171

General: Idiopathic disturbance of the diencephalon; etiology unknown; cerebral gigantism in childhood, Russell syndrome, and total lipodystrophy are related forms of the same entity (see Russell Syndrome).

Ocular: Hypertelorism; antimongoloid lid aperture; high refractive error (hyperopia); nystagmus; strabismus.

Clinical: Acromegaly; large skull with frontal bossing; mental retardation; incoordination; abnormal excessive growth, mainly during first 2 years of life.

Maino DM, et al. Ocular manifestations of Sotos' syndrome. *J Am Optom Assoc* 1994; 65:339-346.

Milunski A, et al. Cerebral gigantism in childhood: a report of two cases and review of the literature. *Pediatrics* 1967; 40:395.

Sotos JF, et al. Cerebral gigantism in childhood: a syndrome of excessively rapid growth with acromegalic features and a nonprogressive neurologic disorder. *N Engl J Med* 1964; 271:109.

Yeh H, et al. Cerebral gigantism (Sotos' syndrome) and cataracts. *J Pediatr Ophthalmol Strabismus* 1978; 15: 231-232.

Spanlang-Tappeiner Syndrome (Keratosis Palmoplantaris and Corneal Dystrophy Syndrome) 1172

General: Autosomal dominant; etiology unknown; occurs in both sexes; onset age 5 to 20 years; linear palmar and diffuse plantar keratosis with dystrophy of the cornea.

Ocular: Corneal opacities (yellowish, tongue-shaped, and not always involving the center of the cornea).

Clinical: Hyperkeratosis of palms and soles; nail dystrophy; hyperhidrosis.

Spanlang H. Beitrage zur Klinik und Pathologie Seltener Hornhauterkrankungen (Dystrophia Adiposo Comeae; Dyskeratosis Comaea Congenitalis). *Z Augenheilkd* 1927; 62:21.

Thomas CI. *The Cornea.* Springfield, IL: Charles C. Thomas, 1972.

Sparganosis 1173

General: Infestations in humans by the plerocercoid larvae of the cestodes *Spirometra mansoni* and *Diphyllobothrium;* usually occurs in tropical countries.

Ocular: Conjunctivitis; blepharospasm; orbital cellulitis; ptosis.

Clinical: Marked eosinophilia; intraventricular hemorrhage; hydrocephalus; a case of sparganosis in the spinal canal with partial block has been reported.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*, 11th ed. New York: McGraw-Hill, 1987.

Cho YD, et al. Sparganosis in the spinal canal with partial block: an uncommon infection. *Neuroradiology* 1992; 34:241-244.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Leon LA, et al. A case of ocular sparganosis in Ecuador. *J Parasitol* 1972; 58:184-185.

Wong CW, Ho YS. Intraventricular haemorrhage and hydrocephalus caused by intraventricular parasitic granuloma suggesting cerebral sparganosis. *Acta Neurochir* 1994; 129:205-208.

Spasmus Nutans Syndrome 1174

General: Etiology unknown; both sexes affected; onset between ages 6 and 18 months; disappears during sleep; aggravated by cold weather; spontaneous disappearance by age 3 or 4 years; it is not clear whether head nodding is a compensatory mechanism to control the nystagmus or an involuntary movement of pathologic origin.

Ocular: Bilateral nystagmus; attempt at gaze fixation intensifies manifestations.

Clinical: Rhythmic movements of head in upright position.

Gottlob I, et al. Head nodding is compensatory in spasmus nutans. *Ophthalmology* 1992; 99: 1024-1 031.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Spastic Paraplegia, Optic Atrophy, Dementia 1175

General: Autosomal dominant.

Ocular: Pallor of optic disk; constricted visual fields; optic atrophy; visible deficit in retinal fiber layer; deficit in color vision; slight decrease in visual acuity; pupillary reflex sluggishness to light.

Clinical: Dementia; spastic paraparesis; stiff gait; increased deep tendon reflexes; bilateral extensor plantar responses; euphoria; pseudobulbar speech; incontinence.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Rothner AD, et al. Familial spastic paraparesis, optic atrophy, and dementia: *Clinical* observations of affected kindred. *NY J Med* 1976; 76:756-758.

Spastic Paraplegia, X-Linked (SPPX) 1176

General: X-linked; early onset; slow progression and long survival with eventual involvement of the cerebellum, cerebral cortex, and optic nerves.

Ocular: Nystagmus; optic atrophy; poor vision; cataracts; convergent strabismus; red-green color vision defects.

Clinical: Spastic paraplegia; athetosis; mental retardation; dysarthria, ankle clonus; clubfeet; slow speech; spasticity of the legs; hyperactive reflexes; bilateral Babinski signs; toe walking; bilateral pes cavus; knee clonus; upward plantar reflexes; urinary incontinence; recurrent urinary infection; hematuria; progressive spastic gait disorder; hyperreflexia.

Gutmann DH, et al. Complicated hereditary spastic paraparesis with cerebral white matter lesions. *Am J Med Genet* 1990; 36:251-257.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Raggio JF, et al. X-linked hereditary spastic paraplegia. *J La State Med Soc* 1973; 125: 4-6.

Ulku A, et al. X-linked spastic paraplegia. *Acta Neurol Scand* 1991; 83:403-406.

Spastic Quadriplegia, Retinitis Pigmentosa, Mental Retardation 1177

General: Autosomal recessive; consanguineous parents.

Ocular: Granular pigmented retina; pale optic disk; retinal degeneration; exotropia; miotic pupils; ptosis; nystagmus; small optic disk; retinitis pigmentosa.

Clinical: Expressionless face; drooling; spastic contractures; scissoring; spastic gait; mental retardation; brachydactyly; hypoplasia; tremors; hearing impairment.

Gordon AM, et al. Progressive quadriparesis, mental retardation, retinitis pigmentosa and hearing loss: report of two siblings. *Johns Hopkins Med J* 1976; 138:142-145.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Spatial Visualization Aptitude 1178

General: Sex-linked; more prevalent in males.

Ocular: Aptitude for visualizing space.

Clinical: None.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Stafford RE. Sex differences in spatial visualization as evidence of sex-linked inheritance. *Percept Mot Skills* 1961; 13:428.

Sphenocavernous Syndrome 1179

General: Lesion in the cavernous sinus; similar to the superior orbital fissure syndrome (Rochon-Duvigneaud) and orbital apex syndrome (see Rochon-Duvigneaud Syndrome).

Ocular: Proptosis; edema; paresis of cranial nerves III, IV, and VI (paralysis of the abducens nerve precedes paralysis of the oculomotor nerve, because the abducens is situated between the internal carotid artery and the cavernous sinus wall); conjunctival edema.

Clinical: Paresis of the first (sometimes second and third) division of cranial nerve V; sinusitis.

Geeraets WJ. *Ocular Syndromes.* 3rd ed. Philadelphia: Lea & Febiger, 1976:404.

Jefferson G. Concerning injuries, aneurysms and tumours involving the cavernous sinus. *Trans Ophthalmol Soc UK* 1953; 73:117.

Sekhar LN, et al. Surgical management of lesions within the cavernous sinus. *Clin Neurosurg* 1991; 37:440-489.

Watson NJ, et al. A case of sinusitis presenting with spheno-cavernous syndrome: discussion of the differential diagnosis. *Scott Med J* 1991; 36:179-180.

Sphenomaxillary Fossa Syndrome (Pterygopalatine Fossa Syndrome) 1180

General: Malignant tumor, second division of which involves the sphenopalatine fossa, causing paralysis of cranial nerve V; similar to Trotter syndrome with unilateral deafness, mandibular pain, facial pain, defective mobility of the palate, and trismus.

Ocular: Infraorbital anesthesia; optic nerve atrophy.

Clinical: Maxillary neuralgia with pain in the upper teeth; mandibular pain; displaced jaw toward involved side because of pterygoid muscle paralysis; deafness (middle ear, ipsilateral).

Ford FR, Walsh FB. Raeder's paratrigeminal syndrome. *Bull Johns Hopkins Hasp* 1958; 103:296.

Klossek JM, et al. Endoscopic approach of the pterygopalatine fossa: report of one case. *Rhinology* 1994; 32: 208-210.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Spider Bites 1181

General: Venom of several different spiders can cause systemic poisoning in humans.

Ocular: Conjunctivitis; subconjunctival hemorrhages; conjunctival chemosis; lid edema; lid gangrene; necrosis of lid; ptosis; pupil constriction; retinal cyanosis; visual loss.

Clinical: Localized itching; vesicle and necrosis of tissue; secondary infection; abdominal rigidity; headache; sweating; nausea; facial congestion.

Edwards JJ, et al. Loxoscelism of the eyelids. *Arch Ophthalmol* 1980; 98:1997-2000.

Grant WM. *Toxicology of the eye,* 2nd ed. Springfield, IL: Charles C. Thomas, 1974:37-57.

Reifsnyder DN. Spider bites. *Hosp Pract (Off Ed)* 1994; 29: 15.

Spina Bifida (Rachischisis) 1182

General: Defect of the bony spinal canal without defect of cord or meninges; myelocele sac containing meninges may protrude; failure of neural tube of embryo to close and separate from surface ectoderm.

Ocular: Anophthalmos; microphthalmos; choroidal coloboma; aplasia of retinal ganglion cells and optic nerve; macular aplasia; Homer syndrome; strabismus; lateral rectus palsy; papilledema; optic nerve atrophy.

Clinical: Progressive motor, sensory, vasomotor, and trophic disturbances; hydrocephalus.

Duke-Elder S, MacFaul PA, eds. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Gaston H. Ophthalmic complications of spina bifida and hydrocephalus. *Eye* 1991; 5[Part 3]:279-290.

Welch JP, Aterman K. The syndrome of caudal dysplasia: a review including etiologic considerations and evidence of heterogeneity. *Pediatr Pathol* 1984; 2:313-327.

Spinocerebellar Atrophy with Pupillary Paralysis 1183

General: Autosomal dominant; rare.

Ocular: Absence of pupillary reaction to light or convergence.

Clinical: Spinocerebellar atrophy.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Sutherland JM, et al. Atrophic Spino-Cerebelleuse (HDSC) Familiale avec Mydriase Fixe. *Rev Neurol* 1963; 108: 439-42.

Spinocerebellar Degeneration and Corneal Dystrophy (Corneal Cerebellar Syndrome; Corneal Dystrophy with Spinocerebellar Degeneration) 1184

General: Autosomal recessive; consanguineous parents.

Ocular: Corneal opacification; thickened Descemet membrane; degeneration of pannus; congenital cataracts; myopia; tilted optic disks.

Clinical: Mental retardation; progressive cerebellar abnormalities with variable dorsal column lesions; upper motor neuron involvement; histologic muscle abnormalities.

De Kaloustian VM, et al. Familial spinocerebellar degeneration with corneal dystrophy. *Am J Med Genet* 1985; 20: 325-339.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Mousa AR, et al. Autosomally inherited recessive spastic ataxia, macular dystrophy, congenital cataracts, myopia and vertically oval temporally tilted discs: report of a Bedouin family-a new syndrome. *J Neurol Sci* 1986; 76: 105-121.

Spongy Degeneration of the White Matter (Canavan Disease; Van Bogaert-Bertrand Syndrome) 1185

General: Neurologic disorder of childhood; Jews; familial; autosomal recessive; this is a severe leukodystrophy caused by the deficiency of aspartoacylase (ASPA) and accumulation of N-acetylaspartic acid; a missense mutation recently was identified in the human ASPA coding sequence from patients with this disorder.

Ocular: Optic atrophy; nystagmus; strabismus; roving eye movements.

Clinical: Progressive megalocephaly; psychomotor deterioration; death.

Cogan DG. Ocular manifestations of spongy degeneration. *Birth Defects* 1976; 12:527-534.

Evans OB. Inborn errors of metabolism of the nervous system: Canavan's disease. In: Bradley WG, ed. *Neurology in Clinical Practice*, 2nd ed. Boston: Butterworth-Heinemann, 1995:1506.

Fenichel GM. Spongy degeneration of infancy (Canavan). In: Fenichel GM, ed. *Clinical pediatric neurology*, 2nd ed. Philadelphia: WB Saunders, 1993:136.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Kaul R, et al. Canavan disease: genomic organization and localization of human ASPA to 17p13-ter and conservation of the ASPA gene during evolution. *Genomics* 1994; 21:364-370.

Matalon R, et al. Canavan disease: biochemical and molecular studies. *J Inherit Metab Dis* 1993; 16:744-752.

Sporadic Cretinism (Congenital Hypothyroidism) 1186

General: Variable, from complete lack of thyroid function to reduced function because of enzyme defects; endemic in particular areas (Crete, Beotia, Alpine Valley); affects both sexes; occurs at birth; normal physical and mental development possible with correct treatment.

Ocular: Nystagmus; piglike eyes.

Clinical: Excessive weight; lethargy; facies with heavy expression; large tongue; open mouth; drooling; yellowish tint on cheeks; hypothermia; altered tone of voice; persistent neonatal jaundice; protuberant stomach; umbilical hernia; dry skin; coarse hair; failure to thrive; poor appetite; constipation; cardiomegaly; slow pulse; delayed sexual development; dwarfism; imbecility; reported coexistence with the CHARGE association (bilateral papillary coloboma, congenital heart disease, dysmorphic ears, sensorineural deafness, psychomotor retardation, cryptorchidism, facial palsy, and vesicoureteral reflux).

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Marin JF, et al. The CHARGE association and athyreosis. *J Med Genet* 1991; 28:207-208.

Sporotrichosis 1187

General: Chronic fungal infection caused by *Sporothrix schenckii;* lesion usually occurs on exposed skin and is characterized by nodules or pustules that may develop into small ulcers; infectious agent usually gains entrance into the skin by traumatic implantation of soil or plant materials; disseminated sporotrichosis is uncommon, usually occurring in alcoholics or immunosuppressed patients.

Ocular: Conjunctivitis; keratitis; corneal ulcer; blepharitis; endophthalmitis; iris atrophy; dacryocystitis; osteitis; periosteitis; scleritis; erosion of bony walls of the orbit.

Clinical: Enlargement of regional lymph nodes; pulmonary lesions; granulomas in the joints and genitourinary system.

Agger WA, et al. Ocular sporotrichosis mimicking mucormycosis in a diabetic. *Ann Ophthalmol* 1978; 10:767-771.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Heller HM, Fuhrer J. Disseminated sporotrichosis in patients with AIDS. Case report and review of the literature. *AIDS* 1991; 5:1243-1246.

Mercurio MG, Elewski BE. Therapy of sporotrichosis. *Semin Dermatol* 1993; 12:285-289.

Stern GA, et al. Intravitreal amphotericin B treatment of candida endophthalmitis. *Arch Ophthalmol* 1977; 95:89-93.

Streeten BW; et al. Sporotrichosis of the orbital margin. *Am J Ophthalmol* 1974; 77:750.

Sprengel Syndrome (High Scapula Congenita) 1188

General: Etiology unknown; nonprogressive.

Ocular: Hypertelorism.

Clinical: One scapula short in vertical axis and wider in transverse and closer to the midline than the other scapula; scoliosis; torticollis; vertebral malformations.

Beals RK, et al. Anomalies associated with vertebral malformations. *Spine* 1993; 18:1329-1332.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Sprengel AE. Die Angeborene Verschiebung des Schulterblattes Nach Oben. *Arch Klin Chir (Berlin)* 1891; 42:545-549.

Stanesco Syndrome (Stanesco Dysostosis Syndrome; Osteochondrosis-Osteopetrosis) 1189

General: Autosomal dominant; present from birth; both sexes affected.

Ocular: Exophthalmos.

Clinical: Small stature; brachycephaly; depression at frontoparietal sutures; narrow maxilla; small mandible; crowded teeth; exostoses; fractures.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Stanesco V, et al. Syndrome Hereditaire Dominant Reussissant une Dyostose Cranio-Faciale de Type Particulier, Une Insuffisance de Croissance d' Aspect Chondrodystrophlque et un Epaississement Massif de la Corticale des Os Longs. *Rev Fr Endocrinol Clin* 1963; 4:219-231.

Stannus Cerebellar Syndrome 1190

General: Vitamin B (riboflavin) deficiency.

Ocular: Nystagmus; increased lacrimation; asthenopia; blepharitis; angular conjunctivitis; iris nodules; perilimbal vasodilation and pigmentation; corneal vascularization; superficial- diffuse keratitis; epithelial edema and corneal opacities; cataracts; brownish retinal patches.

Clinical: Muscular asthenia; hypotonia; ataxia; dysdiadochokinesia; mucocutaneous lesions resembling monilial intertrigo and glossitis.

Osuntokun BO, et al. Controlled trial of combinations of hydroxocobalamin-cyst me and riboflavin-cystine, in Nigerian ataxic neuropathy. *J Neurol Neurosurg Psychiatry* 1974; 37:102.

Roe DA. Riboflavin deficiency: mucocutaneous signs of acute and chronic deficiency. *Semin Dermatol* 1991; 10: 293-295.

Stannus HS. Some problems in riboflavin and allied deficiencies. *Br Med J* 1944; 2:103,140.

Staphylococcus 1191

General: Gram-positive coccus *Staphylococcus aureus;* most common cause of suppurative infection in humans; more common in patients with a previous disorder, such as diabetes, thyroid disease, renal failure, or malnutrition; although most *S. aureus* isolates from other sources are encapsulated, capsules have not been noted in ocular isolates.

Ocular: Uveitis; hypopyon; conjunctivitis; keratitis; cellulitis of lid; meibomianitis; ptosis; blepharitis; endophthalmitis; dacryocystitis; increased intraocular pressure; orbital periosteitis.

Clinical: Tissues hypertonic, edematous, and painful; lesion liquefies, forming creamy yellow pus; fever; nausea; vomiting; cough; dyspnea; abdominal pain; diarrhea; bloody stools; dehydration; shock.

Aragones JV. The treatment of blepharitis: a controlled double blind study of combination therapy. *Ann Ophthalmol* 1973; 5:49-52.

Cokingtin CD, Hyndiuk RA. Insights for experimental data on ciprofloxacin in the treatment of bacterial keratitis and ocular infections. *Am J Ophthalmol* 1991; 112:255.

Fox KR, Golomb HS. Staphylococcal ophthalmia neonatorum and the staphylococcal scalded skin syndrome. *Am J Ophthalmol* 1979; 88:1052-1055.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Smolin G, Okumoto M. Staphylococcal blepharitis. *Arch Ophthalmol* 1977; 95:812-816.

Stargardt Disease (Juvenile Macular Degeneration) 1192

General: Onset between ages 8 and 14 years; variable appearance in different families.

Ocular: Heredomacular dystrophy; bilateral lesions showing some degree of symmetry; chorioretinal heredodegeneration; abnormal color vision.

Clinical: Possible association with neurologic deficits, including spastic tetraparesis and cerebellar involvement.

Cibis GV, et al. Dominantly inherited macular dystrophy with flecks (Stargardt). *Arch Ophthalmol* 1980; 98: 1785-1789.

Hadden OB, Gass JDM. Fundus flavimaculatus and Stargardt's disease. *Am J Ophthalmol* 1976; 82:527.

Kalfakis N, et al. Stargardt's disease with neurological involvement: case report. *Funct Neurol* 1994; 9:97-100.

Mantyjarvi M, Tuppurainen K. Color vision in Stargardt's disease. *Int Ophthalmol* 1992; 16:423-428.

Moloney JBM, et al. Retinal function in Stargardt's disease and fundus flavimaculatus. *Am J Ophthalmol* 1983; 96:57-65.

Noble KG, Carr RE. Stargardt's diseases and fundus flavimaculatus. *Arch Ophthalmol* 1979; 97:1281-1285.

Steele-Richardson-Olszewski Syndrome (Progressive Supranuclear Palsy) 1193

General: Nerve cell degeneration centered in the brainstem; resemblance to Lhermitte pyramidopallidal syndrome and to Jakob disease with dementia and rigidity; onset in the sixth decade of life; prominent in males.

Ocular: Supranuclear ophthalmoplegia affecting chiefly vertical gaze, especially downward.

Clinical: Pseudobulbar palsy; dysarthria; dystonic rigidity of neck and upper trunk; axial rigidity; bradykinesia; pyramidal signs; parkinsonism; frontal lobe-type dementia.

Fearnley JM, et al. Diffuse Lewy body disease presenting with a supranuclear gaze palsy. *J Neurol Neurosurg Psychiatry* 1991; 54:159-161.

Mastaglia FL, et al. Progressive supranuclear palsy (the Steele-Richardson-Olszewski syndrome). Clinical and electrophysiological observations in eleven cases. *Proc Aust Assoc Neurol* 1973; 10:35.

Steele JC, Richardson JC, Olszewski J. Progressive supranuclear palsy. *Arch Neurol* 1964; 10:333.

Tolosa E, et al. Clinical diagnosis and diagnostic criteria of progressive supranuclear palsy (Steele-Richardson-Olszewski syndrome). *J Neural Transm Suppl* 1994; 42:15-31.

Stevens-Johnson Syndrome (Dermatostomatitis; Erythema Multiforme Exudativum; Syndroma Mucocutaneo-Oculare; Baader Dermatostomatitis Syndrome; Mucosal-Respiratory Syndrome; Fuchs [2] Syndrome; Mucocutaneous Ocular Syndrome) 1194

General: Etiology unknown; affects all ages; most frequently seen in first and third decades of life; prevalent in males; drugs are the most commonly identified etiologic factor in this condition.

Ocular: Hypopyon; iritis; keratitis; corneal ulcers; keratoconjunctivitis sicca; chemosis; conjunctivitis; widespread fibrinoid necrosis of conjunctival vessels; blepharitis; endophthalmitis; phthisis bulbi; uveitis; cataracts; pannus; optic neuritis; keratoconus; adenoviral conjunctivitis has been reported to have precipitated Stevens-Johnson syndrome; orbital cyst may be a complication.

Clinical: General malaise, headaches, chills, and fever; severe skin and mucous membrane eruptions (erythema multiforme); dorsa of hands and feet are most frequently affected; rhinitis; balanitis; vulvovaginitis; urethritis (nonspecific); cystitis; patients with AIDS are at higher risk of developing Stevens-Johnson syndrome.

Arstikaitis MJ. Ocular aftermath of Stevens-Johnson syndrome. *Arch Ophthalmol* 1973; 90:376.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Gottschalk HR, Stone OJ. Stevens-Johnson syndrome from ophthalmic sulfonamide. *Arch Dermatol* 1976; 112: 513-514.

Hodge WG, et al. Adenoviral keratoconjunctivitis precipitating Stevens-Johnson syndrome. *Can J Ophthalmol* 1994; 29:198-200.

Kaido M, Goto E, Dogru M, et al. Punctal occlusion in the management of chronic Stevens-Johnson syndrome. *Ophthalmology* 2004; 111: 895-900.

Power WJ, et al. Analysis of the acute ophthalmic manifestations of the erythema multiforme/Stevens-Johnson syndrome/toxic epidermal necrolysis disease spectrum. *Ophthalmology* 1995; 102:1669-1676.

Stevens AM, Johnson CF. A new eruptive fever associated with stomatitis and ophthalmia. *Am J Dis Child* 1922; 24:526.

Tabbara KF, Shammas HF. Bilateral corneal perforations in Stevens-Johnson syndrome. *Can J Ophthalmol* 1975; 10: 514-517.

Stickler Syndrome (Hereditary Progressive Arthroophthalmopathy) 1195

General: Autosomal dominant; onset in childhood; severe and debilitating connective tissue disorder inherited as an autosomal dominant syndrome with a variable phenotype; linkage analysis has provided statistical evidence for linkage of collagen type II (COL2A1) gene with this syndrome in some but not all families.

Ocular: Phthisis bulbi; glaucoma; chronic uveitis; keratopathy; complicated cataracts; chorioretinal degeneration; total retinal detachment during first decade of life; myopia; giant retinal tears.

Clinical: Bony enlargement of joints with abnormal development of the articular surfaces and premature degenerative changes; hypermobility of joints with abnormality in connective tissues supporting the joints; possible skeletal deformities.

Ahmad NN, et al. Stickler syndrome. A mutation in the non-helical 3' end of type II procollagen gene. *Arch Ophthalmol* 1995; 113:1454-1457.

Hall J. Stickler's syndrome presenting as a syndrome of cleft palate, myopia and blindness inherited as a dominant trait. *Birth Defects* 1974; 10: 157-171.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Schreiner RL, et al. Stickler's syndrome in a pedigree of Pierre-Robin syndrome. *Am J Dis Child* 1973; 126:86.

Spallone A. Stickler's syndrome. A study of 12 families. *Br J Ophthalmol* 1987; 71:504.

Stickler GB, et al. Hereditary progressive arthro-*Ophthalmology*. *Mayo Clin Proc* 1965; 40:433.

Vlutiner GM, et al. Genetic and *Clinical* heterogeneity of stickler syndrome. *Am J Med Genet* 1991; 41:44-48.

Vu CD, Brown J, Korkko J, et al. Posterior chorioretinal atrophy and vitreous phenotype in a family with Stickler syndrome from a mutation in COL2A1 gene. *Ophthalmology* 2003; 110: 70-77.

Zlotogora J, et al. Variability of Stickler syndrome. *Am J Med Genet* 1992; 42:337-339?

Strachan Syndrome (Tropical Nutritional Neuropathy) 1196

General: Possibly nutritional vitamin A deficiency, chronic cyanide poisoning, and/or infectious agents.

Ocular: Optic atrophy; blurred vision, usually bilateral and symmetric; retrobulbar neuritis; scotoma; decreased vision; bilateral and symmetrical central or cecocentral scotomata; loss of color vision due to selective lesion of the maculopapillary bundles.

Clinical: Ataxic neuropathy; sensorineural deafness; high prevalence of goiter.

Dreyfus PM. Nutritional disorder of obscure etiology. *Med Sci* 1966; 17:44-48.

Strachan H. On a form of multiple neuritis prevalent in the West Indies. *Practitioner (London)* 1897; 59:477-484.

Straw Peter Syndrome (Slovenly Peter Syndrome; Struwwelpeter Syndrome) 1197

General: Etiology unknown; combination of organic and environmental factors; affects 5% to 20% of the general school population; in preschool-aged children, manifested as clumsiness, abnormal activity level, and disorganized thought process; in school-aged children, manifested as distractibility and learning disability.

Ocular: Strabismus; impaired visual perception.

Clinical: Hyperkinesia; restlessness; fidgetiness; disorganized thought process; hypokinesia; impulsivity; poor coordination of fingers; motor awkwardness; impaired auditory perception.

Clements SD, Peter JE. Minimal brain dysfunctions in school-age child. *Arch Gen Psychiatry* 1962; 6:185-197.

Hoffmann H. "Struwwelpeter." *JAMA* 1967; 202:28-29.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Streptococcus (Scarlet Fever) 1198

General: Gram-positive bacteria that can invade any tissue.

Ocular: Conjunctivitis; corneal ulcer; blepharitis; scarlatinal rash of lid; erysipelas dermatitis of lid; gangrene of lid; endophthalmitis; proptosis; dacryocystitis; optic neuritis; orbital cellulitis; uveitis; hypopyon; secondary glaucoma; paralysis of extraocular muscles; infectious crystalline keratopathy; scleritis.

Clinical: Pharyngitis; impetigo; scarlet fever; pneumonia; bacteremia; rheumatic fever; glomerulonephritis.

Altman AJ, et al. Scleritis and Streptococcus pneumoniae. *Cornea* 1991; 10:341-345.

Matoba AY, et al. Infectious crystalline keratopathy due to Streptococcus pneumoniae. Possible association with serotype. *Ophthalmology* 1994; 101:1000-1004.

Ostler HB, Okumoto M. Anaerobic streptococcal corneal ulcer. *Am J Ophthalmol* 1976; 81:518-519.

Peter G, Smith AL. Group A streptococcal infections of skin and pharynx. *N Engl J Med* 1977; 297:311-317.

String Syndrome 1199

General: Following encircling operations or circular diathermy, chemosis, and excess protein exudation into the anterior chamber; onset between postoperative days 4 and 19; suture causing vascular obstruction; configuration of eye is a predisposing factor.

Ocular: Necrosis of iris; necrosis of ciliary body; corneal ring abscess; iritis; deep anterior chamber; proptosis; lid edema; chemosis of conjunctiva; ocular hypertension; iris assumes a green color; retinal detachment.

Clinical: None.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Mason N. The string syndrome: seen as a complication of Arruga's cerclage suture. *Br J Ophthalmol* 1964; 48:70-74.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Strumpell-Leichtenstern Syndrome (Acute Hemorrhagic Encephalitis) 1200

General: Etiology viral, postvaccinal, drug-induced, or allergic; both sexes affected; onset at all ages but prevalent in children.

Ocular: Optic atrophy; acute retinal necrosis syndrome.

Clinical: Fever; convulsions; mental dullness; delirium; coma; ataxia; neck rigidity; tachypnea; myoclonus; aphasia; acute disseminated encephalomyelitis.

Ahmadieh H, et al. Association of herpetic encephalitis with acute retinal necrosis syndrome. *Ann Ophthalmol* 1991; 23:215-219.

Dangond F, et al. Acute disseminated encephalomyelitis progressing to hemorrhagic encephalitis. *Neurology* 1991; 41:1697-1698.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

von Strumpell A. Ueber Primaere Acute Encephlitis. *Dtsch Arch Klin Med* 1890; 47:53-74.

Sturge-Weber Syndrome (Meningocutaneous Syndrome; Vascular Encephalotrigeminal Syndrome; Neuro-Oculocutaneous Angiomatosis; Encephalofacial Angiomatosis; Encephalotrigeminal Syndrome) 1201

General: Trisomy 22 or partial trisomy inheritance. Variations include Jahnke syndrome (neuro-oculocutaneous angiomatosis without glaucoma), Schirmer syndrome (oculocutaneous angiomatosis with early glaucoma), Lawford syndrome (oculocutaneous angiomatosis with late glaucoma and no increase in volume of globe), and Mille syndrome (oculocutaneous syndrome with choroidal angioma but no glaucoma).

Ocular: Unilateral hydrophthalmos; secondary glaucoma (late) conjunctival angiomata (telangiectases); iris decoloration; nevoid marks or vascular dilation of the episclera; glioma; serous retinal detachment; choroidal angiomata; deep anterior chamber angle; port-wine stain of eyelid; buphthalmos; optic nerve cupping; anisometropia; hemianopsia; increased corneal diameter; enophthalmos; exophthalmos; optic atrophy; choroidal hemangioma; anterior chamber angle vascularization.

Clinical: Vascular port-wine nevus (face, scalp, limbs, trunk, leptomeninges); acromegaly; facial hemihypertrophy; intracranial angiomas; convulsion; mental retardation; obesity; limb atrophy.

Cibis GW, et al. Glaucoma in Sturge-Weber syndrome. *Ophthalmology* 1984; 91:1061-1071.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 2000.

Mwinula JH, et al. Anterior chamber angle vascularization in Sturge-Weber syndrome. Report of a case. *Graefes Arch Clin Exp Ophthalmol* 1994; 232:387-391.

Stevenson RF, Morin JD. Ocular findings in nevus flammeus. *Can J Ophthalmol* 1975; 10:136-139.

Sturge WA. A case of partial epilepsy, apparently due to a lesion of one of the vasomotor centers of the brain. *Trans Clin Soc Lond* 1879; 12:1962.

Sullivan TJ, et al. The ocular manifestations of the Sturge-Weber syndrome. *J Pediatr Ophthalmol Strabismus* 1992; 29:349-356.

Susac JO, et al. The "tomato-catsup" fundus in Sturge-Weber syndrome. *Arch Ophthalmol* 1974; 92:69.

Weber FP. A Note on the association of extensive haemangiomatous naevus of the skin with cerebral (meningeal) haemangioma. *Proc Roy Soc Med* 1929; 22:25.

Subacute Bacterial Endocarditis 1202

General: Inflammation of the lining of the heart typically caused by *Staphylococcus epidermidis.*

Ocular: Optic neuritis; papillitis; choroiditis; conjunctival and retinal petechiae; retinal hemorrhages; Roth spots; floaters in aqueous and vitreous; rare embolic occlusion of the central retinal artery; ophthalmoplegia; papilledema.

Clinical: Fever; anemia; splenomegaly; heart murmur.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Gelfand MS, Threkeld MG. Subacute bacterial endocarditis secondary to Streptococcus pneumoniae. *Am J Med* 1992; 93:91-93.

Hermans PE. The *Clinical* manifestations of infective endocarditis. *Mayo Clin Proc* 1982; 57:15-21.

Kitts D, et al. Septic embolism complicating infective endocarditis. *J Vasc Surg* 1991; 14:480.

Manor RS, et al. Prepapillary metastatic abscess in a case of subacute bacterial endocarditis. *Ophthalmologica* 1975; 170:22-29.

Subclavian Steal Syndrome 1203

General: Reversal of blood flow through the vertebral artery caused by stenosis of one subclavian artery proximal to the origin of the vertebral artery; arteriosclerosis; atresia of the proximal subclavian artery; aortic coarctation; subclavian artery may siphon (steal) blood from the vertebral artery; causes fluctuating symptoms of basilar artery insufficiency.

Ocular: Ptosis; nystagmus; temporary visual loss during activity of one arm; visual disturbances.

Clinical: Brachial vascular insufficiency; claudication; tingling of the arm; syncope; decrease of blood pressure in affected arm by at least 20 mm Hg; numbness; coldness; pain; facial paresthesia; headache; syncopal attacks; vertigo; intermittent claudication of the involved upper extremity.

Breall JA, et al. Atherectomy of the subclavian artery for patients with symptomatic coronary-subclavian steal syndrome. *J Am Coil Cardiol* 1993; 21:1564-1567.

Delaney CP, et al. Investigation and management of subclavian steal syndrome. *Br J Surg* 1994; 81: 1093-1 095.

Dwyer-Joyce P. The fields in subclavian steal. *Trans Ophthalmol Soc UK* 1972; 92:819.

Garland JA. New vascular syndrome-the "subclavian steal." *N Engl J Med* 1961; 265:912.

Mannick JA, et al. The "subclavian steal" syndrome: a further documentation. *JAMA* 1962; 182:254.

Milliare A, et al. Subclavian angioplasty immediate and late results in 50 patients. *Cathet Cardiovasc Diagn* 1993; 29:8-17.

Smith JM. Subclavian steal syndrome. A review of 59 consecutive cases. *J Cardiovasc Surg (Torino)* 1994; 35: 11-14.

Submandibular, Ocular, and Rectal Pain with Flushing 1204

General: Autosomal dominant; brief, severe pain of submandibular, ocular, and rectal areas with flushing of surrounding skin.

Ocular: Ocular pain.

Clinical: Jaw aches; severe rectal pain.

Haydon R, et al. Rectal ocular and submaxillary pain: a familiar autonomic disorder related to proctalgia fugax. *Am J Dis Child* 1959; 97:479-482.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Sulfite Oxidase Deficiency 1205

General: Rare abnormality of sulfur metabolism in which there is an accumulation of S-sulfocysteine, thiosulfate, and sulfate and a deficiency of sulfite; present at birth; death occurs before age 3 years.

Ocular: Dislocated lens; nystagmus; ectopia lentis.

Clinical: Muscular rigidity; eccentric behavior; coarse face; broad nasal bridge; long philtrum (in association with Leigh syndrome).

Amiel J, et al. Deficit en sulfite oxydase se presentat comme un syndrome de Leigh. *Arch Pediatr* 1994; 1: 1023-1027.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Harley RD, ed. *Pediatric Ophthalmology*, 4th ed. Philadelphia: WB Saunders, 1998.

Sunrise Syndrome 1206

General: Occurs when the intraocular lens (IOL) optic is displaced superiorly out of the visual axis.

Ocular: Edge of IOL in pupil; decreased visual acuity; glare.

Clinical: None.

Akkin C, et al. Tilt and decentration of bag-fixated intraocular lenses: a comparative study between capsulorhexis and envelope techniques. *Doc Ophthalmol* 1994; 87:199-209.

Davison JA. Capsule contraction syndrome. *J Cataract Refract Surg* 1993; 19:582-589.

Smiddy WE, et al. Surgical management of dislocated intraocular lenses. *J Cataract Refract Surg* 1995; 21:64-69.

Smith SG, Lindstrom RL. Malpositioned posterior chamber lenses: etiology, prevention and management. *Am Intraocul Implant Soc J* 1985; 11:584-591.

Smith SG, Lindstrom RL. Report and management of the sunrise syndrome. *Am Intraocul Implant Soc J* 1984; 7: 55-57.

Wilson DJ, et al. Effects of extracapsular cataract extraction on the lens zonules. *Ophthalmology* 1987; 94:467-470.

Sunset Syndrome 1207

General: Occurs when the capsule or zonules have been sufficiently damaged to allow the posterior chamber IOL to slip gradually into the inferior vitreous in the postoperative period.

Ocular: Superior edge of IOL in pupil; IOL in vitreous body; retinal detachment; decreased visual acuity; glare; zonular disinsertions.

Clinical: None.

Akkin C, et al. Tilt and decentration of bag-fixated intraocular lenses: a comparative study between capsulorhexis and envelope techniques. *Doc Ophthalmol* 1994; 87:199-209.

Allara GW, Weinstein GWA new surgical technique for managing sunset syndrome. *Ophthalmic Surg* 1987; 18:811.

Davison JA. Capsule contraction syndrome. *J Cataract Refract Surg* 1993; 19:582-589.

Smiddy WE, et al. Surgical management of dislocated intraocular lenses. *J Cataract Refract Surg* 1995; 21:64-69.

Smith SG, Lindstrom RL. Malpositioned posterior chamber lenses: etiology, prevention and management. *Am Intraocul Implant Soc J* 1985; 11:584-591.

Superior Vena Cava Syndrome (Vena Cava Superior Syndrome) 1208

General: Compression or obstruction of the superior vena cava by aortic aneurysms, mediastinal neoplasms, thyroid adenoma, or carcinoma of the lung; found in males in their 50s.

Ocular: Glaucoma; conjunctival vasodilation; retinal hemorrhages (Valsalva retinopathy); eyelid edema; optic disk edema; engorgement of the conjunctival and episcleral vessels; periorbital edema.

Clinical: Cyanosis and edema of face, neck, and upper trunk with a rather sharp demarcation line (short cape edema); dysphagia; epistaxis; hoarseness; vertigo; tinnitus; may be caused by Graves disease and as a presentation of Behçet disease.

Collins JF. *Handbook of Clinical Ophthalmology* New York: Masson, 1982.

Dyken JR, et al. Superior vena cava syndrome presenting as periorbital edema. *J Am Acad Dermatol* 1994; 31 [2 Part 1]:281-283.

Ishihara T, et al. Superior vena cava syndrome due to Graves' disease. *Intern Med* 1993; 32:80-83.

Lokich JJ, Goodman R. Superior vena cava syndrome: *Clinical* management. *JAMA* 1975; 231:58.

McArt BA, et al. Surgical reversal of superior vena cava syndrome: report of a case caused by intrathoracic goiter and associated with roentgenographic hilar vascular shadow simulating neoplasm of chest. *Arch Surg* 1954; 69:4.

Thomas I, et al. Behcet's disease presenting as superior vena cava syndrome. *J Am Acad Dermatol* 1992; 26[5 Part 2]:863-865.

Suprarenal Sympathetic Syndrome (Pheochromocytoma Syndrome; Adrenal Sympathetic Syndrome; Adrenal Medulla Tumor Syndrome) 1209

General: Tumors producing increased secretion of norepinephrine and epinephrine deriving from chromaffin cells of the adrenal medulla; more common in males (3:2); symptoms occur in paroxysms or attacks; precipitated by emotional upsets; predilection for the right adrenal.

Ocular: Spasm of retinal arteries with associated cotton-wool exudates; flame-shaped hemorrhages; papilledema; pupillary dilation; neovascularization of retina; following removal of the tumor there may be macular scarring and optic atrophy.

Clinical: Hypertension; tachycardia; severe anxiety; headache; nervous tension; sweating; pallor; nausea; polyuria; polydipsia; association with neurofibromatosis and von Hippel disease.

Bruce GM. Changes in the ocular fundus associated with pheochromocytoma of the adrenal gland. *Arch Ophthalmol* 1948; 39:707.

Lowden BA, Harris GS. Pheochromocytoma and von Hippel-Lindau's disease. *Can J Ophthalmol* 1976; 11:282-289.

Maher ER, et al. Clinical features and natural history of von Hippel-Lindau disease. *Q J Med* 1990; 66:233.

Modlin IM, et al. Phaechromocytoma in 72 patients: *Clinical* and diagnostic features, treatment and long-term results. *Br J Surg* 1979; 66:456.

Saadat H, Bahrami V. Blindness: a postoperative complication of pheochromocytoma. *Va Med* 1977; 104:38-40.

Susac Syndrome 1210

General: Rare; unknown origin; characterized by the triad of encephalopathy, fluctuation hearing loss and visual loass resulting from microangiopathy of the brain, cochlea and retina.

Ocular: Cotton wool spot; central retinal vein occlusion;

Clinical: Hearing loss; encephalopathy

Gross M, Banin E, Eliashar R, et al. Susac syndrome. *Otol Neurotol* 2004; 25: 470-473.

Xu MS, Tan CB, Umapathi T, et al. Susac syndrome: serial diffusion-weighted MR imaging. *Magn Reson Imaging* 2004; 22: 1295-1298

Sweet Syndrome (Acute Febrile Neutrophilic Dermatosis) 1211

General: Cause unknown; common in middle-aged women; associated with acute leukemia; acute febrile neutrophilic dermatosis; neurologic symptoms.

Ocular: Conjunctivitis; episcleritis; glaucoma; limbal nodules.

Clinical: Cutaneous eruption with fever; nondeforming, asymmetrical, large-joint arthritis; albuminuria; anemia; oral aphthae; genital ulcers; association with Behçet disease has been reported.

Chen TC, Goldstein DA Quinn JP, et al. Scleritis associated with acute febrile neutrophilic dermatosis (Sweet’s syndrome). Brit J Ophthalmol. 1998; 82: 328-329.

Cohen PR. Sweet's syndrome presenting as conjunctivitis. *Arch Ophthalmol* 1993; 111:587-588.

Fourman S. Inflammatory glaucoma associated with Sweet's syndrome. *Am J Ophthalmol* 1988; 105:691-692.

Medenblik-Frysch S, et al. Ocular complications in Sweet's syndrome. *Am J Ophthalmol* 1992; 114:210-231.

Sweet RD. Acute febrile neutrophilic dermatosis. *Br J Dermatol* 1964; 76:349-356.

Uysal H, et al. Acute febrile neutrophilic dermatosis (Sweet's syndrome) in neuro-Behcet's disease. *Clin Neurol Neurosurg* 1993; 9594:319-322.

Sylvester Disease 1212

General: Dominant inheritance.

Ocular: Optic atrophy.

Clinical: Ataxia; moderate and slowly progressive hearing loss.

Konigsmark BW, et al. Dominant congenital deafness and progressive optic nerve atrophy. *Arch Ophthalmol* 1974; 91:99-103.

Symonds Syndrome (Otitic Hydrocephalus Syndrome; Serous Meningitis Syndrome; Benign Intracranial Hypertension; Pseudotumor Cerebri) 1213

General: Children and adolescents; protracted course; increased cerebrospinal fluid, but without increase in protein or cells.

Ocular: Sixth nerve palsy, ipsilateral side with otitis media; retinal hemorrhages and exudates; moderate-to-marked papilledema followed by secondary optic atrophy; unilateral or bilateral swelling of the optic nerve head have been reported; cranial nerve III and IV involvement; bilateral retinal vein occlusion.

Clinical: Greatly increased pressure of spinal fluid, often greater than 300 mm, without increased cells or protein; intermittent headaches; otitis media; chronic renal failure; chronic myeloid leukemia.

Chang D, et al. Benign intracranial hypertension and chronic renal failure. *Cleve Clin J Med* 1992; 59:419-422.

Chari C, Rao NS. Benign intracranial hypertension-its unusual manifestations. *Headache* 1991; 31:599-600.

Chern S, et al. Bilateral central retinal vein occlusion as an initial manifestation of pseudotumor cerebri. *Ann Ophthalmol* 199l; 23:54-57.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Venable HP. Pseudo-tumor cerebri. *J Natl Med Assoc* 1970; 62:435-440.

Venable HP. Pseudo-tumor cerebri: further studies. *J Natl Med Assoc* 1973; 65:194-197.

Sympathetic Ophthalmia 1214

General: Trauma or injury to one eye and later onset of inflammation in the other eye.

Ocular: Iridocyclitis (acute inflammation of iris, ciliary body, and anterior chamber); choroiditis; chronic persistent keratitic precipitates; posterior synechiae; phthisis bulbi; has been reported following laser cyclocoagulation.

Clinical: None.

Bechrakis NE, et al. Sympathetic ophthalmia following laser cyclocoagulation. *Arch Ophthalmol* 1994; 112:80-84.

Bonuik J. The incidence of phthisis bulbi as a complication of cataract surgery in congenital rubella syndrome. *Trans Am Acad Ophthalmol Otolaryngol* 1970; 74:360.

Duane TD. *Clinical Ophthalmology*. Philadelphia: JB Lippincott, 1987.

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Takayasu Syndrome (Martorell Syndrome; Aortic Arch Syndrome; Pulseless Disease; Reversed Coarctation Syndrome) 1215

General: Two types are (i) occlusive inflammatory lesion (seen in young Japanese women) and (ii) occlusive vascular disease without inflammation, associated with atherosclerosis and syphilis; onset in fifth and sixth decades; both sexes affected; can involve the aorta and its major branches as well as the coronary, hepatic, mesenteric, pulmonary, and renal arteries.

Ocular: Iris atrophy; cataracts; retinal microaneurysms; sausage-shaped venous dilations; reduced central retinal artery pressure; optic atrophy; cotton-wool spots; anterior segment ischemia; retinal arteriovenous shunts.

Clinical: Diminished or absent pulsation of arteries (head, neck, upper limbs); orthostatic syncope; facial atrophy; epileptiform seizures; intermittent claudication.

Font TG, Naumann G. Ocular histopathology in pulseless disease. *Arch Ophthalmol* 1969; 877:784.

Koo J, Boldrey EE. Takayasu's disease. *Arch Ophthalmol* 2000; 118:858-859.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Takayasu S. A case with unusual changes in the central vessels of the retina. *Acta Soc Ophthalmol Jpn* 1908; 12:554.

Tananka T, Shimizu K. Retinal arteriovenous shunts in Takayasu disease. *Ophthalmology* 1987; 94:1380-1388.

Winiecki MW Takayasu's arteritis: a case study. *AANA J* 1993; 61:133-136.

Tangier Syndrome (Lipoprotein Deficiency; Familial High-Density Lipoprotein Deficiency; α-Lipoprotein Deficiency; Fish Eye Disease) 1216

General: First seen on Chesapeake Bay Island; rare; autosomal recessive; inability to synthesize polypeptide required in the elaboration of high-density lipoprotein; choles-terol esters are stored; occurs in both sexes; onset from childhood to fifth decade of life; disorder appears to be a variant of familial lecithin-cholesterol acyltransferase (LCAT) deficiency in which the enzyme remains partly active.

Ocular: Corneal infiltrates; fine, dotted stromal opacities, most marked in posterior central third of corneal stroma; wasting of orbicularis oculi muscle.

Clinical: Maculopapular rash; orange-yellow striped tonsils; hepatosplenomegaly; lymphadenopathy; intermittent diarrhea; bilateral motor weakness.

Chu FC, et al. Ocular manifestations of familial high-density lipoprotein deficiency (Tangier disease). *Arch Ophthalmol* 1979; 97:1926-1928.

Hoffman HN, Fredrickson DS. Tangier disease (familial high density lipoprotein deficiency). Clinical and genetic features in two adults. *Am J Med* 1965; 39:582.

Kastelein JJ, et al. Familial high density-lipoprotein deficiency causing corneal opacities (fish eye disease) in a family of Dutch descent. *J Intern Med* 1992; 231:413-419.

Tapetal-Like Reflex Syndrome 1217

General: Rare.

Ocular: Ring scotoma; discrete bright yellow spots in posterior polar region deep to the retinal vessels; tapetal-like reflex and retinitis pigmentosa may be present in members of the same family.

Clinical: None.

Ciccarelli EC. A new syndrome of tapetal-like fundic reflexes with ring scotoma. *Arch Ophthalmol* 1962; 67:316.

Falls HF, Cotterman CW. Choroidal degeneration. *Arch Ophthalmol* 1948; 40:685.

TAR Syndrome (Thrombocytopenia Absent Radius Syndrome) 1218

General: Bilateral absence of the radius and hypomegakaryocytic thrombocytopenia.

Ocular: May have cataracts, glaucoma, megalocornea, and blue sclera.

Clinical: Patients have foreshortened forearms and radially deviated hands; infrequently associated with mental retardation (7%); also may have lower extremity deformity.

Gounder DS, et al. Clinical manifestations of the thrombocytopenia and absent radii (TAR) syndrome. *Aust N* Z *J Med* 1989; 19:479-482.

MacDonald MR, et al. Hypoplasia of the cerebellar vermis and corpus callosum in thrombocytopenia with absent radius syndrome on MR1 studies. *Am J Med Genet* 1994; 50:46-50.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders,* 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Tay-Sachs Syndrome (Norman-Wood Syndrome; Familial Amaurotic Idiocy; Gangliosidosis G_{M2} Type 1; Hexosaminidase Deficiency) 1219

General: Similar to ceroid lipofuscinosis; autosomal recessive; occasional dominant inheritance; onset birth to 10 months; affects Jewish females; death during first 2 years; stored ganglioside (see Ceroid Lipofuscinosis); decreased hexosaminidase A localized to chromosome 15 (15q22-15q25.1).

Ocular: Nystagmus; strabismus; whitish-gray macular area with cherry-red spot in the center; retinal pigmentary changes with involvement of the macula occasionally may be seen instead of the typical red spot; grayish coloration of the macula is due to

the swollen ganglion cells in the perifoveal macular region; retinal vessels become narrowed; progressive ascending optic atrophy; cortical blindness by age 12 to 18 months with reactive pupils; deterioration of ocular motor function.

Clinical: Hyperacusis; mental retardation; convulsions; muscles, initially flaccid, becoming spastic with progression; infants normal at birth but fail to thrive after 4 to 8 months; hypotonia; death; occurs primarily in Jewish children; biochemical heterogeneity; absence of hexosaminidase A most common (type 1); absence of hexosaminidase A and B in Sandhoff variant (type 2); feeding difficulties; doll-like facies; fine hair; macrocephaly; abnormal acousticomotor reaction.

Gravel RA, et al. Biochemistry and genetics of Tay-Sachs disease. *Can J Neurol Sci* 1991; 18[3 Suppl]:419-423.

Honda Y. Sudo M. Electroretinogram and visually evoked cortical potential in Tay-Sachs' disease: a report of two cases. *J Pediatr Ophthalmol* 1976; 13:226-229.

Nagashima K, et al. Retinal amacrine cell involvement in Tay-Sachs disease. *Acta Neuropathol* 1981; 53: 333-633.

Sachs B. An arrested cerebral development with special reference to its cortical pathology. *J Nerv Ment Dis* 1887; 14:541.

Schmitt HP. Peripheral intraaxonal storage in Tay-Sachs' disease (GM2-gangliosidosis type 1). *J Neural Sci* 1979; 44: 115-124.

Smith LH. Inherited metabolic disease with pediatric ocular manifestations. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. IV.* Philadelphia: WB Saunders, 1994:2777-2790.

Tay W. Symmetrical changes in the yellow spot in each eye of an infant. *Trans Ophthalmol Soc UK* 1881; 1:55.

Temporal Arteritis Syndrome (Cranial Arteritis Syndrome; Giant Cell Arteritis; Hutchinson-Horton-Magath-Brown Syndrome) 1220

General: Etiology unknown; mainly females; mainly whites; ages 55 to 80 years; temporal artery shows inflammatory thickening; arteritis of the vessels supplying the optic nerve.

Ocular: Transient ptosis; partial or complete loss of vision on the affected side; retinal detachment; exudates and hemorrhages; narrowing of retinal vessels; obstruction of the central retinal artery; optic atrophy; ischemic optic neuropathy; acute decreased intraocular pressure; corneal hypesthesia; palsies of extraocular muscles; hemorrhagic glaucoma; diplopia; hemorrhages on or around the disk.

Clinical: Throbbing headache; hyperalgesia of the scalp; malaise; anorexia; weakness; weight loss; fever; nodular pulmonary nodules; cough; otitis with deafness.

Cullen JF, Coleiro JA. Ophthalmic complications of giant cell arteritis. *Surv Ophthalmol* 1976; 20:247-260.

Cullen JF. Occult temporal arteritis. *Br J Ophthalmol* 1967; 51:513.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Healey LA, Wilske KR. Manifestations of giant cell arteritis. *Med Clin North Am* 1977; 61:261-270.

Sorensen S, Lorenzen I. Giant-cell arteritis, temporal arteritis and polymyalgia rheumatica. *Acta Med Scand* 1977; 201:207-213.

Zenone T, et al. Unusual manifestations of giant cell arteritis: pulmonary nodules, cough, conjunctivitis and otitis with deafness. *Eur Respir J* 1994; 7:2252-2254.

10q-Syndrome (10q Deletion Syndrome) 1221

General: Chromosome 10q deletion syndrome.

Ocular: Microphthalmia.

Clinical: Intrauterine growth retardation; microcephaly; truncus arteriosus type I; respiratory distress; craniofacial dysmorphism.

Glanz AS, et al. Lenz microphthalmia: a malformation syndrome with variable expression of multiple congenital anomalies. *Can J Ophthalmol* 1983; 18:41-44.

Lewandowski RC, et al. Partial deletion 10q. *Hum Genet* 1978; 42:339-343.

Taysi K, et al. Terminal deletion of the long arm of chromosome 10:q26 to qter. Case report and review of literature. *Ann Genet* 1982; 25:141-144.

Terrien Disease (Terrien Marginal Degeneration; Gutter Dystrophy; Peripheral Furrow Keratitis; Senile Marginal Atrophy) 1222

General: Rare; no known cause; 75% of patients are males from age 10 to 70 years.

Ocular: Usually bilateral; may be asymmetrical; peripheral, fine, yellow-white, punctate stromal opacities associated with mild, superficial corneal vascularization; progressive thinning leads to peripheral gutter formation; decrease in visual acuity; loss of Bowman membrane and anterior stromal lamella with partial replacement of these tissues by a vascularized connective tissue; fatty deposits; thin stroma; thickness changes in Descemet membrane; regular recurring attacks of pain and inflammation; keratoconus; reported association with Terrien marginal degeneration.

Clinical: None.

Ashenhurst M, Slomovic A. Corneal hydrops in Terrien's marginal degeneration: an unusual complication. *Can J Ophthalmol* 1987; 22:328.

Austin P, Brown SI. Inflammatory Terrien's marginal corneal disease. *Am J Ophthalmol* 1981; 92: 189-192.

Friedlaender MH. *Allergy and immunology of the eye*. Hagerstown, MD: Harper & Row, 1979:190.

Kremer I. Terrien's marginal degeneration associated with vernal conjunctivitis. *Am J Ophthalmol* 1991; 111: 517-518.

Lopez JS, et al. Immunohistochemistry of Terrien's and Mooren's corneal degeneration. *Arch Ophthalmol* 1991; 109: 988-992.

Terson Syndrome (Subarachnoid Hemorrhage Syndrome) 1223

General: Spontaneous rupture of aneurysm or traumatic intracerebral hemorrhage; onset at all ages.

Ocular: Weakness of extraocular muscles; disarranged and uncoordinated gaze; severe intraocular hemorrhage; preretinal hemorrhages; peripapillary hemorrhages; papilledema secondary to optic nerve sheath hemorrhages; pigmentary changes in macula and retina; preretinal membrane formation; vitreous detachment; amblyopia; anisocoria; bilateral retinal detachments have been associated with this disorder; epiretinal membranes (sequelae).

Clinical: Sudden unconsciousness; elevated cerebrospinal fluid pressure.

Fahmy JA. Vitreous hemorrhage in subarachnoid hemorrhage-Terson's syndrome: report of a case with macular degeneration as a complication. *Acta Ophthalmol* 1972; 50:137.

Garcia-Arumi J, et al. Epiretinal membranes in Terson's syndrome. A clinicopathologic study. *Retina* 1994; 14: 351-355.

McRae M, et al. Bilateral retinal detachments associated with Terson's syndrome. *Retina* 1994; 14:467-469.

Terson LL. Lesions of the fundus associated with brain hemorrhage. *Arch Neural Psychiatry* 1939; 42:664.

Toosi SH, Malton M. Terson's syndrome: significance of ocular findings. *Ann Ophthalmol* 1987; 19:656-660.

Weingeist TA, et al. Terson's syndrome. *Ophthalmology* 1986; 93:1435-1442.

Thalasselis Syndrome 1224

General: Keratoconus-tetany-menopause.

Ocular: Spontaneous keratoconus.

Clinical: Syndrome describes an association between hormone deficiency, magnesium deficiency, allergy, keratoconus, and type B behavior.

Rabinowitz YS. Major review: keratoconus. *Surv Ophthalmol* 1998; 42:297-319.

Thalasselis A, Selin AA. Keratoconus-tetany-menopause: the new association. *Optom Vis Sci* 1991; 68:357-363.

Thanatophoric Dwarfism 1225

General: Etiology unknown, but autosomal recessive inheritance is suspected; seen most frequently in males; onset in fetal life; death usually within first 3 days of life; possible association with drug administration has been reported.

Ocular: Exophthalmos.

Clinical: Head enlarged; small face; enlarged fontanelles; high forehead; frontal bossing; saddle nose; abdomen protuberant; thorax narrow; short rib; hypotonia; respiratory distress; cardiac failure; marked shortening of the extremities; macrocephaly.

Kassanos D, et al. Prenatal sonographic diagnosis of thanatophoric dwarfism. *Int J Gynaecol Obstet* 1991; 34: 373-376.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981. Maroteaux, P., et al. Lenanisme Thanatophore. *Presse Med* 1967; 775:2519-2522.

Rafla NM, Meehan FP. Thanatophoric dwarfism; drugs and antenatal; a case report. *Eur J Obstet Gynecol Biol* 1991; 38:161-165.

Thelaziasis 1226

General: Ocular infection caused mainly by the nematode *Thelazia callipardon;* natural habitat is the lacrimal gland of the dog.

Ocular: Uveitis; parasites in the conjunctiva, cornea, lacrimal system, and anterior chamber; corneal opacity; corneal abrasion; increased tear secretion.

Clinical: None.

Duke-Elder S, ed. *System of Ophthalmology, vol VIII* St. Louis: CV Mosby, 1976: 193.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th Ed. Philadelphia: WB Saunders, 2000.

Schultz GR. Intraocular nematode in man. *Am J Ophthalmol* 1970; 70:826-829.

Singh TS, Singh KN. Thelaziasis: report of two cases. *Br J Ophthalmol* 1993; 77:528-529.

Thermal Burns 1227

General: May occur to any body tissue.

Ocular: Conjunctival necrosis; corneal ulcer; exposure keratitis; ectropion; contracture deformity of lids; lid edema; entropion; endophthalmitis; proptosis; dacryocystitis; chronic epiphora; cellulitis; corneal perforation; symblepharon.

Clinical: Bums of any body tissue; edema; contractures; secondary infections.

Guy RJ, et al. Three-years' experience ill regional burn center with burns of the eye and eyelids. *Ophthalmic Surg* 1982; 13:383-386.

Huang TT, et al. Burn injuries of the eyelids. *Clin Plast Surg* 1978; 5:571-581.

Reim M, Kuckelkorn R. Chemical and thermal lesions in the orbital region. *Bull Soc Belg Ophthalmol* 1992; 245: 21-28.

Vajpayee RB, et al. Contact thermal burns of the cornea. *Can J Ophthalmol* 1991; 26:215-218.

13q-Syndrome (13q Deletion Syndrome) 1228

General: Chromosome 13q deletion syndrome.

Ocular: Retinoblastoma; telecanthus; hypertelorism; optic nerve hypoplasia; retinal dysplasia.

Clinical: Holoprosencephaly; abnormal lower extremity configuration; atrial septal defect; microcephaly; ambiguous genitalia; hypotonia; low-set ears; growth retardation; mild mental retardation; intestinal atresia.

Nishikawa A, et al. A 13q-syndrome with extensive intestinal atresia. *Acta Paediatr Scand* 1985; 74:305-308.

Santolaya J, et al. Ultrasonographic prenatal diagnosis of the 13q- syndrome. *Fetal Diagn Ther* 1993; 8:261-267.

Stoll C, Alembik Y. A patient with 13q-syndrome with mild mental retardation and with growth retardation. *Ann Genet* 1998; 41:209-212.

Weichselbaum RR, et al. New findings in the chromosome 13 long-arm deletion syndrome and retinoblastoma. *Ophthalmology* 1979; 86:1191-1201.

Thompson Syndrome 1229

General: Autosomal dominant inheritance.

Ocular: Congenital optic atrophy; nystagmus; blindness.

Clinical: None.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Thompson AH, Cashell GTW A pedigree congenital optic atrophy entrancing sixteen affected cases in six generations. *Proc R Soc Med* 1935; 28:1415-1426.

Thomsen Syndrome (Congenital Myotonia Syndrome; Myotonia Congenita) 1230

General: Dominant; inheritance manifestations before age 5 years; prevalent in males; possibly caused by excessive production of acetylcholine neuromuscular junction; emotions and cold enhance symptoms; warmth decreases symptoms; there are two types of this disorder, an autosomal dominant and an autosomal recessive, both with the same Clinical features; has been linked to chromosome 7q35 in the region of the human skeletal muscle chloride channel gene (HUMCLC).

Ocular: Inability to open eyelids for a few seconds after closure; spasm of the orbicularis oculi muscle; extraocular muscle paresis.

Clinical: Myotonia with muscles of upper and lower extremities primarily affected; muscle hypertrophy; pronounced delay in relaxation of contracted voluntary muscles.

Adams RD, Victor M. Congenital myotonia. In: Adams RD, Victor M. *Principles of neurology*, 5th ed. New York: McGraw-Hill, 1993:1275-1278.

Ashworth B. Ocular myotonia. *Bristol Med Chir J* 1975; 90:31-35.

Brooke NM, Cwik VE. Disorders of skeletal muscle: myotonia congenita. In: Bradley WG, ed. *Neurology in Clinical practice*, 2nd ed. Boston: Butterworth-Heinemann, 1995:2026-2027.

George AJ Jr, et al. Molecular basis of Thomsen's disease (autosomal dominant myotonia congenital. *Nat Genet* 1993; 3:305-310.

Miller NR, ed. *Walsh and Hoyt's Clinical Ophthalmology*, 4th ed. Baltimore: Williams & Wilkins, 1987.

Thomsen AJT. Myotonia Congenita: Tonische Krampfe in Willkurlich Beweglichen Muskeln in Folge von Ererbter Physischer Disposition (Ataxia Muscularis?). *Arch Psych* 1876; 6:702.

3B Translocation Syndrome 1231

General: Chromosomal anomaly transmitted by the female but not the male carrier.

Ocular: Iris coloboma; corneal opacity; proptosis; strabismus.

Clinical: Low birth weight; micrognathia; small ears; cleft lip and palate; cardiac defects; ventricular septal defect; atrial septal defect; absent ductus arteriosus; pulmonary arterial diverticulum; right aortic arch; absent pulmonic valve.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Walzer S, et al. A new translocation syndrome (3/B). *N Engl J Med* 1966; 275:290-298.

3p-Syndrome (3p Deletion Syndrome) 1232

General: Chromosome 3p deletion syndrome.

Ocular: Blepharoptosis, telecanthus, mongoloid (down-slanting) palpebral fissures.

Clinical: Mental retardation, profound growth failure, characteristic facies, low birth weight, trigonocephaly, psychomotor delay, micrognathia.

Drumheller T, et al. Precise localisation of 3p25 breakpoints in four patients with the 3p-syndrome. *J Med Genet* 1996; 33:842-847.

Higgins JJ, et al. A gene for nonsyndromic mental retardation maps to chromosome 3p25-pter. *Neurology* 2000; 55: 335-340.

Thrombocytopenia 1233

General: Decrease in platelets.

Ocular: Retinal hemorrhages; papilledema; visual field defects; oculomotor nerve palsy; optic nerve atrophy.

Clinical: Anemia; cranial nerve palsies; thrombotic thrombocytopenic purpura; intracranial hemorrhage; neuroophthalmologic signs and symptoms.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982:235.

Elewaut C, et al. Optic atrophy as a complication of neonatal alloimmune thrombocytopenia. *Bull Soc Belg Ophthalmol* 1991: 241:85-88.

Miyao S. et al. Oculomotor nerve palsy due to intraneural hemorrhage in idiopathic thrombocytopenic purpura: a case report. *Eur Neurol* 1993; 33:20-22.

Thygeson Syndrome (Keratitis Superficialis Punctata) 1234

General: Etiology probably of viral origin; recurrence every 3 to 4 years.

Ocular: Punctate lesions of cornea; keratitis.

Clinical: None.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Thygeson P. Superficial punctate keratitis. *J Am Med Assoc* 1950; 144:1544-1549.

Thyrocerebroretinal Syndrome (Familial Thyrocerebral Retinal Syndrome) 1235

General: Autosomal recessive; renal, neurologic, and thyroid disease.

Ocular: Retinal hemorrhages; central vision defect; retinal edema; optic atrophy.

Clinical: Thrombocytopenia; chronic renal disease; colloid goiter.

Cutler EA, et al. Familial thyrocerebral retinal syndrome: a newly recognized disorder. *Birth Defects 1978:14:* 265-274.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Tic Douloureux (Trigeminal Neuralgia) 1236

General: Brief, sharp, unilateral facial pain that usually occurs in the middle or lower face; occurs more often in females; occurs most frequently in persons over age 40 years; right side affected more than left side.

Ocular: Ipsilateral hyperemia with the pain of conjunctiva; periorbital pain; ipsilateral lacrimation during the pain; decreased corneal sensitivity; photophobia.

Clinical: Pain triggered by chewing, swallowing, laughing, brushing teeth, shaving, or combing hair; may be present with multiple sclerosis.

Ackermann-Korner M, Draeger J. Quantitative Messungen der Hornhautsensibilitat bei idiopathischer Trigeminusneuralgie und nach neurochirurgischen Eingriffen am N. trigeminus. *Klin Monatsbl Augenheilkd* 1991; 199:84-90.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Grimson BS, Boone SC. Sixth nerve palsy complicating percutaneous thermal ablation of the trigeminal nerve rootlet. *Am J Ophthalmol* 1981; 92:225-229.

Gutrecht JA, Lessell 1M. Photophobia in trigeminal neuralgia. *J Neuro-Ophthalmol* 1994; 14:122-123.

Voorhies R, Patterson RH. Management of trigeminal neuralgia (tic douloureux). *JAMA* 1981; 245:2521-2523.

Tilted Disk Syndrome 1237

General: Relatively common congenital anomaly (l% to 2% of the population) consisting of inferonasal "tilting" of the disk, with the upper and temporal portion of the disk laying anterior to the inferonasal portion.

Ocular: "Tilting" of the disk with associated findings of an obliquely directed long axis of the disk, inferonasal crescent, posterior staphyloma of the affected inferonasal region of the fundus, and upper and temporal emergence of the retinal vessels rather than the nasal (situs inversus); lenticular astigmatism.

Clinical: Patients tend to have myopic astigmatism and superotemporal or bitemporal visual field depression; patients are at increased risk for retinal serous detachment.

Apple DJ, Rabb MF, Walsh PM. Congenital anomalies of the optic disc. *Surv Ophthalmol* 1982; 27:3-41.

Brazitikos PD, et al. Threshold perimetry in tilted disc syndrome. *Arch Ophthalmol* 1990; 108:1698-1700.

Cohen SY, et al. Macular serous retinal detachment caused by subretinal leakage in tilted disc syndrome. *Ophthalmology* 1998; 105:1831-1834.

Dorrell D. The tilted disc. *Br J Ophthalmol* 1978; 62:16-20.

Gass JDM. Stereoscopic atlas of macular diseases: diagnosis and treatment, 4th ed. St. Louis: CV Mosby, 1997: 986-987.

Gunduz A, Everekliogiu C, Er H, et al. Lenticular astigmatism in tilted disc syndrome. J Cataract Refract Srug 2002; 28: 1836-1840.

Young SE, Walsh FB. Knox DL. The tilted disk syndrome. *Am J Ophthalmol* 1976;82: 16-23.

Tolosa-Hunt Syndrome (Painful Ophthalmoplegia) 1238

General: Symptoms last from days to weeks; attacks recur at intervals of months or years; inflammatory lesion of cavernous sinus; onset most frequent in fifth decade of life; recurrent Tolosa-Hunt syndrome has been observed in some patients.

Ocular: Steadily "growing" retroorbital pain; ptosis; involvement of cranial nerves III, IV, VI, and first division of V; scintillating scotomata; sluggish pupil reaction to light; corneal sensitivity diminished; optic neuritis.

Clinical: Inflammatory lesions of cavernous sinus.

Fowler TJ, et al. Tolosa-Hunt syndrome: the dangers of an eponym. *Br J Ophthalmol* 1975; 59: 149-154.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Hannerz J. Recurrent Tolosa-Hunt syndrome. *Cephalalgia* 1992; 12:45-51.

Hunt WE, et al. Painful ophthalmoplegia. *Neurology* 1961; 11:56.

Lesser R, Jampol LM. Tolosa-Hunt syndrome and anti-nuclear factor. *Am J Ophthalmol* 1974; 77:732.

Smith JL, Taxdal DSR. Painful ophthalmoplegia: the Tolosa-Hunt syndrome. *Am J Ophthalmol* 1966; 61:1466.

Tolosa E. Periarteritic lesions of carotid siphon with *Clinical* features of carotid infraclinoidal aneurysm. *J Neurol Neurosurg Psychiatry* 1954; 17:300.

Toluene Abuse 1239

General: Caused by inhalation of toluene and other toluene-containing substances, such as glue.

Ocular: Jerking movements of the eyes; bilateral optic neuropathy.

Clinical: Progressive tremor of the limbs, trunk, and head; dizziness; slurred speech; bilateral hearing loss; mild impairment of memory and concentration; reduced magnetic resonance signal intensity in the brain; distal renal tubular acidosis.

Carlisle EJ, et al. Glue-sniffing and distal renal tubular acidosis: sticking to the facts. *J Am Soc Nephrol* 1991:1: 1019-1027.

Keene JR. Toluene optic neuropathy. *Am Neurol* 1978; 4:390.

Lagar RB. Multifocal central nervous system damage caused by toluene abuse. *Neurology* 1983; 33:1337-1340.

Wilkins-Haug L, Gabow PA. Toluene abuse during pregnancy: obstetric complications and perinatal outcomes. *Obstet Gynecol* 1991; 77:504-509.

Topless Optic Disk Syndrome 1240

General: Superior segmental optic hypoplasia.

Ocular: Relative superior entrance of the central retinal artery; thinning of the superior peripapillary nerve fiber layer; superior peripapillary scleral halo; pallor of the superior disk.

Clinical: Patients usually have good visual acuity and an inferior altitudinal or sector-like field defect; all reported patients either are children of mothers with type I diabetes mellitus or are Japanese.

Hashimoto M, et al. Topless optic disk syndrome without maternal diabetes mellitus. *Am J Ophthalmol* 1999; 128: 111-112.

Kim RY, et al. Superior segmental optic hypoplasia: a sign of maternal diabetes. *Arch Ophthalmol* 1989; 107: 1312-1315.

Landau K, Bajka JD, Kirchschlager BM. Topless optic disks m children of mothers with type I diabetes mellitus. *Am J Ophthalmol* 1997; 125:605-611.

Peterson RA, Walton DS. Optic nerve hypoplasia with a good, visual acuity and visual field defects. *Arch Ophthalmol* 1977; 95:254-258.

Torre Syndrome 1241

General: Multiple sebaceous gland tumors of nonglabrous skin and visceral malignancy (primarily colonic).

Ocular: Sebaceous cell carcinoma of lid; carcinoma of caruncle; tumor of the meibomian gland.

Clinical: Visceral carcinoma of gastrointestinal tract, breast, and prostate.

Descalze M, Rosenthal S. Sebaceous adenomas and keratoacanthoma m patient with malignant lymphoma: a new form of Torre's syndrome. *Cutis* 1981; 28:169.

Jakobiec FA. Sebaceous adenoma of the eyelid and visceral malignancy. *Am J Ophthalmol* 1974; 78:952.

Tillawi L et al. Solitary tumors of meibomian gland origin and Torre's syndrome. *Am J Ophthalmol* 1987; 104: 179-182.

Torre D. Multiple sebaceous tumors. *Arch Dermatol* 1968; 98:549.

Torre-Muir Syndrome 1242

General: Multiple sebaceous gland tumors of nonglabrous skin and visceral malignancy (primarily colonic) and keratoacanthomas; like Torre syndrome but associated with keratoacanthomas; benign skin tumors accompany and sometimes precede development of internal visceral malignancy; autosomal dominant inheritance.

Ocular: Sebaceous cell carcinoma of lid; carcinoma of caruncle; keratoacanthomas; tumor of the meibomian gland.

Clinical: Visceral carcinoma of gastrointestinal tract, breast, and prostate.

Hauck RM, Manders EK. Familial syndromes with skin tumor markers. *Ann Plast Surg* 1994; 33:102-111.

Jakobiec FA, et al. Unusual eyelid tumors with sebaceous differentiation in the Muir-Torre syndrome. *Ophthalmology* 1988; 95:1543-1548.

Jakobiec FA. Sebaceous adenoma of the eyelid and visceral, malignancy. *Am J Ophthalmol* 1974; 78:952.

Muir EG, et al. Multiple primary carcinomata of colon, duodenum and larynx associated with keratoacanthomata of the face. *Br J Surg* 1967; 54: 19l.

Narita H, et al. Muir-Torre syndrome. *J Dermatol* 1992; 19:105-108.

Tillawi I, et al. Solitary tumors of meibomian gland origin and Torre's syndrome. *Am J Ophthalmol* 1987; 104: 179-182.

Torre D. Multiple sebaceous tumors. *Arch Dermatol* 1968; 98:549.

Touraine-Solente-Gole Syndrome (Pachydermoperiostosis; Acropachyderma; Audry II Syndrome; Brugsch Syndrome; Friedrich-Erb-Arnold Syndrome; Hehlinger Syndrome) 1243

General: Rare; hereditary; predominant in males; onset in puberty to third decade.

Ocular: Elephantiasis of the lids caused by meibomian gland cysts and connective tissue hypertrophy; ptosis.

Clinical: Thick and furrowed skin of forehead, face, scalp, hands, and feet; hyperhidrosis of hands and feet; increased subcutaneous secretion; enormous hands and feet; watch crystal-like nails; cylindrical arms and legs; effusions of ankles, knees, and other joints; finger clubbing; facial enlargement; periostitis; cutaneous mucinosis.

Kirkpatrick JN, et al. Ptosis caused by pachydermoperiostosis. *Br J Ophthalmol* 1991; 75:442-446.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Martinez M. Hypertrophic osteoarthropathy. In: Scumacher HR, ed. *Primer on the rheumatic diseases,* 10th ed. Arthritis Foundation, 1993:262-263.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Touraine A, et al. Un Syndrome Osteodermopathique: La Pachyderme Plicaturee avec Pachyperiostose des Extremites. *Presse Med* 1935; 42:1820-1824.

Tourette Syndrome (Gilles de la Tourette Syndrome; Brissaud II Syndrome; Caprolalia Generalized Tic; Guinon Myospasia Impulsiva) 1244

General: Etiology unknown; occurs at ages 7 to 8 years; emotional trauma is frequent precipitating factor; disturbed parent-child relationship frequently encountered.

Ocular: Blepharospasm; oculogyric deviations; dystonic neck movements.

Clinical: Chorea; caprolalia; echolalia tic; blinking and facial twitching.

Braunwald E, et al. *Harrison's Principles of Internal Medicine*, 11th ed. New York: McGraw-Hill, 1987.

Jankovic J, Stone L. Dystonic tics in patients with Tourette's syndrome. *Mov Disord* 1991; 6:248-252.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981:317.

Toxic Lens Syndrome (Toxic Anterior Segment Syndrome; TASS) 1245

General: Syndrome occurs within a few days to several weeks of implantation of an intraocular lens; with therapy, vision is restored in the majority of cases; increased incidence of disease caused by use of ethylene oxide sterilization (dry pack intraocular lenses); toxic lens syndrome may be prevented by treating the lens with sodium hydroxide and by using modem lathe-cut or compression-molded lenses with polypropylene loops; risk factors include uveitis in history, pseudoexfoliation syndrome, inadequate mydriasis at the start of surgery, problems with intraocular lens implantation, and pigment effusion during surgery.

Ocular: Pigment precipitation on the surface of an intraocular lens; hypopyon; vitreous opacification; chronic uveitis; secondary glaucoma.

Clinical: None.

Alpar JJ. Toxic lens syndrome. *J Ocul Ther Surg* 1982; 5:306-308.

Drolsum L et al. Risk factors for an inflammatory response after extracapsular cataract extraction and posterior chamber IOL. *Acta Ophthalmol* 1994; 72:21-26.

Hellinger Wc, Hasan SA, Bacalis LP, et al. Outbreak of toxic anterior segment syndrome following cataract surgery associated with impurities in autoclave steam moisture. Infect Control Hosp Epidemiol 2006; 27: 294-298

Mamalis, N, Edelhauser HF, Dawson DG, et al. Toxic anterior segment syndrome. *J Cataract Refract Surg* 2006; 32: 324-333

Werner,L, Sher JH, Taylor JR, et al. Toxic anterior segment syndrome and possible association with ointment in the anterior chamber following cataract surgery. *J Cataract Refract Surg* 2006; 32: 227-235

Toxic Shock Syndrome 1246

General: Multisystem illness caused by toxin-producing *Staphylococcus aureus* infection; occurs in all sexes and ages but most frequently in women; seen in association with tampon use; onset may be sudden.

Ocular: Eyelid necrosis; periorbital abscess; neonatal conjunctivitis; conjunctival hyperemia; preseptal cellulitis.

Clinical: Fever; rash; desquamation; hypotension; syncope; dizziness; shock; cardiovascular collapse; coma; death.

Brown MF, Levine RA. Preseptal cellulitis complicated by toxic shock syndrome. *Arch Ophthalmol* 1987; 105: 1631-1632.

Scott DM, Bloome MA. Lid necrosis secondary to streptococcal periorbital cellulitis. *Ann Ophthalmol* 1981;13: 461-465.

Strausbaugh LJ. Toxic shock syndrome. Are you recognizing its changing presentations? *Postgrad Med* 1993; 94: 107-108, 111-113, 117-118.

Weiss A, et al. Bacterial periorbital and orbital cellulitis m childhood. *Ophthalmology* 1985; 90:195-203.

Wolf JE, Rabinowitz LG. Streptococcal toxic shock-like syndrome. *Arch Dermatol* 1995; 131:73-77.

Trachoma 1247

General: Most common in rural communities of the Middle East, Africa, Asia, and South and Central America; caused by *Chlamydia trachomatis;* associated with poor sanitation and medical care.

Ocular: Chronic keratoconjunctivitis; papillae follicles; keratitis; opacities of cornea; scars of palpebral conjunctiva; ptosis; tearing; entropion.

Clinical: Rhinitis; otitis media; upper respiratory tract infection.

Darougar S, et al. A double-blind comparison of topical therapy of chlamydial ocular infection (TRIC infection) chlortetracycline. *Br J Ophthalmol* 1981; 65:549-552.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Jones BR. The prevention of blindness from trachoma. *Trans Ophthalmol Soc UK* 1975; 95:16-33.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Taylor HR. Trachoma-the future for a disease of the past. *Br J Ophthalmol* 1993; 77:66-67.

Transient Light Sensitivity Syndrome 1248

General: Associated with femtosecond laser keratome; may be related to the pulse energy used in flap creastion

Ocular: Transient postoperative photosensitivity

Kurtz RM, Liu XZ, Elner VM, et al. Photodisruption in the human cornea as a function of laser pulse width. J Catract Refract Surg 1997; 13: 653-658.

Stonecipher KG, Dishler JG, Ignacio TS,et al. Transient light sensitivity after femtosecond laser flap creation: *Clinical* findings and management. *J Cataract Refract Surg* . 2006 32: 91-94.

Traumatic Encephalopathy Syndrome (Posttraumatic General Cerebral Syndrome; Postconcussion Syndrome; Punch-Drunk Syndrome) 1249

General: Small focal hemorrhages within the cerebrum and/or cerebellum causing functional brain damage; minor traumatic brain injury is the most common type of traumatic encephalopathy.

Ocular: Nystagmus or nystagmoid ocular movements.

Clinical: Personality change; rigid face without expression; staggering gait; dysphonia.

Katz RT, DeLuca J. Sequelae of minor traumatic brain injury. *Am Fam Physician* 1992; 46:1491-1498.

Martland HS. Punch-drunk. *J Am Med Assoc* 1928; 91:1103.

Ruesch J. Psychophysiological relations in cases with head injuries. *Psychosomat Med* 1945; 7:158.

Treft Syndrome 1250

General: Autosomal dominant; usually appears by age 11 years.

Ocular: Optic atrophy; visual loss; ptosis; ophthalmoplegia.

Clinical: Hearing loss by age 14 years; myopathic changes; balance difficulty.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Treft RL, et al. Dominant optic atrophy, deafness, ptosis, ophthalmoplegia, dystaxia, and myopathy: a new syndrome. *Ophthalmology* 1984; 91:908-915.

Triangular Syndrome 1251

General: Rare; follows trauma of excessive compression during surgery, such as Faden operation.

Ocular: Choroidal ischemia; retinal tear; sectorial infarction of choroid; occlusion of posterior ciliary vessel with triangular chorioretinal scar.

Clinical: None.

Alio JL, et al. Fundus changes following Faden operation. *Arch Ophthalmol* 1984; 102:211-213.

Brine A, et al. Post-contusional triangular syndrome of posterior pole. *Bull Soc Ophthalmol* 1973; 73:985-988.

Trichinellosis (Trichinosis) 1252

General: Parasite *Trichinella* enters the body by ingestion of infected meat (usually poorly cooked pork).

Ocular: Conjunctivitis; splinter hemorrhages of conjunctiva; paralysis of sixth nerve; exophthalmos; proptosis; uveitis; optic neuritis; papilledema; retinal hemorrhages; dyschromatopsia; scotoma; secondary glaucoma; encysted parasites in the extraocular muscles.

Clinical: Fever; urticaria; respiratory symptoms; muscle pain; myalgias and severe proximal muscle weakness; impaired coordination.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Duane TD. Clinical *Ophthalmology*. Philadelphia: JB Lippincott, 1987.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th Ed. Philadelphia: WB Saunders, 2000.

Harms G, et al. Trichinosis: a prospective controlled study of patients ten years after acute infection. *Clin Infect Dis* 1993; 17:637-643.

Santos Duran-Ortiz J, et al. Trichinosis with severe myopathic involvement mimicking polymyositis. Report of a family outbreak. *J Rheumatol* 1992; 19:310-312.

Trichomegaly with Mental Retardation, Dwarfism, and Pigmentary Degeneration of the Retina (Oliver McFarlane Syndrome) 1253

General: Autosomal recessive.

Ocular: Excessive growth of eyelashes and brow hair; pigmentary degeneration of retina; horizontal nystagmus; bilateral choroidoretinal pigmentary degeneration; ring heterochromia of the iris.

Clinical: Bulging of occipital and frontal bones; low birth weight; dwarfism; cryptorchidism; underdevelopment of penis; frontal alopecia.

Delleman JW, Van Walbeek K. The syndrome of trichomegaly, tapetoretinal degeneration and growth disturbances. *Ophthalmologica* 1975; 171:313-315.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Triploidy Syndrome 1254

General: Extra set of chromosomes due to diandry or digyny; stillbirth or early neonatal death.

Ocular: Iris coloboma; microphthalmia; hypertelorism.

Clinical: Large placenta; prenatal growth deficits; large fontanelles; syndactyly; heart defects; cleft lip; genital, brain, ear, and kidney malformations; meningomyelocele; micrognathia.

Arvidsson CG, et al. A boy with complete triploidy and unusually long survival. *Acta Paediatr Scand* 1986; 75: 507-510.

Crane JP, Beaver HA, Cheung SW. Antenatal ultrasound findings in fetal triploidy syndrome. *J Ultrasound Med* 1985; 4:519-524.

Kaufman MH. New insights into triploidy and tetraploidy, from an analysis of model systems for these conditions. *Hum Reprod* 1991; 6:8-16.

Magalim SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

O'Brien WF, et al. Elevated maternal serum alpha-fetoprotein in triploidy. *Obstet Gynecol* 1988; 71[6 Part 2]: 994-995.

Rubenstein JB, et al. Placental changes in fetal triploidy syndrome. *J Ultrasound Med* 1986; 5:545-550.

Strobel SL, Brandt JT. Abnormal hematologic features in a live-born female with triploidy. *Arch Pathol Lab Med* 1985; 109:775-777.

Walker S. et al. Three further cases of triploidy in man surviving to birth. *J Med Genet* 1973; 10:135-141.

Trisomy 8 Mosaicism Syndrome 1255

General: Chromosomally abnormal cell line with each cell containing an extra chromosome 8; other cell lines normal; both sexes affected; present from birth.

Ocular: Strabismus; hypertelorism; deep-set eyes.

Clinical: Mild-to-moderate mental retardation; low-set or malformed ears; broad, bulbous nose; palatal deformity; congenital cardiovascular disorders; hydronephrosis; cryptorchidism; poor coordination; prominent forehead; enlarged nares; full lips; cupped ears; camptodactyly of fingers and toes; reported as a nonrandom secondary change in myxoid liposarcoma.

Fineman RM, et al. Trisomy 8 mosaicism syndrome. *Pediatrics* 1975; 56:762-767.

Geeraets WJ *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Trisomy 18 Syndrome (E Syndrome; Edwards Syndrome) 1256

General: Chromosome 18 present in triplicate; more common in females (3:1); age of mother over 40 years; onset from fetal life.

Ocular: Unilateral ptosis; epicanthal folds; congenital glaucoma; corneal opacities; lens opacities; optic atrophy.

Clinical: Low-set ears; micrognathia; high-arched palate; prominent occiput; cryptorchidism; failure to thrive; ventricular septal defect; hypertonicity with rigidity in flexion of limbs; mental retardation; umbilical and inguinal hernias.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Menkes JH. Trisomy chromosome 18. In: Menkes JH, ed. *Textbook of Child Neurology,* 5th ed. Baltimore: Williams & Wilkins, 1995:221.

Townes PL, et al. Trisomy 18 (16-18) associated with congenital glaucoma and optic atrophy. *J Pediatr* 1962; 61:755.

Trisomy 9q Syndrome 1257

General: Congenital mental retardation syndrome due to 9p trisomy.

Ocular: Hypertelorism; deep-set eyes; antimongoloid (up-slanting) eyes.

Clinical: Mental retardation; short stature; down-turned comers of the mouth; slightly or moderately bulbous nose; moderately large ears; nail dysplasia and hypoplasias; clinodactyly; abnormal dermatoglyphs.

Centerwall WR, Miller KS, Reeves LM. Familial "partial 9q" trisomy: six cases and four carriers in three generations. *J Med Genet* 1976; 13:57-61.

Wahlstrom J, Gustavasson KH. Trisomy 9p syndrome in siblings. *Clin Genet* 1978; 13:511.

Young RS, et al. The dermatoglyphic and *Clinical* features of the 9p trisomy and partial 9p monosomy syndromes. *Hum Genet* 1985; 62:31-39.

Trisomy 17p Syndrome 1258

General: Trisomy 17p duplication syndrome.

Ocular: Hypertelorism, antimongoloid (up-slanting) palpebral fissures.

Clinical: Growth retardation; microcephaly; long philtrum with a thin upper lip; micrognathia; high-arched palate.

Schrander-Stumpel C, et al. Trisomy 17p due to a t(8;17) (p23;p11.2)pat translocation. Case report and review of the literature. *Clin Genet* 1990; 37:148-152.

Trisomy 6p Syndrome (6p+ Syndrome, 6p Duplication Syndrome) 1259

General: Chromosome 6p trisomy syndrome.

Ocular: Blepharochalasis.

Clinical: Low birth weight; psychomotor retardation; craniofacial abnormalities (prominent forehead, large fontanelle, wide sagittal suture, low-set and/or malformed ears); congenital heart malformation; small kidneys; proteinuria.

Breunin MH, Bijlmsma JB, deFrance HF. Partial trisomy 6p due to familial translocation t(6;20)(p21;q13). A new syndrome? *Hum Genet* 1977; 38:7-13.

Rosi G, et al. Trisomy 6p22 leads to 6pter due to familial t(6;13) (p22; q34 or 33) translocation. *Hum Genet* 1979; 51:67-72.

Trisomy 6q Syndrome (Duplication 6q+6q Syndrome) 1260

General: Chromosome 6q trisomy syndrome.

Ocular: Hypertelorism; mongoloid (down-slanting) palpebral fissures.

Clinical: Cleft soft palate; bow-shaped mouth; micrognathia; short, laterally webbed neck; microcephaly; clubbing of hands and feet; syndactyly; growth retardation; mental retardation; "carp" mouth.

Chen H, et al. Familial partial trisomy 6q syndromes resulting from inherited ins (5;6) (q33; q15q27). *Clin Genet* 1976; 9:631-637.

Franchino CJ, et al. Partial trisomy 6q: case report with necropsy findings. *J Med Genet* 1987; 24:300-303.

Zneimer SM. Zeil B, Bachman R. Partial trisomy of chromosome 6q: an interstitial duplication of the long arm. *Am J Med Genet* 1998; 80:133-135.

Trisomy 10q Syndrome (10q+ Syndrome) 1261

General: Chromosome 10q trisomy (duplication) syndrome.

Ocular: Microphthalmia; deep-set eyes; epicanthus; bilateral, enlarged, gray optic disks; distended retinal vessels; bilateral punctate yellow deposits near the macula and optic disk.

Clinical: Mental retardation; microcephaly; prominent forehead; upturned nose; bow shaped mouth; micrognathia; thick and flat helices of the ears; long slender limbs.

Neely K, et al. Ocular findings in partial trisomy 10q syndrome. *Am J Ophthalmol* l988; 106:82-87.

Trisomy 13 Syndrome (Trisomy Dl Syndrome, Patau Syndrome, Reese Syndrome) 1262

General: Extra chromosome in the D group; fatal in the first few months of life; trisomy 13-15 resembles trisomy D1.

Ocular: Anophthalmia; microphthalmia; iris coloboma; cataracts; retinal dysplasia; optic nerve coloboma; optic atrophy; iris dyplasia; calcified lens; retinal detachment; optic nerve hypoplasia; orbital cysts.

Clinical: Apneic spells; developmental deficiency of the nervous system; seizures (minor motor); deafness; cleft lip and palate; hemangiomata; horizontal palmar creases; hyperconvex fingernails; interventricular septal defects; renal abnormalities; cardiovascular changes; respiratory involvement; gastrointestinal disease; urogenital involvement; cerebral hypoplasia with hydrocephalus; mental retardation.

Apple DJ. Patau syndrome. *Am J Ophthalmol* 1970; 70:383.

Cogan DG, Kuwabara T. Ocular pathology of the 13-15 trisomy syndrome. *Arch Ophthalmol* 1964; 72:246.

Ginsberg J, Bove KE. Ocular pathology of trisomy 13. *Ann Ophthalmol* 1974; 6: 113.

Siliaeva NF. Eye developmental defects in Patau's syndrome (trisomy 13). *Oftalmol Z* 1990; 7:423-426.

Trisomy 20 Syndrome 1263

General: Trisomy 20q (duplication) syndrome.

Ocular: Oblique palpebral fissures, strabismus.

Clinical: Round face; cardiac and vertebral abnormalities; mild psychomotor retardation with poor coordination and speech impediment; anencephaly.

Archidiacono N, et al. Trisomy 20q from maternal t(3;20) translocation. *J Med Genet* 1979; 16:229-232.

Zumel RM, et al. Trisomy 20q from maternal translocation and anencephaly. Case report and genetic review. *Ann Genet* 1989; 32:247-249.

Trisomy 21q- Syndrome (21q Deletion Syndrome) 1264

General: Chromosome 21q deletion syndrome.

Ocular: Blepharochalasis; microphthalmia with persistent hypoplastic primary vitreous.

Clinical: Mental and physical retardation; generalized hypertonia; high nasal bridge; micrognathia; malformed ears with preauricular pits, and overlying fingers.

Flaherty L, et al. A case of monosomy 21 found to be an unbalanced de novo t(5q;21q) by fluorescence in situ hybridization. *J Intellect Dis Res* 1998; 42[Part 3]:254-258.

Philip N, et al. Three new cases of partial monosomy 21 resulting from one right 21 chromosome and two unbalanced reciprocal translocations. *Eur J Pediatr* 1984; 142:61-64.

Yamamoto Y, et al. A case of 21q-syndrome with normal SOD-1 activity. *Hum Genet* 1979; 48:321-327.

Trisomy 2q Syndrome (q33-qter) 1265

General: Associated with monosomy 9p (p24-pter); autosomal recessive or X-linked inheritance.

Ocular: Congenital glaucoma; hypertelorism; epicanthus.

Clinical: Low-set and malformed ears; short saddle nose with anteverted nostrils; long, hypoplastic philtrum; thin upper lip; hypospadias; short fingers; muscular hypotonia; psychomotor retardation; clinodactyly; scoliosis; broad, flat nasal bridge; short neck; short esophagus; tubular stomach.

Degrouchy J, Turleau C. *Clinical atlas of human chromosomes.* 2nd ed. New York: John Wiley and Sons, 1984.

Katsushima H, et al. Primary congenital glaucoma in a patient with trisomy 2q and monosomy 9p. *Arch Ophthalmol* 1987; 105:323.

Porter J, et al. Gastrointestinal malformation in genetic disorders: a case of partial trisomy 2q with short esophagus and tubular stomach. *Clin Pediatr* 1991; 30:559-562.

Romain DR, et al. Partial trisomy for 2q in a patient with dir dup (2) (q33.1q35). *J Med Genet* 1994; 31:652-653.

Trisomy 22 Syndrome 1266

General: Trisomy for chromosome 21/22; trisomy 22 may be very mild form of Down syndrome (trisomy 21).

Ocular: High myopia.

Clinical: Schizophrenia; micrognathia; large nostrils; flat occiput; hyperextension of elbows; macrocephaly; hydrocephalus; holoprosencephaly; facioauriculovertebral (Goldenhar) sequence.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fahmi F, et al. Hydrocephalus in an infant with trisomy 22. *J Med Genet* 1994; 31:141-144.

Hayward MD, Bower BD. Chromosomal trisomy associated with the Sturge-Weber syndrome. *Lancet* 1960; 2:844.

Kobrynski L, et al. Trisomy 22 and facioauriculovertebral (Goldenhar) sequence. *Am J Med Genet* 1993; 46:68-71.

Kucerowa M, Polivkova Z. A case of a girl with a 21 ring chromosome. *Hum Hered* 1974; 24:100.

Trisomy DI Syndrome (Trisomy 13; Patau Syndrome; Reese Syndrome) 1267

General: Extra chromosome in the D group; fatal in first few months of life; trisomy 13-15 resembles trisomy D1.

Ocular: Anophthalmia; microphthalmia; iris coloboma; cataracts; retinal dysplasia; optic nerve coloboma; optic atrophy; iris dysplasia; calcified lens; retinal detachment; optic nerve hypoplasia; orbital cysts.

Clinical: Apneic spells; developmental deficiency of nervous system; seizures (minor motor); deafness; cleft lip and palate; hemangiomata (capillary type); horizontal palmar creases; hyperconvex fingernails; interventricular septal defects; renal abnormalities; cardiovascular changes; respiratory involvement; gastrointestinal disease; urogenital involvement; cerebral hypoplasia with hydrocephalus; mental retardation.

Apple DJ, et al. Patau syndrome. *Am J Ophthalmol* 1970; 70:383.

Cogan DG, Kuwabara T. Ocular pathology of the 13-15 trisomy syndrome. *Arch Ophthalmol* 1964; 72:246.

Ginsberg J, Bove KE. Ocular pathology of trisomy 13. *Ann Ophthalmol* 1974; 6:113.

Kukharenko V: et al. Down's syndrome, Edward's syndrome, Patau's syndrome-synthesis of glycosaminoglycans. *Hum Genet* 1994; 94:80-82.

Tristichiasis 1268

General: Autosomal dominant.

Ocular: Three rows of eyelashes.

Clinical: None.

Loeffler L. Erbbiologie des Menschlichen Hautorgans. In: *Hanbuch der Erbbiologie des Menschen.* Berlin: Springer Verlag, 1940.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Tritanomalous Color Blindness (Color Blindness, Partial Tritanomaly) 1269

General: Sex-linked; rare; lacking blue and yellow sensory mechanisms while retaining those for red and green.

Ocular: Blue-yellow color blindness.

Clinical: None.

Kalmus H. *Diagnosis and genetics of defective color vision*. Oxford: Pergamon, 1959.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Tritanopia (Color Blindness; Blue Color Blindness) 1270

General: Autosomal dominant; more common in males; defective blue color vision is characteristic; two amino acid substitutions in the gene encoding the blue-sensitive opsin have been detected.

Ocular: Lacking blue and yellow sensory mechanisms while retaining those for red and green; optic atrophy.

Clinical: None.

Boger WP, Petersen RA. Pediatric *Ophthalmology*. Protan and deutan color blindness. In: Pavan-Langston D, ed. *Manual of ocular diagnosis and therapy*, 4th ed. Boston: Little, Brown and Company, 1995:285-286.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Merin S. The cone dystrophies and color vision disorders. In: Merin S, ed. *Inherited eye diseases.* New York: Marcel Dekker, 1991:176-196.

Miyake Y, et al. Differential diagnosis of congenital tritanopia and tritanopia and dominantly inherited juvenile optic atrophy. *Arch Ophthalmol* 1985; 103:1496-1501.

Weitz CJ, et al. Human tritanopia associated with two amino acid substitutions in the blue sensitive opsin. *Am J Hum Genet* 1992; 50:498-507.

Tropical Pancreatic Diabetes (TPD) 1271

General: Secondary diabetes as a result of chronic calcific pancreatitis; limited geographically to a few tropical countries; highest prevalence in southern India; male predominance; onset at young age; associated with protein calorie malnutrition; possible cause is cassava ingestion; malnutrition has been postulated as a possible etiology.

Ocular: Background retinopathy; proliferative retinopathy; fibrous retinitis proliferans; microaneurysms; macular edema; hemorrhages; exudates; decreased visual acuity.

Clinical: Chronic pancreatitis; recurrent abdominal pain; steatorrhea.

Jarrett RJ, Keen H. Hyperglycemia and diabetic mellitus. *Lancet* 1976; 2: 1009-1112.

Mohan R, et al. Retinopathy in tropical pancreatic diabetes. *Arch Ophthalmol* 1986; 103:1487-1489.

Rao RH. Is tropical pancreatic diabetes malnutrition related? *Diabetes Care* 1993; 16:941-945.

Thomas SV, et al. Visual evoked potential changes in patients with tropical pancreatic diabetes. *Acta Neurol* 1993; 15:427-432.

Tuberculosis 1272

General: Communicable disease caused by the acid-fast bacillus *Mycobacterium tuberculosis.*

Ocular: Conjunctivitis; subconjunctival nodules (tuberculomas); keratitis; pannus; corneal ulcer; blepharitis; cellulitis; meibomianitis; uveitis; dacryocystitis; chronic orbital cellulitis; retinitis; scleritis; scleral perforation; hypopyon; vitreous hemorrhages; optic neuritis; optic atrophy; tuberculous panophthalmitis; choroidal tubercles; intraorbital extraocular lesions.

Clinical: Pulmonary infection; pyuria; hematuria; epididymitis; dysuria; flank pain; distorted calyces; productive cough.

Collins JK. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

D'Souza P, et al. Orbital tuberculosis. *Int Ophthalmol* 1994; 18:149-152.

DeVoe AG, Locatcher-Khorazo D. The external manifestations of ocular tuberculosis. *Trans Am Ophthalmol Soc* 1964; 62:203-212.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Gupta V, Gupta A, Arora S, et al. Presumed tubercular serpiginouslike choroiditis. *Ophthalmology* 2003; 110 1744-1749.

Patkar S, et al. Intraorbital extraocular tuberculosis: a report of three cases. *Surg Neurol* 1994; 42:320-321.

Tejada P, et al. Choroidal tubercles with tuberculous meningitis. *Int Ophthalmol* 1994; 18:115-118.

Tunbridge-Paley Disease 1273

General: Onset in childhood; familial; optic atrophy and deafness seen in conjunction with juvenile diabetes mellitus.

Ocular: Optic atrophy; ptosis; retinal pigmentation.

Clinical: Hearing loss; perceptive deafness; juvenile diabetes mellitus; neurogenic bladder; Friedreich ataxia; Refsum syndrome; amentia; epilepsy; Laurence-Moon-Biedl-Bardet syndrome.

Ikkos DG, et al. Association of juvenile diabetes mellitus, primary optic atrophy, and perceptive hearing loss in three sibs, with additional idiopathic diabetes insipidus in one case. *Acta Endocrinol* 1970; 65:95-102.

Konigsmark BW, et al. Dominant congenital deafness and progressive optic nerve atrophy. *Arch Ophthalmol* 1974; 91:99-103.

Tunbridge RE, Paley RG. Primary optic atrophy in diabetes mellitus. *Diabetes* 1956; 5:295-296.

Tuomaala-Haapanen Syndrome 1274

General: Unknown etiology; features similar to pseudohypoparathyroidism.

Ocular: Antimongoloid lid fissures; hypoplastic tarsus; distichiasis; nystagmus; strabismus; myopia; cataract; hypoplasia of the fovea.

Clinical: Dwarfism; short fingers and toes; wide nose bridge; small maxilla; oxycephaly; cutaneous depigmentation; alopecia; micrognathia; anodontia.

Albright R, et al. Pseudo-hypoparathyroidism - example of "Seabright-Bantam syndrome," report of 3 cases. *Endocrinology* 1942; 30:922.

Tuomaala P, Haapanen E. Three siblings with similar anomalies in the eyes, bones and skin. *Acta Ophthalmol* 1968; 46:365.

Turner Syndrome (Turner-Albright Syndrome; Gonadal Dysgenesis; Genital Dwarfism Syndrome; Ullrich-Turner Syndrome; Bonnevie-Ullrich Syndrome; Pterygolymphangiectasia Syndrome; Ullrich-Bonnevie Syndrome) 1275

General: Ovarian or gonadal agenesis; 45 chromosomes with an XO sex chromosome constitution; females; rare in males; onset in childhood.

Ocular: Exophthalmos; hypertelorism; ptosis; epicanthal folds; blue sclera; corneal nebulae; cataracts; conjunctival lymphoedema; keratoconus.

Clinical: Webbed neck (pterygium colli); diminished growth; mandibulofacial disproportion; cubitus valgus; masculine chest and trunk; late appearance of pubic and axillary hair; congenital deafness; mental retardation; coarctation of aorta.

Austin MW, et al. Conjunctival lymphoedema in Turner's syndrome. *Eye* 1992; 6[Part 3]:335-336.

Buckley CA, Cheng H. Intraocular melanoma, diabetes, and Turner's syndrome: presentation with proptosis. *Br J Ophthalmol* 1981; 65:460-463.

Khodadoust A, Paton D. Turner's syndrome in a male. *Arch Ophthalmol* 1967; 77:309.

Lessell S. Forbes AP. Eye Signs in Turner's syndrome. *Arch Ophthalmol* 1966;76:211.

Turner HH. Syndrome of infantilism, congenital webbed neck and cubitus valgus. *Endocrinology* 1938; 23:566.

Wilson JD, Griffin JE. Disorders of sexual differentiation. In: Braunwald E, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:2039-2051.

U

UGH Syndrome (Uveitis-Glaucoma-Hyphema Syndrome) 1276

General: Caused by a defective anterior chamber lens; can be caused by toxic substance incorporated into the plastic of lens during manufacture or warped intraocular lens; syndrome may rarely occur after extra-capsular cataract extraction (ECCE) with implantation of a posterior chamber intraocular lens.

Ocular: Uveitis; glaucoma; hyphema (UGH).

Clinical: None.

Beehler CC. UGH syndrome after posterior chamber lens implantation. *J Am Intraocul Implant Soc* 1983; 9: 200-201.

Masket S. Pseudophakic posterior iris chafing syndrome. *J Cataract Refract Surg* 1986; 12:252-256.

Van Liefferinge T, et al. Uveitis-glaucoma-hyphema syndrome: a late complication of posterior chamber lenses. *Bull Soc Belg Ophthalmol* 1994; 252:61-65.

Ulcerative Colitis (Regional Enteritis; Inflammatory Bowel Disease) 1277

General: Chronic inflammatory disease of unknown etiology; both sexes affected; onset at all ages, most frequently between ages 20 and 40 years; usually abrupt onset; psychosomatic pathogenesis possible.

Ocular: Iritis; uveitis; episcleritis; papillomatous changes of palpebral conjunctiva; scleritis; serous retinal detachment; choroidal infiltrates; retrobulbar neuritis; papillitis; retinal pigment epithelium disturbance; choroidal folds.

Clinical: Abdominal pain; cramps; diarrhea; arthritis; weight loss; erythema nodosum; aphthous stomatitis; pallor; tenderness over colon; nutritional deficiency; carcinoma; associations with Sjögren syndrome and Takayasu disease have been reported.

Ernst BB, et al. Posterior segment manifestations of inflammatory bowel disease. *Ophthalmology* 1991; 98: 1272-1280.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders. 2000.

Korelitz BI, Coles RS. Uveitis (iritis) associated with ulcerative and granulomatous colitis. *Gastroenterology* 1967; 52:78-82.

Oyanagi H, et al. Ulcerative colitis associated with Takayasu's disease. *Int Med* 1994; 33:127-129.

Petrelli EA. Ocular manifestations of inflammatory bowel disease. *Ann Ophthalmol* 1982; 14:356-360.

Triantafillidis JK, et al. Ulcerative colitis and Sjogren's syndrome in the same patient: report of two cases and a review of the literature. *Ital J Gastroenterol* 1994; 26:299-302.

Ullrich Syndrome (Ullrich-Feichtiger Syndrome; Dyscraniopylophalangy) 1278

General: Belongs to trisomy 13-15; unknown etiology; sporadic occurrence.

Ocular: Microphthalmia to anophthalmia; hypertelorism; narrow lid fissures; strabismus; glaucoma; aniridia; cloudy cornea; corneal ulcers; chorioretinal coloboma.

Clinical: Hypoplastic mandible; broad nose; polydactyly; spina bifida; bicornuate uterus or septa vagina; congenital heart disease.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Ullrich O. Der Status Bonnevie-Ullrich im Rahmen Anderer "Dyscranio-Dysphalangien." *Ergeb Inn Med Kinderheilkd* 1951; 2:412.

Weber JW, Schwarz H. Der Typus Rostockiensis Ullrich-Feichtiger. Dyskranio-pygo-phalangie. *Helv Paediatr Acta* 1960; 15:163.

Ultraviolet Radiation 1279

General: Eye and skin are the only organs of the body particularly sensitive to the nonionizing wavelengths of radiation normally present in the environment.

Ocular: Photokeratitis; pterygia; band keratopathy; herpes simplex keratitis; recurrent corneal erosions; discoloring of lens; retinal degeneration; cataract formation; questionable alterations to the corneal endothelium.

Clinical: Actinic keratosis; edema; erythema of skin, blisters of skin; depigmentation of skin; skin carcinoma.

Bergmanson JP. Ultraviolet radiation damage to the corneal endothelium? *Ophthalmology* 1993; 100:442-443.

Dolin PJ. Ultraviolet radiation and cataract: a review of the epidemiological evidence. *Br J Ophthalmol* 1994; 78: 478-482.

Hockwin O, Lerman S. Clinical evaluation of direct and photosensitized ultraviolet radiation damage to the lens. *Ann Ophthalmol* 1982; 14:220-223.

Lerman S, ed. *Radiant energy and the eye*. New York: Macmillan, 1980.

Unna II Syndrome (Marie-Unna Syndrome) 1280

General: Both sexes affected; rare; autosomal dominant; onset in children.

Ocular: Eyebrows and eyelashes missing.

Clinical: Loss of hair; scant growth of axillary and pubic hair, teeth, and nails; loss of hair in the eyebrows, eyelashes, and body.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Unna M. Ueber Hypotricosis congenita hereditaria. *Dermatol Wochenschr* 1925; 81:1167-1178.

Unverricht Syndrome (Familial Myoclonia Syndrome; Lafora Disease) 1281

General: Fatal hereditary form of diffuse neuronal disease; autosomal recessive; late childhood; death within 2 to 10 years from onset of symptoms.

Ocular: Amaurosis; laminated Lafora bodies in ganglion cell and inner nuclear layers of the retina, either intracellular or extracellular, in inner plexiform and nerve fiber layers, and in the optic nerve.

Clinical: Major epilepsy; widespread myoclonus; dementia; tetraplegia; pseudobulbar palsy; generalized tonic-clonic seizure; behavioral changes; brisk tendon reflexes; cerebellar signs.

Acharya JN, et al. Lafora's disease in south India: a *Clinical*, electrophysiologic, and pathologic study. *Epilepsia* 1993; 34:476-487.

Kaufman MA, et al. Late-onset Lafora's disease with typical intraneural inclusions. *Neurology* 1993; 43:1246-1248.

Riedl JL, et al. Electrophysiological and neuropharmacological studies in a patient with Unverricht-Lafora's disease. *Neurology* 1967; 17:502.

Unverricht H. *Die myokionie*. Berlin: Franz Denticke, 1891.

Urbach-Wiethe Syndrome (Rossle-Urbach-Wiethe Syndrome; Lipoproteinosis; Hyalinosis Cutis et Mucosae; Lipoid Proteinosis; Proteinosis-Lipoidosis) 1282

General: Rare autosomal recessive disorder in which hyaline material is deposited in the skin, mucous membranes, and brain; both sexes affected; onset in infancy; relatively benign progressive course; association with diabetes mellitus.

Ocular: Margin of eyelids may show beadlike excrescences with loss of cilia; itching of eyes; dry eyes.

Clinical: Skin about face covered with small, yellowish-white, waxy nodules; alopecia; hoarseness of voice at birth or within first few years of life; tongue large, thick; hyper-keratotic lesions on knees, elbows, and fingers; inability to cry; dry mouth.

Cinaz P, et al. Lipoid proteinosis: Urbach-Wiethe disease. *Acta Paediatr* 1993; 82:892-893.

Disdier P, et al. Specific xerostomia during Urbach-Wiethe disease. *Dermatology* 1994; 188:50-51.

Feiler-Ofry V, et al. Lipoid proteinosis (Urbach-Wiethe syndrome). *Br J Ophthalmol* 1979; 63:694-698.

Urbach E, Wiethe C. Lipoidosis Cutis et Mucosae, Veichous. *Arch Pathol Anat* 1929; 273:285.

Usher Syndrome (Hereditary Retinitis Pigmentosa-Deafness Syndrome) 1283

General: Retinitis pigmentosa associated with deaf-mutism; dominantly inherited; anatomic and metabolic condition; onset unknown (see Hallgren Syndrome).

Ocular: Concentric contraction of visual fields; retinitis pigmentosa with dotted, fine pigmentation in midperiphery; bone-corpuscle configured pigment deposits mainly along the vessels toward the periphery; yellow-white dots in outer retina and choroid; poor night vision.

Clinical: Deaf-mutism; however, deafness is not always complete; multiple sclerosis.

Berson EL, Adamian M. Ultrastructural findings in an autopsy eye from a patient with Usher's syndrome type II. *Am J Ophthalmol* 1992;114:748-757.

Holland MG, et al. An evaluation of genetic carriers of Usher's syndrome. *Am J Ophthalmol* 1972; 74:940.

Lynch SG, et al. Usher's syndrome and multiple sclerosis. Review of an individual with Usher's syndrome with a multiple sclerosislike illness. *J Neuro-Ophthalmol* 1994; 14:34-37.

Muftuoglu AU, et al. Polycythemia vera associated with Usher's syndrome. *Am J Ophthalmol* 1975;80:93-95.

Usher CH. On the inheritance of retinitis pigmentosa with notes of cases. *R Lond Ophthalmol Hosp Rep* 1913/1914; 19:130.

Uvea Touch Syndrome 1284

General: Caused by intraocular lens coming in contact with uveal tissue; seen most frequently with intracapsular cataract extraction and anterior chamber intraocular lens implant.

Ocular: Corneal decompensation; endothelial dystrophy; retinal edema; pigment dispersion; painful eye; disorders of motility.

Clinical: None.

Binkhorst CD. The uvea-touch syndrome and how to avoid it: personal thoughts about lens implantation. *Acta Ophthalmol* 1985; 63:609-623.

Smith SG, et al. An experimental model for uveal touch syndrome. *J Cataract Refract Surg* 1988; 14:182.

Uveal Effusion Syndrome 1285

General: Congenital anomaly of sclera and, in some cases, the vortex vein; inability to transport extravascular protein across abnormal sclera; condition typically affects middle-aged men and causes recurrent, spontaneous serous retinal and ciliochoroidal detachments, often resulting in significant visual impairment.

Ocular: Exudative retinal detachment; sclera abnormally thick; vortex vein obstruction; idiopathic central serous choroidopathy; vitreous cells; ciliochoroidal detachment; nanophthalmos.

Clinical: Viral infection; elevated blood pressure; allergic reaction; minor trauma.

Brubaker RF, Pederson JE. Review-ciliochoroidal detachment. *Surv Ophthalmol* 1983; 27:281-289.

Gass JD. Uveal effusion syndrome. *Retina* 1983; 3:159-163.

Morita H, et al. Recurrence of nanophthalmic uveal effusion. *Ophthalmologica* 1993; 207:30-36.

Stelmach MZ, et al. Uveal effusion syndrome. *Aust N Z J Ophthalmol* 1994; 22:139-143.

Uveitis Masquerade Syndrome(s) (VMS) 1286

General: Uveitis masquerade syndrome is a group of disorders that mimic uveitis; cells seen may be of noninflammatory origin or are inflammatory and secondary to another disorder.

Ocular: Uveitis; panuveitis; pars planitis; vitreitis; papillitis; anterior segment cells; hypopyon; vitreal infiltrates.

Clinical: Causes may be malignant, such as lymphoma, leukemia, retinoblastoma, melanoma, and lung cancer metastasis, or nonmalignant, such as ocular toxoplasmosis, diabetic retinopathy, hypertension, and radiation retinopathy.

Nussenblatt RB, Witcup SM, Palestine AG. *Uveitis: fundamentals and Clinical practice,* 2nd ed. St. Louis: Mosby, 1906:385-395.

Ranking GA, Jakobiec FA, Hidayat AA. Intraocular lymphoproliferations simulating uveitis. In: Albert DM, Jacobiec FA, eds. *Principles and Practices of Ophthalmology, vol. 1.* Philadelphia: WB Saunders, 1994:524-548.

Rothova A, et al. Uveitis masquerade syndromes. *Ophthalmology* 2001; 108:386-399.

Uyemura Syndrome (Fundus Albipunctatus with Hemeralopia and Xerosis) 1287

General: Rare; resembles retinitis punctata albescens, fundus albipunctatus, and congenital idiopathic night blindness or Oguchi disease; avitaminosis A; affects both sexes.

Ocular: Night blindness; conjunctival xerosis; Bitot spots; white spots on the fundus.

Clinical: None.

Krill AE, Martin D. Photopic abnormalities in congenital stationary nightblindness. *Invest Ophthalmol* 1971; 10:625.

Uyemura M. Uber eine Merkwurdige Augenhintergrundsveranderung bei zwei Fallen von Idiopathischer Hemeralopie. *Klin Monatsbl Augenheilkd* 1928; 81:471.

Venkataswamy G. Ocular manifestations of vitamin A deficiency. *Br J Ophthalmol* 1967; 51:854.

V

V Esotropia Syndrome 1288

Esotropia greater looking down by 15 prism diopters than looking up; may have underaction of superior oblique or overaction of inferior oblique; antimongoloid (downward) slant of lid fissures; may have accommodative, nonaccommodative, or paralytic esotropia components.

Clinical: Fusion obtained by chin depression.

Duane TD. *Clinical Ophthalmology.* Philadelphia: JB Lippincott, 1987.

Roy FH. *Practical management of eye problems: glaucoma, strabismus, visual fields*. Philadelphia: Lea & Febiger, 1975:134-135.

V Exotropia Syndrome 1289

Exotropia greater looking up by 15 diopters than looking down; underaction of superior oblique or overaction of inferior oblique muscles; antimongoloid (downward) slant of lid fissures.

Clinical: Fusion obtained by chin elevation.

Helveston EM. A-exotropia, alternating sursumduction and superior oblique over-action. *Am J Ophthalmol* 1969; 67:377-380.

Roy FH. *Practical management of eye problems: glaucoma, strabismus, visual fields*. Philadelphia: Lea & Febiger, 1975:146-147.

Vaccinia 1290

General: Laboratory virus used for vaccination against smallpox.

Ocular: Pustules of lids; edema of lids; conjunctivitis; orbital cellulitis; keratitis; pannus; corneal perforation; iridocyclitis; central serous retinopathy; perivasculitis; pseudoretinitis pigmentosa; ocular palsies papillitis; optic atrophy.

Clinical: Vesicles; pustules; erythema; fever; malaise; axillary lymphadenopathy; necrosis of skin; vaccinia gangrenosa; encephalomyelitis; drowsiness; vomiting; coma; death.

Goldstein JA, et al. Smallpox vaccination reactions, prophylaxis, and therapy of complications. *Pediatrics* 1975; 55: 342-347.

Harley RD, ed. *Pediatric Ophthalmology*. 4th ed. Philadelphia: WB Saunders, 1998.

Lee SF, et al. Vaccinia keratouveitis manifesting as a masquerade syndrome. *Am J Ophthalmol* 1994; 117: 480-487.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*. 5th ed. Philadelphia: WB Saunders, 1995: 105.

Pettit TH. The poxviruses: vaccinia and variola. *Int Ophthalmol Clin* 1975; 15:203-210.

Van Bogaert-Hozay Syndrome 1291

General: Manifest after age 3 years; similar to Rubinstein-Taybi syndrome; etiology unknown; affects both sexes.

Ocular: Hypertelorism; hypoplastic cilia and eyebrows; ptosis; esotropia; astigmatism; myopia.

Clinical: Facial dysplasia; broad nasal bridge and zygomatic arch; flat, wide nose; arched palate; skeletal anomalies with short, thick phalangeal joints; finger and toes appear infantile; flat nasal bridge; thickened cheeks; deformed ears; micrognathia.

Hozay J. Sur une Dystrophic Familiale Particuliere. Inhibition Precoce de la Croissance et Osteolyse Non Mutilante Acrales avec Dysmorphie Faciale. *Rev Neural* 1953; 89:245.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Roy FH, et al. Ocular manifestations of the Rubinstein-Taybi syndrome. *Arch Ophthalmol* 1968; 79:272.

van Bogaert L. Essai de Classement et d'lnterpretation de Quelques Acro-osteolyses Mutilantes et Non Mutilantes Actuellement Connues. *Acta Neural Psychiatr Belg* 1953; 53:90.

Van Bogaert-Scherer-Epstein Syndrome (Primary Hyperlipidemia; Familial Hypercholesterolemia Syndrome) 1292

General: Autosomal dominant; high serum lipoprotein; both sexes affected; onset at all ages.

Ocular: Xanthelasma; arcus juveniles of the cornea; lipid keratopathy; cataract; retinopathy with yellowish deposits and cholesterol crystals have been reported but are more rare manifestations.

Clinical: Xanthelasmatosis of skin and tendons; progressive atherosclerosis; coronary insufficiency; cardiac infarcts; dementia; progressive ataxia; cerebral infarction; poly-neuropathy.

Blodi FC, Yarbrough JC. Ocular manifestation of familial hypercholesterolemia. *Am J Ophthalmol* 1963; 55:714.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Hoeg JM. Familial hypercholesterolemia. What the zebra can teach us about the horse. *JAMA* 1994; 271:543-546.

Ihara Y, et al. A family of familial hypercholesterolemia with cerebral infarction and without coronary heart disease. An unusual case with corneal opacity, polyneuropathy and carpal tunnel syndrome in the family: therapy with probucol and tocopherol nicotinate. *J Neural Sci* 1991; 106:10-18.

van Bogaert A, et al. *Une Forme Cerebrale de Cholesterinose Generalisee*. Paris: Masson, 1937.

Van Der Hoeve Syndrome (Osteogenesis Imperfecta; Osteopsathyrosis; Ekman Syndrome; Lobstein Syndrome; Spurway Syndrome; Vrolik Syndrome; Eddowes Syndrome; Brittle Bone Disease) 1293

General: Autosomal dominant.

Ocular: Glaucoma; blue sclera; keratoconus; cataract; optic nerve atrophy; retinopathy; retinal detachment.

Clinical: Brittle bones; deafness; hyperflexibility of ligaments; dental defects; developmental delay.

al Gazali LI, et al. A syndrome of osteogenesis imperfecta, optic atrophy, retinopathy and severe developmental delay in two sibs of consanguineous parents. *Clin Dysmorphol* 1994;3:55-62.

Buyse M, Bull MJ. A syndrome of osteogenesis imperfecta, microcephaly, and cataracts. *Birth Defects* 1975; 14:95-98.

Madigan WP, et al. Retinal detachment in osteogenesis imperfecta. *J Pediatr Ophthalmol Strabismus* 1994; 31:268-269.

Ruedemann AD. Osteogenesis imperfecta congenita and blue sclerotics. *Arch Ophthalmol* 1953; 49:6.

Tobelem G, et al. C2 deficiency, platelet aggregation reduction and Lobstein disease in the same family. *Biomedicine* 1974; 2 1:190-193.

Velocardiofacial Syndrome 1294

General: Autosomal dominant; anomaly of neural crest derivatives; most common syndrome of clefting; possible association with microdeletion at 22q11.

Ocular: Retinal vascular tortuosity; posterior embryotoxon; narrow palpebral fissures; suborbital discoloration; small optic nerves; iris nodules; cataracts; prominent corneal nerves; strabismus; hyperopia; myopia; astigmatism; anisometropic astigmatism.

Clinical: Cleft palate; learning disability; ventricular septal defect with or without the tetralogy of Fallot; right-sided aortic arch; prominent nose; retrognathia; helical thickening; small auricles; auricular protrusion; microcephaly; small stature; inguinal or umbilical hernia; scoliosis; slender hands and digits; small vermis; small posterior fossa; developmental delay; heart malformations; late-onset psychosis.

Goldberg R, et al. Velo-cardio-facial syndrome: a review of 120 patients. *Am J Med Genet* 1993; 45:313-319.

Mansour AM, et al. Ocular findings in velo-cardio-facial syndrome. *J Pediatr Ophthalmol Strabismus* 1987; 24: 263-266.

Mitnick RJ, et al. Brain anomalies in velo-cardio-facial syndrome. *Am J Med Genet* 1994; 54:100-106.

Williams MA, et al. Male-to-male transmission of velo-cardio-facial syndrome: a case report and review of 60 cases. *Am J Med Genet* 1983; 15:669-671.

Vermis Syndrome 1295

General: Medulloblastomas arising primarily in the posterior vermis but invading the fourth ventricle secondarily and compressing the bulb; frequent in children.

Ocular: Nystagmus; papilledema; abnormal saccades.

Clinical: Enlargement of the head; vomiting (early sign); stiffness and pain of the neck and shoulders; equilibratory disturbances; incoordination of the limbs.

Baker GS. Physiologic abnormalities encountered after removal of brain tumours from the floor of the fourth ventricle. *J Neurosurg* 1965; 23:338.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Halmagyi GM. Central eye movement disorders. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:2411.

Ranalli PJ, Sharpe JA. Contrepulsion of saccades and ipsilateral ataxia: a unilateral disorder of the rostral cerebellum. *Ann Neurol* 1986; 20:311.

van Bogaert, van L, Martin P. Les Tumeurs du IV. Vent, et le Syndrome Cerebelleux de la Ligne Mediane. *Rev Neurol* 1928; 2:431.

Vertebral Basilar Artery Syndrome 1296

General: "Whiplash" injury with hyperextension of the neck followed by rapid forward movement of the head or osteoarthritis of the cervical spine, cervical ribs (see Cranio-cervical Syndrome).

Ocular: Nystagmus (postural); internuclear ophthalmoplegia; visual deterioration; visual hallucinations may be associated with a decrease in consciousness; homonymous hemianopsia (bilateral); contralateral hemianopic visual field defect.

Clinical: Severe, throbbing occipital headache associated with neck pain; vertigo from ischemia of the internal auditory artery, from the temporoparietal cortex or from ischemia in the lateral tegmentum of the pons; nausea and vomiting; ataxia; hemiparesis; quadriplegia; dysarthria; dysphagia; deafness; dyslexia; atonia; confusion; coma; tremor.

Caplan LR. Posterior cerebral artery syndrome. In: Vinken PJ, Bruyn GW; Klawans HL, eds. *Handbook of Clinical neurology, vascular diseases, part I, vol.* 53. Amsterdam: Elsevier Science, 1988.

Caplan LR. "Top of the basilar" syndrome. *Neurology* 1980; 30:72.

Hoyt WF. Transient bilateral blurring of vision: considerations of an episodic ischemic symptom of vertebral-basilar insufficiency. *Arch Ophthalmol* 1963; 70:746.

Millikan CH, Siekert RG. Studies in cerebrovascular disease: I. the syndrome of intermittent insufficiency of the basilar arterial system. *Proc Mayo Clin* 1965; 30:61.

Vesell Syndrome (Deafness-Strabismus Symphalangia Syndrome) 1297

General: Etiology unknown; possibly autosomal dominant inheritance.

Ocular: Strabismus.

Clinical: Symphalangism; hearing loss.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981.

Vesell ES. Symphalangism, strabismus and hearing loss in mother and daughter. *N Engl J Med* 1960; 263:839-842.

Vestibular Paralysis, Bilateral 1298

General: Infective process bilaterally affecting the vestibular system in any site between the semicircular canals and the vestibular nuclei in the brain; usually vasculitic-type lesion.

Ocular: Nystagmus; no depth perception with eyes closed, no difficulty with eyes open.

Clinical: Staggering; inability to swim; disorientation in water; loss of hearing; vertigo.

Chusid J, et al. Syndrome of bilateral vestibular paralysis. *J Nerv Ment Dis* 1946; 103:172-180.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Ryu JH. Vestibular neuritis: an overview using a classical case. *Acta Otolaryngol Suppl* 1993; 503:25-30.

Vincent Infection (Gingivitis; Trench Mouth) 1299

General: Onset usually sudden; remission, but may reoccur; young adults.

Ocular: Conjunctivitis; corneal ulcer; dacryocystitis; dacryoadenitis; orbititis; uveitis.

Clinical: Painful, bleeding gingiva characterized by necrosis and ulceration of gingival papillae and margins; lymphadenopathy; foul mouth odor; fever; leukocytosis.

Kardachi BJR, Clarke NG. Etiology of acute necrotizing ulcerative gingivitis: a hypothetical explanation. *J Periodont Res* 1974; 45:830.

Shields C, et al. Hand infections secondary to human bites. *J Trauma* 1975; 15:235.

Visual Disorientation Syndrome (Riddoch Syndrome) 1300

General: Parietal lobe lesion.

Ocular: Visual agnosia; stereoscopic vision and central vision unimpaired; homonymous quadrantanopsia.

Clinical: Contralateral numbness and tingling; loss of static or postural sensation when postcentral convolution affected.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Riddoch G. Dissociation of visual perceptions due to occipital injuries, with especial reference to appreciation of movement. *Brain* 1917; 40:15.

Riddoch G. Visual disorientation in homonymous half-fields. *Brain* 1935; 58:376.

Visual Paraneoplastic Syndrome 1301

General: Loss of visual acuity and loss of visual field due to malignant disease without metastasis to visual system; most often occurs with small cell carcinoma of lung; visual symptoms may be only symptom, before neoplastic disease is diagnosed; can occur associated with cutaneous melanoma.

Ocular: Visual field defects; impaired color vision; narrowing of arterioles; changes in retinal pigment epithelium; orbital myositis; optic neuropathy.

Clinical: Endometrial carcinoma, small cell lung carcinoma.

Apple DJ, et al. *Ocular pathology.* St. Louis: CV Mosby, 1985.

Crofts JW, et al. Visual paraneoplastic syndrome associated with undifferentiated endometrial carcinoma. *Can J Ophthalmol* 1988; 23:293-309.

Harris GJ, et al. Orbital myositis as a paraneoplastic syndrome. *Arch Ophthalmol* 1994; 112:380-386.

Kim RY, et al. Cutaneous melanoma-associated renopathy. *Ophthalmology* 1994; 101: 1837-1843.

Malik S, et al. Optic neuropathy: a rare paraneoplastic syndrome. *J Clin Neuro-Ophthalmol* 1992; 12: 137-141.

Vitamin A Deficiency 1302

General: Worldwide cause of blindness; onset in childhood; dietary vitamin A insufficiency; interference of absorption from the intestinal tract and transport or storage in the liver, as with diarrhea or vomiting; most frequent in young children.

Ocular: Bitot spots; conjunctival xerosis; corneal xerosis; keratomalacia with perforation; photophobia; enlarged tarsal glands; corneal ulcer; nyctalopia; hemeralopia; obstruction of the tear ducts.

Clinical: Diarrhea; malabsorption syndrome; follicular hyperkeratosis; lesions on buttocks, legs, and arms; xerosis of skin; tracheitis; bronchitis; pneumonia; chronic obstruction of pancreatic or biliary ducts; increased infant mortality.

Duke-Elder S, MacFaul PA, eds. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Stanton BF, et al. Risk factors for developing mild nutritional blindness in urban Bangladesh. *Am J Dis Child 1986;* 140:584-588.

World Health Organization. Global situation-vitamin A deficiency. In: *Expanded program on immunization update.* Geneva: WHO, 1988.

Vitiligo 1303

General: Patchy depigmentary disorder of the skin; progressive; etiology unknown; autosomal dominant; associated with Vogt-Koyanagi-Harada disease and uveitis.

Ocular: Posterior uveitis; depigmentation of lids, lashes, iris, and retina; retinal atrophy.

Clinical: Loss of pigment in hair; depigmentation of skin; halo nevi.

Albert DM, et al. Ocular abnormalities occurring with vitiligo. *Ophthalmology* 1979; 86:1145-1158.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Park S, et al. Ocular manifestations of pigmentary disorders. *Dermatol Clin* 1992; 10:609-622.

Vitreocorneal Touch Syndrome 1304

General: After cataract extraction, vitreous bulges through the pupillary space, coming in contact with and finally attaching to the corneal endothelium; onset 2 to 3 weeks after cataract extraction.

Ocular: Area of decreased transparency of the central cornea; Descemet folds; striate keratopathy where the intact vitreous face is touching the corneal endothelium; corneal edema; iris bombs; bullous keratopathy.

Clinical: None.

Gostin SB. Vitreocorneal touch syndrome. *South Med J* 1972; 65:741.

Leahey BD. Bullous keratitis from vitreous contact. *Arch Ophthalmol* 1951; 46:22.

Vitreoretinal Skeletal Syndrome 1305

General: Associated with various systemic skeletal abnormalities; autosomal dominant.

Ocular: Optically empty posterior vitreous; fibrillar anterior vitreous preretinal membrane; retinal breaks.

Clinical: Cleft palate; hypermobility of joints; mild skeletal abnormalities; pelvic bone deformities; hearing loss.

Jones KL. Older paternal age and fresh gene mutation: data on additional disorders. *J Pediatr* 1975; 86:84.

Regenbogen L, Godel V. Hereditary vitreoretinal degeneration, cleft lip and palate, deafness, and skeletal dysplasia. *Am J Ophthalmol* 1980; 89:414-418.

Vitreoretinochoroidopathy (VRCP; Autosomal Dominant Vitreoretinochoroidopathy; ADVIRC) 1306

General: Autosomal dominant.

Ocular: Chorioretinal hypopigmentation or hyperpigmentation; preretinal punctate opacities; retinal arteriolar narrowing and occlusion; choroidal atrophy; diffuse retinal vascular incompetence; cystoid macular edema; presenile cataracts; fibrillar condensation and moderate pleocytosis of vitreous; myopia; optically empty vitreous; lattice degeneration; retinal breaks; retinal detachment; glaucoma; spontaneous vitreous hemorrhage.

Clinical: None.

Blair NP, et al. Autosomal dominant vitreoretinochoroidopathy. *Br J Ophthalmol* 1984; 68:2-9.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Traboulsi EL, Payne JW. Autosomal dominant vitreoretinopathy: report of the third family. *Arch Ophthalmol* 1993; 111:194-196.

Vitreous Tug Syndrome (Vitreous Wick Syndrome) 1307

General: Vitreous strand passes through the pupillary space and becomes attached or incarcerated in a corneal wound either posttraumatic or after intraocular surgery (see Irvine Syndrome); associated with endophthalmitis.

Ocular: Sensation of light flashes due to vitreous pull on the retina; irregular pupil; vitreous strands passing through pupil to attach to corneal wound or scar; loss of foveal reflex on ophthalmoscopic examination; circumscribed retinal edema; occasional posterior retinal detachment.

Clinical: None.

Irvine SR. Newly defined syndrome following cataract surgery; interpreted according to recent concepts of structure of vitreous: seventh Francis P. Proctor lecture. *Am J Ophthalmol* 1953; 36:599.

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Ruiz RS, van Teeters W. A late complication following cataract extraction. *Am J Ophthalmol* 1970; 70:483.

Stainer GA, Binder PS. Vitreous wick syndrome following a corneal relaxing incision. *Ophthalmic Surg* 1981; 12: 567-570.

Vogt-Koyanagi-Harada Disease (Harada Disease; Uveitis-Vitiligo-Alopecia-Poliosis Syndrome) 1308

General: Viral infection; occurs predominantly among Italian and Japanese individuals; young adults; chronic.

Ocular: White lashes; secondary glaucoma; bilateral uveitis; sympathetic ophthalmitis; exudative iridocyclitis; vitreous opacities; bilateral serous retinal detachment and edema with spontaneous reattachment after weeks; depigmentation and patches of scattered pigment later; bilateral acute diffuse exudative choroiditis; papilledema; macular hemorrhage; cataracts; phthisis bulbi; poliosis; scleromalacia; intraocular lymphoma.

Clinical: Poliosis; vitiligo; hearing defect; headache; vomiting; meningeal irritation; reported to occur rarely in children.

Carlson MR, Kerman BM. Hemorrhagic macular detachment in the Vogt-Koyanagi-Harada syndrome. *Am J Ophthalmol* 1977; 84:632.

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Manger CC III, Ober RR. Retinal arteriovenous anastomoses in the Vogt-Koyanagi-Harada syndrome. *Am J Ophthalmol* 1980; 89:186-191.

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Walker J, et al. Intraocular lymphoma developing in a patient with Vogt-Koyanagi-Harada syndrome. *Int Ophthalmol* 1993-1994; 17:331-336.

Von Bekhterev-Strumpell Syndrome (Marie-Strumpell Spondylitis; Ankylosing Spondylitis; Pierre-Marie Syndrome; Bekhterev Disease; Rheumatoid Spondylitis) 1309

General: Variant of rheumatoid arthritis; etiology unknown; autosomal dominant; male preponderance; onset at age 20 to 40 years; although genetic background determines susceptibility to uveitis, the disease pattern suggests the possibility of random environmental triggers unrelated to the course of the underlying rheumatologic disorder.

Ocular: Nongranulomatous anterior uveitis; optic nerve atrophy (occasionally); hypopyon; band keratopathy; spontaneous hyphema.

Clinical: Spondylitis of vertebra and sacroiliac joints; ankylosis; general arthralgia; kyphosis; scoliosis; displaced head and total rigidity of spine.

Amor B. Isolated uveitis-relationship with ankylosing spondylitis and Reiter's syndrome. *Mod Probl Ophthalmol* 1976; 16:225-227.

Edmunds L, et al. New light on uveitis on ankylosing spondylitis. *J Rheumatic* 1991; 18:50-52.

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Zervas J, et al. HLA-B27 frequency in Greek patients with acute anterior uveitis. *Br J Ophthalmol* 1977; 61:699-701.

Von Economo Syndrome (Encephalitis Lethargica; Sleeping Sickness; Iceland Disease) 1310

General: Etiology not understood; may be caused by filterable virus; both sexes affected; onset at all ages; epidemic form.

Ocular: Nystagmus; strabismus; diplopia; muscle imbalance; lid retraction; homonymous hemianopsia; cortical blindness.

Clinical: Fever; headache; cramps; lethargy; insomnia; athetoid or choreiform movements; convulsions; depression; unsteady gait; fatigue; parkinsonism; oculogyric crisis; behavior disorder.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

Pruskauer-Apostol B, et al. The present status of encephalitis lethargica. *Neurol Psychiatr (Bucur)* 1977; 15: 125-128.

von Economo C. Encephalitis lethargica. *Wien Klin Wochenschr* 1917; 30:581.

Von Gierke Disease (Glycogen Storage Disease Type I; Glycogenosis Type I; Glucose-6-Phosphatase Deficiency) 1311

General: Condition simulating congenital glaucoma; affects both sexes during first year of life.

Ocular: Corneal clouding; discrete, nonelevated, yellow flecks in macular area.

Clinical: Convulsions; failure to thrive; epistaxis; bleeding tendency; steatorrhea; lumbar lordosis; adiposity; hepatomegaly; enlarged kidney; renal tubular acidosis type I and kidney stones.

Geeraets WJ *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Kanematsu A, et al. Multiple calcium oxalate stone formation in a patient with glycogen storage disease type I (von Gierke's disease) and renal tubular acidosis type I: a case report. *Hinyokika Kiyo* 1993; 39:645-648.

von Gierke E. Hepato-Nephromegalia Glykogenica (Glycogenspeicherkrankeit der Leber und Nieren). *Beitr Pathol Anal* 1929; 82:497-513.

Von Herrenschwand Syndrome (Sympathetic Heterochromia) 1312

General: Congenital anomaly; heterochromia with Horner syndrome; sympathetic palsy from cervical ribs, tumor of the thyroid gland, enlarged cervical lymph nodes, scars following tuberculosis, or syringomyelia in apex of the pleura.

Ocular: Enophthalmos; ptosis; heterochromia (ipsilateral iris); miosis; iris on the side of the sympathetic denervation usually shows subtle hypochromia.

Clinical: Decrease of sweating ipsilateral side of face as part of the sympathetic paralysis.

Durham DG. Congenital heredity in Homer's syndrome. *Arch Ophthalmol* 1958; 60:939.

Eagle RC. Congenital, developmental and degenerative disorders of the iris and ciliary body. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:367.

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Von Hippel-Lindau Syndrome (Retinocerebral Angiomatosis; Angiomatosis Retinae; Cerebelloretinal Hemangioblastomatosis; Lindau Syndrome; Retinal Capillary Hamartoma) 1313

General: Dominant inheritance; angiomata in the cerebellum and the walls of the fourth ventricle; young adults.

Ocular: Secondary glaucoma; angiomatosis of the iris; vitreous hemorrhages; tortuosity of dilated retinal artery and vein (feeder vessels); retinal exudates and hemorrhages; retinitis proliferans; angiomata of optic nerve and retina; papilledema; retinal detachment; lipid accumulation in macula; keratoconus; bilateral macular holes; choroid plexus papilloma; bilateral optic nerve hemangioblastomas.

Clinical: Cerebellar angiomatosis; epilepsy; psychic disturbances to dementia.

Annesley WJ Jr, et al. Fifteen year review of treated cases of retinal angiomatosis. *Ophthalmol Otolaryngol* 1977; 83: 446-453.

Blamires TL, Maher ER. Choroid plexus papilloma. A new presentation of von Hippel-Lindau (VHL) disease. *Eye* 1992; 6[Part 1]:90-92.

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Ginzburg BM, et al. Diagnosis of von Hippel-Lindau disease in a patient with blindness resulting from bilateral optic nerve hemangioblastomas. *AJR Am J Roentgenol* 1992; 159:403-405.

Goldberg MF, Koenig S. Argon laser treatment of von Hippel-Lindau retinal angiomas. I. Clinical and angiographic findings. *Arch Ophthalmol* 1974; 92:121.

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Kilmartin DJ, Mooney DJ, Acheson RW, et al. von hippel-Lindau disease and familial polyposis coli in the same family. Arch Ophthalmol. 1996; 114: 1294

Loewenstein n. Bilateral macular holes in von Hippel Lindau disease. *Arch Ophthalmol* 1995; 113:143-144.

Lowden BA, Harris GS. Pheochromocytoma and von Hippel Lindau's disease. *Can J Ophthalmol* 1976; 11:282-289.

Raka D. Benz MS, Murray TG, et al. Salvage external beam radiotherapy of retinal capillary hemangiomas secondary to von Hippel-Lindau disease. *Ophthalmology*. 2004; 111: 150-153.

Von Monakow Syndrome (Monakow Anterior Choroidal Artery Syndrome) 1314

General: Rupture or thrombosis of anterior choroidal artery; aneurysm; tumor.

Ocular: Visual field defect; hemianopia.

Clinical: Hemiplegia; hemianesthesia.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Steegmann AT, Roberts DJ. The syndrome of the anterior choroidal artery. *J Am Med Assoc* 1935; 104:1695-1697.

Takahashi S, et al. The anterior choroidal artery syndrome I. Microangiography of the anterior choroidal artery. *Neuroradiology* 1994; 36:337-339.

Von Recklinghausen Syndrome (Neurofibromatosis Type I; Neurinomatosis) 1315

General: Dominant inheritance activated at puberty, during pregnancy, and at menopause; strong evidence supports the existence of NFl as a tumor suppresser gene.

Ocular: Proptosis; displacement of the globe; pulsation of the globe; ptosis; elephantiasis of the lids; pigment spots on lids; hydrophthalmos; nodular swelling of corneal nerves; cataracts; optic atrophy; choroidal melanoma; neurofibroma of the choroid, iris, eyelid, and ciliary body; enlarged optic foramen; underdevelopment of orbital bones; cafe-au-lait spots on fundus; hamartoma of retina; congenital glaucoma; focal iris nodules; choroidal nevi; optic nerve gliomas; orbital neurofibroma; keratoconus.

Clinical: Cafe-au-lait skin pigmentations; fibroma molluscum; lipomas and sebaceous adenomas; schwannomas; growth abnormalities; spontaneous fractures; facial hemihypertrophy.

Bickler-Bluth ME, et al. Neurilemoma as a presenting feature of neurofibromatosis. *Arch Ophthalmol* 1988; 106: 665-667.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Legius E, et al. Somatic deletion of the neurofibromatosis type I gene in a neurofibrosarcoma supports a tumor suppresser gene hypothesis. *Nat Genet* 1993; 3:122-125.

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Seiff SR, et al. Orbital optic glioma in neurofibromatosis. *Arch Ophthalmol* 1987; 105:1689-1692.

Smith B, English FP. Classical eyelid border sign of neurofibromatosis. *Br J Ophthalmol* 1979; 54: 134-135.

Von Reuss Syndrome (Galactosemic Syndrome; Galactokinase Deficiency; Galactosemia) 1316

General: Autosomal recessive; consanguinity; conversion of galactose into glucose is blocked, leading to galactosemia; onset after a few days or weeks of milk ingestion; deficiency of galactose-1-phosphate uridyltransferase.

Ocular: Searching-type nystagmus; bilateral nuclear or cortical cataracts appear clinically as oil droplets; bilateral zonular cataracts with fine punctate opacities in the lens periphery.

Clinical: Vomiting; refusal of food; diarrhea; weight loss; hepatomegaly with ascites; jaundice; galactosuria; aminoaciduria; dehydration; hypoglycemic crisis; failure to thrive; hypotonia; lethargy; severe mental and neurologic manifestations.

Cordes FC. Galactosemia cataract: a review. *Am J Ophthalmol* 1960; 50: 1151.

Fraunfelder FT, Roy FH. *Current Ocular Therapy,* 5th ed. Philadelphia: WB Saunders, 2000.

Lerman S. The lens in congenital galactosemia. *Arch Ophthalmol* 1960; 61:88.

Okajima K, et al. Thimidase kinase in individuals with galactokinase deficiency [Letter]. *Am J Hum Genet* 1987; 41: 503-504.

Von Sallmann-Paton-Witkop Syndrome (Hereditary Benign Intraepithelial Dyskeratosis; Witkop-von Sallmann Syndrome; HBID Syndrome) 1317

General: Autosomal dominant; conjunctival and. oral lesions; found in whites.

Ocular: Foamy gelatinous plaques located in a typical horseshoe fashion at 3 and 9 o'clock positions of the perilimbal area; superficial hyperemia of bulbar conjunctiva; corneal vascularization and consequent visual decrease.

Clinical: Thickening of oral mucosa with whitish plaques and folds of a spongy character (asymptomatic), with slow progression from birth into second decade of life.

McLean IW; et al. Hereditary benign intraepithelial dyskeratosis. *Ophthalmology* 1981; 88:164-168.

von Sallmann L, Paton D. Hereditary benign intraepithelial dyskeratosis: I. Ocular manifestations. *Arch Ophthalmol* 1960; 63:421.

Witkop CJ Jr, et al. Hereditary benign intraepithelial dyskeratosis: II. Oral manifestations and hereditary transmission. *Arch Pathol* 1960; 70:696.

Waardenburg Syndrome (Van Der Hoeve-Halberstam-Waardenburg Syndrome; Waardenburg-Klein Syndrome; Embryonic Fixation Syndrome; Interoculo-Iridodermato-Auditive Dysplasia) 1318

General: Irregular dominant inheritance; developmental fault in neural crest with absence of the organ of Corti, aplasia of the spiral ganglion, and pigmentary changes; no sex preference; onset at birth.

Ocular: Hyperplasia of the medial portions of the eyebrows; hypertelorism; blepharophimosis; strabismus; heterochromia iridis; aniridia; microcornea; cornea plana; microphakia; abnormal fundus pigmentation; hypoplasia of optic nerve; synophrys; poliosis; hypopigmentation and hypoplasia of retina and choroid; epicanthus; lateral displacement of inferior puncta; lenticonus; underdevelopment of orbital bones; lateral displacement of inner canthi; hypopigmented iris.

Clinical: Congenital deafness; unilateral deafness or deaf-mutism; broad and high nasal root with absent nasofrontal angle; albinotic hair strain (unilateral); faint patches of skin pigmentation; pituitary tumor; nasal atresia; white forelock.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Goldberg ME Waardenburg's syndrome with fundus and other anomalies. *Arch Ophthalmol* 1966; 76:797.

Hayasaka S, et al. Waardenburg syndrome in Japanese patients. Case reports and literature review. *Ophthalmologica* 1992; 205:46-51.

Holmstrom H, Santanelli E Waardenburg's syndrome. Report of a case with nasal atresia, *Scand J Plast Reconstr Surg Hand Surg* 1991; 25:181-182.

Kimura H, et al. Waardenburg's syndrome and pituitary tumor. *Acta Ophthalmol* 1994; 72:642-644.

Waardenburg PJ. A new syndrome combining development anomalies of the eyelids, eyebrows and nose root with pigmentary defects of the iris and head and with congenital defects. *Am J Hum Genet* 1951; 3:195,

Wagner Syndrome (Hyaloideoretinal Degeneration; Hereditary Hyaloideoretinal Degeneration and Palatoschisis; Clefting Syndrome; Goldmann-Favre Syndrome; Favre Hyaloideoretinal Degeneration; Retinoschisis with Early Hemeralopia) 1319

General: Irregular dominant inheritance; both sexes affected.

Ocular: Epicanthus; nystagmus; myopia; iris atrophy; vitreous opacities with dense streaks and folds in posterior hyaloid membrane; corneal degeneration, including band-shaped keratopathy; cataracts; hyaloideoretinal degeneration (usually apparent after 15 years); narrowing of retinal vessels; pigmentary changes; type of retinal degeneration varies from case to case; retinal detachment and avascular preretinal membranes; marked choroidal sclerosis; pale optic disk; Bergmeister papilla.

Clinical: Palatoschisis; genua valga; facial anomalies; hypoplastic maxilla; saddle nose; hyperextensible fingers, elbows, and knees; tapering fingers.

Black GCM, et al. A novel hereditary developmental vitreoretinopathy with multiple ocular abnormalities localizing to a 5-cm region of chromosome 5q13-q14. *Ophthalmology* 1999; 1006:2074-2081.

Frandsen E. Hereditary hyaloideoretinal degeneration (Wagner) in a Danish family. *Arch Ophthalmol* 1966; 74:223.

Hirose T, et al. Wagner's hereditary vitreoretinal degeneration and retinal detachment. *Arch Ophthalmol* 1973; 89: 176.

Kaiser-Kupfer M. Ectrodactyly, ectodermal dysplasia and clefting syndrome. *Am J Ophthalmol* 1973; 76:992.

Wagner H. Ein Bisher Unbekanntes Erbleiden des Auges (Degeneratio Hyaloideo Hereditaria), Beobachtet im Kanton Zurich. *Klin Monatsbl Augenheilkd* 1938; 100: 840.

Waldenström Syndrome (Macroglobulinemia) 1320

General: Occurs mainly in males over age 50 years; chromosomal abnormalities were reported, with most of the cells having 47 chromosomes.

Ocular: Sludging of conjunctival vessels; crystalline deposits in conjunctiva and cornea; keratoconjunctivitis sicca; retinal venous thrombosis; hemorrhagic glaucoma; papilledema; retinal microaneurysms; cotton-wool spots; dry eye.

Clinical: Adenopathy; hepatosplenomegaly; weakness; fatigue; pallor; dyspnea; weight loss; lymphoid orbital tumor.

Cordido M, et al. Dry eye in Waldenstrom's macroglobulinemia. Improvement after systemic chemotherapy. *Cornea* 1995; 14:210-211.

Ettl AR, et al. Orbital involvement in Waldenstrom's macroglobulinemia: ultrasound, computerized tomography and magnetic resonance findings. *Ophthalmologica* 1992; 205:40-45.

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Friedman AH, et al. Immunofluorescent studies of the eye in Waldenstrom's macroglobulinemia. *Arch Ophthalmol* 1980; 98:743-746.

Lu LW; et al. Sjogren's syndrome and benign hyperglobulinemic purpura of Waldenstrom. *Ann Ophthalmol* 198l; 13: 1285-1287.

Sen HN, Chan CC, Caruso RC, et al. Waldenstrom's macroglobulinemia-associated retinopathy. *Ophthalmology*. 2004; 111: 535-538.

Walker-Clodius Syndrome (Lobster Claw Deformity with Nasolacrimal Obstruction; EEC; Ectrodactyly, Ectodermal Dysplasia, and Cleft Lip/Palate) 1321

General: Autosomal dominant; both sexes affected; onset from birth; association with chromosome 7 abnormalities.

Ocular: Hypertelorism; nasolacrimal obstruction with constant epiphora; mucopurulent conjunctival discharge; keratitis; nanocanalization of the lacrimal duct.

Clinical: Deformities of hands and feet ("lobster claw"); absence of both index and middle fingers and second metacarpals with rudimentary third metacarpals; syndactylism; cleft palate and lips; deafness; ear malformation; renal anomalies.

Fukushima Y, et al. The breakpoints of the EEC syndrome (ectrodactyly, ectodermal dysplasia, and cleft/lip palate confirmed to 7q11.21 and 9p12 by fluorescence in situ hybridization [Letter]. *Clin Genet* 1993; 44:50.

Walker JC, Clodius L. The syndromes of cleft lip, cleft palate and lobster claw deformities of hands and feet. *Plast Reconstr Surg* 1963; 32:627.

Wiegmann OA, Walker FA. The syndrome of lobster claw deformity and nasolacrimal obstruction. *J Pediatr Ophthalmol* 1970; 7:79.

Walker-Warburg Syndrome (Cerebroocular Dysplasia-Muscular Dystrophy; Warburg Syndrome; COD-MD Syndrome; Fukuyama Congenital Muscular Dystrophy; Hard + or - E Syndrome) 1322

General: Rare; encompassing a triad of brain, eye, and muscle abnormalities; probably autosomal recessive.

Ocular: Microphthalmia; cataract; immature anterior chamber angle; retinal dysplasia; retinal detachment; persistent hyperplastic primary vitreous; optic nerve hypoplasia; iris coloboma; opaque cornea; myopia; orbicularis weakness; irregular gray subretinal mottling; optic atrophy.

Clinical: Cerebral and cerebellar agyria-micropolygyria; cortical disorganization; glialmesodermal proliferation; neuronal heterotopias; hypoplasia of nerve tracts; hydrocephalus; encephalocele; muscular dystrophy; seizures; mental retardation; hypotonia; abnormal facies.

Heggie P, et al. Cerebro-ocular dysplasia-muscular dystrophy syndrome. *Arch Ophthalmol* 1987; 105:520-524.

Levine RA, et al. Warburg syndrome. *Ophthalmology* 1983; 90:1600-1603.

Walker AE. Lissencephaly. *Arch Neurol* 1942; 48:13-29.

Warburg M. Hydrocephaly, congenital retinal nonattachment and congenital falciform fold. *Am J Ophthalmol* 1978; 85:88-94.

Wallenberg Syndrome (Dorsolateral Medullary Syndrome; Lateral Bulbar Syndrome) 1323

General: Occlusion of the posterior inferior cerebellar artery; onset after age 40 years; similar to Babinski-Nageotte syndrome but crossed hemiparesis is absent; nystagmus is produced by involvement of the vestibular nuclei or posterior longitudinal bundle.

Ocular: Enophthalmos; ptosis; spontaneous homolateral or contralateral horizontal or torsional nystagmus; miosis; Horner syndrome; skew deviation; impaired contralateral pursuit; saccadic abnormalities; gaze-holding abnormalities.

Clinical: Nausea; vertigo; difficulty in swallowing and speaking; ipsilateral ataxia; muscular hypotonicity; ipsilateral loss of pain and temperature sense of the face; neurotrophic skin ulcers; contralateral hypalgesia; facial weakness.

Brazis PW. Ocular motor abnormalities in Wallenberg's lateral medullary syndrome. *Mayo Clin Proc* 1992; 67:365-368.

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Sacco RL, et al. Wallenberg's lateral medullary syndrome. Clinical-magnetic resonance imaging correlations. *Arch Neurol* 1993; 50:609-614.

Silfverskiold BP. Skew deviation in Wallenberg's syndrome. *Acta Neurol Scand* 1966; 41:381.

Wallenberg A. Anatomische Befunde in Einem als "Akute Bulbaraffektion (Embolie der Arteria Cerebellar. Post. Inf. Sinist.)" Beschriebenen Falle. *Arch F Psychiatr* 1901; 34.

Ward Syndrome (Epitheliomatous Phakomatosis; Nevus-Jaw Cyst Syndrome) 1324

General: Autosomal dominant.

Ocular: Hypertelorism; dystopia canthorum; nevi of eyelids; congenital cataracts; congenital corneal opacities; colobomata.

Clinical: Basal cell nevi with multiple basalomatous nodules on face, neck, and trunk; epithelioma adenoides cysticum.

Font RL, Ferry AP. The phakomatoses. *Int Ophthalmol Clin* 1972; 12:1.

Gorlin RJ, Goltz RW. Multiple nevoid basal-cell epithelioma, jaw cysts and bifid ribs. *N Engl J Med* 1960; 262:908.

Water Intoxication Syndrome (Overhydration Syndrome) 1325

General: Administration of water in excess of kidney excretion capacity; psychogenic water drinker; may be chronic, with slow accumulation of water, or acute.

Ocular: Lacrimation.

Clinical: Chronic symptoms include weakness, sleepiness, apathy, anorexia, nausea, vomiting, sialorrhea, diarrhea, perspiration, behavioral changes, seizures and coma, tendon hyporeflexia, pitting edema, hemiplegia, pulmonary edema, and congestive heart failure; acute symptoms include tendon hyporeflexia, decreased attention, confusion, aphasia, muscle twitching, hemiplegia, incoordination, apathy, violent behavior, and marked muscle weakness.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

Weber Syndrome (Weber-Dubler Syndrome; Cerebellar Peduncle Syndrome; Alternating Oculomotor Paralysis; Ventral Medial Midbrain Syndrome) 1326

General: Lesion of the peduncle (crus), pons, or medulla, which interrupts the third nerve before it emerges from the peduncle and interrupts fibers in the pyramidal tract above the level of the third nuclei; hemorrhage and thrombosis; tumor of the pituitary region, extending posteriorly; also may result secondary to cerebrovascular disease.

Ocular: Ptosis; homolateral third nerve palsy (usually complete); fixed, dilated pupil.

Clinical: Contralateral hemiplegia; contralateral paralysis of face and tongue (supranuclear type).

Kistler JP, Ropper AH, Martin JB. Cerebrovascular diseases. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine,* 13th ed. New York: McGraw-Hill, 1994:2242.

Newman NJ. Third, fourth and sixth-nerve lesions and the cavernous sinus. In: Albert DM, Jakobiec FA, eds. *Principles and Practice of Ophthalmology, vol. V.* Philadelphia: WB Saunders, 1994:2451.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 4, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1991:2384.

Weber HD. A contribution to the pathology of the crura cerebri. *Med Chir Trans* 1863; 46:121.

Wolf S, et al. The posterior inferior cerebellar artery on vertebral angiography. *AJR Am J Roentgenol* 1962; 87:322.

Weber-Christian Syndrome (Pfeiffer-Weber-Christian Syndrome) 1327

General: Etiology unknown; subcutaneous inflammatory lesions; occurs at any age; no gender dominance.

Ocular: Secondary glaucoma; anterior uveitis; acute exudative central choroiditis.

Clinical: Generalized distribution of subcutaneous nodular lesions that vary in size and are located predominantly on the trunk, arms, and legs; recurrent attacks of fever; anorexia; hepatosplenomegaly; malaise; oropharyngeal infections; myocardosis; liver cirrhosis; retroperitoneal fibrosis; ulcerative colitis; myalgia; cardiac dilatation with congestive heart failure.

Christian HA. Relapsing febrile nodular non-suppurative panniculitis. *Arch Intern Med* 1928; 42:338.

Lemley DE, et al. Cardiac manifestation of Weber-Christian disease: report and review of the literature. *J Rheumatal* 1991; 18:756-760.

Nozue M, et al. Ulcerative colitis associated with Weber-Christian panniculitis and musculitis: a case report. *J Gastroenterol* 1994; 29:84-87.

Ohara S, et al. Myalgia as the major symptom in systemic panniculitis (Weber-Christian disease). *Eur Neurol* 1992; 32:321-323.

Popoff N, Weelock M. Relapsing febrile nodular nonsuppurative panniculitis. Pfeiffer- Weber-Christian disease: report of three cases. *Arch Intern Med* 1956; 97:39.

Weber FP. A case of relapsing non-suppurative nodular panniculitis, showing phagocytosis of subcutaneous fat cells by macrophages. *Br J Dermatol Syphil* 1925; 37:301.

WEBINO Syndrome (Wall-Eyed Bilateral Internuclear Ophthalmoplegia) 1328

General: Differential diagnosis includes demyelinating disease, arteriosclerotic cerebrovascular disease, trauma, Arnold-Chiari malformation, syphilis, periarteritis nodosa, glioma, cryptococcal meningitis, and premature infants; usually represents midline involvement of oculomotor nucleus.

Ocular: Wall-eyed internuclear ophthalmoplegia (bilateral internuclear ophthalmoplegia with exotropia).

Clinical: Depends on cause; association with multiple sclerosis in young patients.

Daroff RE, Hoyt WE Supranuclear disorders of ocular control systems in man. In: Bach-Y-Rita P, Collins CC, eds. *The control of eye movements.* Orlando, FL: Academic Press, 1971:223.

Lepore FE, Nissenblatt MJ. Bilateral internuclear ophthalmoplegia after intrathecal chemotherapy and cranial irradiation. *Am J Ophthalmol* 1981; 92:851-853.

Miller NR, ed. *Walsh and Hoyt's Clinical Neuro-Ophthalmology, vol.* 5, *part* 1, 4th ed. Baltimore: Williams & Wilkins, 1995:4323.

Wegener Syndrome (Wegener Granulomatosis) 1329

General: Etiology unknown; occurs in fourth and fifth decades of life; persistent rhinitis or sinusitis; three characteristic features are necrotizing granulomatous lesions in the respiratory tract, generalized focal arthritis, and necrotizing thrombotic glomerulitis,

Ocular: Exophthalmos; lid and conjunctival chemosis; papillitis; conjunctivitis; corneal ulcer; corneal abscess; optic atrophy; optic neuritis; orbital cellulitis; episcleritis; sclerokeratitis; cataract; peripheral ring corneal ulcers; ptosis; dacryocystitis; retinal periphlebitis; cotton-wool spots; retinal and vitreous hemorrhages; rubeosis iridis; neovascular glaucoma.

Clinical: Severe sinusitis; pulmonary inflammation; arteritis; weakness; fever; weight loss; bony destruction; granulomatous vasculitis of the upper and lower respiratory tracts; glomerulonephritis; diffuse pulmonary infiltrates; lymphadenopathy; diffuse pulmonary hemorrhage; overlap with giant cell arteritis.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Flach AJ. Ocular manifestations of Wegener's granulomatosis [Letter]. *JAMA* 1995; 274:1199-1200.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Haynes BF, et al. The ocular manifestations of Wegener's granulomatosis. Fifteen years' experience and review of the literature. *Am J Med* 1977; 63:131-141.

Leavitt RY, Fauci AS. Less common manifestations and presentations of Wegener's granulomatosis. *Curr Opin Rheumatol* 1992; 4:16-22.

Robinson MR, Lee SS, Sneller MC, et al. Tarsal-conjunctival disease associated with Wegener's granulomatosis. *Ophthalmology*. 2003; 110: 1770-1780.

Straatsma BR. Ocular manifestations of Wegener's granulomatosis. *Am J Ophthalmol* 1957; 44:789.

Weil Disease (Leptospirosis) 1330

General: Acute severe infection caused by *Leptospira* transmitted by ingestion of food contaminated by the reservoir bacterium.

Ocular: Acute conjunctivitis; episcleritis; fibrinous iridocyclitis with vitreal haze; hypopyon; keratitis; pain on ocular movement; uveitis; optic neuritis; cataract; hemorrhagic retinitis; ptosis.

Clinical: Jaundice; fever; headaches; chills; vomiting; anemia; psychologic disturbances.

David R, Barkay S. Anterior uveitis and leptospirosis. *Ann Ophthalmol* 1976; 8:958-962.

den Haan PJ, et al. Weil's disease as a cause of jaundice. *Neth J Med* 1993; 42:171-174.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Sanford JP. Leptospirosis. In: Isselbacher KJ, et al., eds. *Harrison's Principles of Internal Medicine*, 13th ed. New York: McGraw-Hill, 1994:743.

Weissenbacher-Zweymuller Syndrome 1331

General: Probably neonatal expression of Stickler syndrome; autosomal dominant.

Ocular: Myopia; congenital glaucoma; cloudy cornea; buphthalmos; Descemet membrane tears; retinal detachment; cataracts.

Clinical: Flattened occiput; flat facies; low nasal bridge; anteverted nostrils; flat vascular nevus-covered glabellar area; small mouth with protruding tip of tongue; high-arched palate; short neck; limbs shorter proximally; dumbbell-shaped long bones; posterior defects in vertebral bodies thoracic region; platyspondylic evidence of coronal cleft lumbar area; deafness; small size at birth; mid face hypoplasia; parietooccipital encephalocele; hearing loss; dwarfism.

Galil A, et al. Weissenbacher-Zweymuller syndrome: long-term follow-up of growth and psychomotor development. *Dev Med Child Neurol* 1991; 33:1104-1109.

Ramer JC, et al. Identical twins with Weissenbacher-Zweymuller syndrome and neural tube defect. *Am J Med Genet* 1993; 45:614-618.

Scribanu N, et al. The Weissenbacher-Zweymuller phenotype in the neonatal period as an expression in the continuum of manifestations of the hereditary arthro-ophthalmopathies. *Ophthalmic Paediatr Genet* 1987; 8:159-163.

Weissenbacher G, Zweymuller E. Gleichzeitiges Vorkommen Eines Syndroms von Pierro Robin und einer Fetalen Chondrodysplasie. *Mschr Kinderheilkd* 1964; 112:315-317.

Winter RM, et al. The Weissenbacher-Zweymuller and Marshall syndromes: further evidence for their identity. *Am J Med Genet* 1983; 16:189-199.

Werlhof Disease (Hemophilia and Thrombocytopenic Purpura) 1332

General: Hemorrhagic disease of unknown etiology.

Ocular: Retinal hemorrhages; degeneration or severe intraocular hemorrhages with resultant retinitis proliferans.

Clinical: Petechiae and ecchymoses of skin and mucous membranes; tendency to bruise; decreased level of circulating platelets with a normal clotting time.

Kobayashi H, Honda Y. Intraocular hemorrhage in a patient with hemophilia. *Metab Ophthalmol* 1985; 8:27-30.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Werlhof PG. *Disquisitio Medica et Philogica de Variolis et Anthracibus*. Brunswick: 1735.

Wermer Syndrome (Multiple Endocrine Neoplasia 1; MEN1; Multiple Endocrine Adenomatosis 1; MEA1; Endocrine Adenoma-Peptic Ulcer Complex; Pluriglandular Adenomatosis n 1333

General: Autosomal dominant; high degree of penetrance; both sexes affected; onset after the first decade (see Zollinger-Ellison Syndrome).

Ocular: Visual field defects secondary to pituitary adenoma.

Clinical: Parathyroid adenomas or hyperplasia; pancreatic adenomas; pituitary adenomas; thyroid adenomas; adrenocortical adenomas; subcutaneous lipomas; hypoglycemic crisis; headaches; amenorrhea; diarrhea; weight loss; acromegaly; Cushing syndrome; hyperthyroidism; ulcer; cerebral aneurysm.

Adachi K, et al. Cerebral aneurysm associated with multiple endocrine neoplasia, type l: case report. *Neurol Med Chir* 1993; 33:309-311.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981:860.

Wermer P. Genetic aspects of adenomatosis of endocrine glands. *Am J Med* 1954; 16:363-371.

Werner Syndrome (progeria of Adults) 1334

General: Etiology unknown; recessive inheritance; consanguinity; second and third decades; possible mechanisms have been proposed to explain mutation of a gene causing inhibition of deoxyribonucleic acid (DNA) synthesis and early cellular senescence.

Ocular: Absence of eyelashes and scanty eyebrows; blue sclera; juvenile cataracts; bullous keratitis; trophic corneal defects; paramacular retinal degeneration; proptosis; telangiectasia of lid; astigmatism; nystagmus; presbyopia; uveitis.

Clinical: Leanness; short stature (160 cm maximum); thin limbs; short, deformed fingers; small mouth; early baldness; stretched, atrophic skin (scleropoikiloderma); telangiectasia and trophic indolent ulcers on toes, heels, and ankles; arteriosclerosis with secondary heart failure.

Bristow JH, Whiting DA. Werner's syndrome: *Clinical* features and cataract surgery. S *Afr Med J* 1973; 47:566-569.

Bullock JD, Howard RO. Werner syndrome. *Arch Ophthalmol* 1973; 90:53-56.

Collins JF. *Handbook of Clinical Ophthalmology*. New York: Masson, 1982.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Thweatt R, Goldstein S. Werner syndrome and biological aging: a molecular genetic hypothesis. *Bioessays* 1993; 15: 421-426.

Wernicke Syndrome I (Superior Hemorrhagic Polioencephalopathic Syndrome; Hemorrhagic Polioencephalitis Superior Syndrome; Encephalitis Hemorrhagica Superioris; Avitaminosis B; Thiamine Deficiency; Beriberi; Gayet-Wernicke Syndrome; Wernicke-Korsakoff Syndrome) 1335

General: Lack of vitamin B or thiamine; focal vascular lesions in the gray matter around third and fourth ventricles and sylvian aqueduct; alcoholics (adults); beriberi of all ages.

Ocular: Ptosis; acute bilateral nuclear ophthalmoplegia; complete ophthalmoplegia; retinal hemorrhages; optic atrophy; optic neuritis; conjunctivitis; blepharitis; nutritional amblyopia; central scotoma; papilledema; nystagmus; absolute pupillary paralysis or Argyll Robertson pupils; accommodative palsy.

Clinical: Early prostration; lethargy; irritability; stupor; delirium; mental disturbances to Korsakoff psychosis; ataxia; tremors; peripheral neuritis; anorexia; vomiting; insomnia; perspiration; tachycardia; hallucinations; retrograde amnesia; apathy; anxiety; fear; defective concentration; cardiomyopathy.

Brust JCM. Environmental neurology: Wernicke Korsakoff syndrome. In: Rowland LP, ed. *Merritt's Textbook of Neurology*, 9th ed. Baltimore: Williams & Wilkins, 1995:971-972.

Cogan G, Victor M. Ocular signs of Wernicke's disease. *Trans Am Ophthalmol Soc* 1953; 51:103.

Djoenaidi W, et al. Beriberi cardiomyopathy. *Eur J Clin Nutr* 1992; 46:227-234.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes.* 2nd ed. Philadelphia: JB Lippincott, 1981:861-862.

Shimojyo S, et al. Cerebral blood flow and metabolism in the Wernicke-Korsakoff syndrome. *J Clin Invest* 1967; 46:849.

Wernicke C. Lehrbuch der Gehirnkrankeiten fur Aerzte und Studirende. Kassel: T. Fischer, 1881.

Yuen TS, Simon RP. Wernicke's encephalopathy. In: Bradley WG, et al., eds. *Neurology in Clinical practice,* 2nd ed. Boston: Butterworth-Heinemann, 1995:1382-1383.

Westphal-Strumpell Disease (Pseudosclerosis of Basal Ganglion) 1336

General: Part of Wilson disease; mostly in men; onset at ages 11 to 25 years (see Wilson Disease).

Ocular: Brown-yellow-green ring in Descemet membrane, about 2 mm wide, which begins directly at the limbus and fades away toward the center; deposition of copper.

Clinical: Jaundice; difficulty in swallowing, speaking, and mastication; extensive muscular rigidity; coarse tremors.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

West Nile Virus Infection 1337

General: Zoonotic disease transmitted by a mosquito vector with wild birds serving as its reservoir; seen worldwide but first seen in North America in 1999

Ocular: Photophobia; chorioretinal lesions; mild vitreous inflammatory reaction; retinal hemorrhage; optic disk swelling

Clinical: Muscle rigidity; meningeal inflammation; encephalitis; poliomyelitis; acute flaccid paralysis

Hershberger VS, Augsburger JJ, Hutchins RK, et al. Chorioretinal lesions in nonfatal cases of west nile virus infection. *Ophthalmology*. 2003; 110: 1732-1736.

Khairallah M, Yahia SB, Ladjimi A, et al. Chorioretinal involvement in patients with west nile virus infection. *Ophthalmology*. 2004; 111: 2065-2070..

West Syndrome (Massive Myoclonia; Jackknife Convulsion) 1338

General: Brain damage from trauma, anoxia, degenerative and metabolic factors and infective agents; onset in first year of life.

Ocular: Nystagmus.

Clinical: Convulsion; nodding of head; opisthotonos; mental retardation.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981.

West WJ. On a peculiar form of infantile convulsions. *Lancet* 1840-1841; 1:724-725.

Weyers Syndrome (2) (Weyers IV Syndrome; lridodental Dysplasia; Dentoirideal Dysplasia; Dysgenesis Iridodentalis; Dysgenesis Mesodermalis Corneae et Irides with Digodontia) 1339

General: Present from birth; hereditary; etiology unknown.

Ocular: Dysplasia; small perforation of iris; pupillary synechiae; microphthalmia; corneal opacity.

Clinical: Dwarfism; myotonic dystrophy; microdontia; oligodontia; hypoplasia of dental enamel.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes,* 2nd ed. Philadelphia: JB Lippincott, 1981:864.

Weyers H. *Dysgenesis Irido-dentalis. Ein Neues Syndrome mit obweichdendem Chromosomalen Geschlecht bei Weiblichen Merkmalträgern. Presented at the Meeting of Deutsche Gessellschaft fuer Kinderheilkunde, Kassel, Germany:* 1960.

Whipple Disease (Intestinal Lipodystrophy) 1340

General: Multisystem disorder; prominent in males; onset between fourth and seventh decades; etiology unknown.

Ocular: Ophthalmoplegia (vertical gaze involved more than horizontal gaze); papilledema; intraocular inflammation; vitreous opacities; bilateral panuveitis; small, round grayish retinal lesions; chemosis; supranuclear ophthalmoplegia; myoclonic ocular jerks; pendular nystagmus.

Clinical: Pneumonia; pleurisy; tonsillitis; sinusitis; cystitis; arthritis; fever; leukocytosis; diarrhea; malabsorption; death; dyspnea; weight loss; lymphadenopathy; polyserositis; gray pigmentation of skin; dementia; facial jerks; rhythmic movement of the mouth, jaw, and extremities.

Durant WJ, et al. Vitrectomy and Whipple's disease. *Arch Ophthalmol* 1984; 102:848-851.

Font RL, et al. Ocular involvement in Whipple's disease: light and electron microscopic observations. *Arch Ophthalmol* 1978; 96:1431-1436.

Gartner J. Whipple's disease of the central nervous system, associated with ophthalmoplegia externa and severe asteroid hyalitis: a clinicopathologic study. *Doc Ophthalmol* 1980; 49:155-187.

Knox DL, et al. Cerebral ocular Whipple's disease: a 62-year odyssey from death to diagnosis. *Neurology* 1995; 45:617-625.

Leland TM, Chambers JK. Ocular findings in Whipple's disease. *South Med J* 1978; 71:335-338.

Schrenk M, et al. Augenbeteiligung be Morbus Whipple. *Klin Monatsbl Augenheilkd* 1994; 204:538-541.

Simpson DA, et al. Oculofacial-skeletal myorhythmia in central nervous system Whipple's disease: additional case and review of the literature. *Mov Disord* 1995; 10:195-200.

Whipple GH. A hitherto undescribed disease characterized anatomically by deposits of fat and fatty acids in the intestinal and mesenteric lymphatic tissues. *Bull Johns Hopkins Hosp* 1907; 18:382-391.

Wildervanck Syndrome (Cervicooculoacousticus Syndrome; Franceschetti-Klein- Wildervanck Syndrome; Wildervanck-Waardenburg Syndrome; Cervicooculofacial Dysmorphia; Cervicooculofacial Syndrome) 1341

General: Etiology unknown; female preponderance; similarities to Klippel-Feil syndrome; onset at birth.

Ocular: Abducens paresis; nystagmus; heterochromia iridis.

Clinical: Deafness or deaf-mutism; torticollis with short, webbed neck; epilepsy; mental retardation; cleft palate; scoliosis; ventricular septal defect; ectopic kidney; hydrocephalus; hypoplastic thumb and growth retardation.

Geeraets WJ. *Ocular Syndromes,* 3rd ed. Philadelphia: Lea & Febiger, 1976.

Jensen J, Rousing H. Dysplasia of the cochlea in a case of Wildervanck's syndrome. *Adv Otorhinolaryngol* 1974; 21:32.

Kose G, et al. Cholelithiasis in cervico-oculo-acoustic (Wildervanck's) syndrome. *Acta Paediatr* 1993; 82:890-891.

Wildervanck LS. A cervico-oculo-acoustic-nerve syndrome. *Ned Tijdschr Geneeskd* 1960; 104:2600.

Williams-Beuren Syndrome (Supravalvular Aortic Stenosis; Beuren Elfin Face; Hypercalcemia Supravalvular Aortic Stenosis; Hypercalcemic Face) 1342

General: Onset at birth or early infancy; occurs in both sexes; etiology unknown; possible abnormality of vitamin D metabolism.

Ocular: Bilateral corneal opacities; hypertelorism; prominent epicanthal folds; strabismus.

Clinical: Anorexia; slow weight gain; retarded physical and mental development; elfin face; absent aortic systolic click; harsh ejection systolic murmur; tooth enamel hypoplasia; malocclusion; cavities.

Greenberg L, Lewis A. The Williams' syndrome, spectrum, and significance of ocular features. *Ophthalmology* 1988; 95:1608-1612. ,

Kapp ME, et al. Strabismus in Williams syndrome. *Am J Ophthalmol* 1995; 119:355-360.

Roy FH, et al. Infantile hypercalcemia and supravalvular aortic stenosis-ocular manifestations of seven cases. *J Pediatr Ophthalmol* 1971; 8:188.

Williams JCP, et al. Supravalvular aortic stenosis. *Circulation* 1961; 24: 1311-1318.

Wilson Disease (Hepatolenticular Degeneration) 1343

General: Lesion in the putamen and lenticular nucleus; familial; occurs in first decade of life; reduced liver function; increase in copper content in tissue.

Ocular: Night blindness; golden, grayish-green, or ruby-red peripheral posterior corneal stromal deposit (Kayser-Fleischer ring); sunflower cataract (same as with retention of copper foreign body); nystagmus; extraocular muscle palsies.

Clinical: Liver cirrhosis; jaundice (early); difficulties in speaking (early); difficulties in swallowing and mastication; extensive muscular rigidity; mouth usually open with salivation ("fixed grin"); coarse tremor.

Johnson BL. Ultrastructure of the Kayser-Fleischer ring. *Am J Ophthalmol* 1973; 776:455.

Tso MOM, et al. Kayser-Fleischer ring and associated cataract in Wilson's disease. *Am J Ophthalmol* 1975; 79:479.

Walshe JM. The eye in Wilson's disease. *Birth Defects* 1976; 12:187.

Wilson SAK. Progressive lenticular degeneration: a familial nervous disease associated with cirrhosis of the liver. *Brain* 1912; 34:295.

Windshield Wiper Syndrome 1344

General: Following cataract extraction with a posterior chamber J loop intraocular lens; intraocular lens too short and moves from side to side with the same motion as windshield wipers.

Ocular: Ruptured zonules; lateral intraocular lens tilt; lateral decentration of intraocular lens; decreased visual acuity; glare.

Clinical: None.

Simcoe CW. Simcoe Posterior chamber lens: theory, techniques and results. *Am Intraocul Implant Soc J* 1981; 7: 154-157.

Smith SG, Lindstrom RL. Malpositioned posterior chamber lenses: etiology, prevention, and management. *Am Intraocul Implant Soc J* 1985; 11:584-591.

Wiskott-Aldrich Syndrome 1345

General: Sex-linked recessive; early infancy with death in the first decade of life; abnormal immune responses; expression of CD43 is defective in this X-chromosome-linked immunodeficiency disorder, suggesting that CD43 might have a role in T-cell activation.

Ocular: Periorbital hemorrhages; vesicular skin eruptions; blepharitis; lid nodules; episcleritis; scleral icterus; conjunctival hemorrhages and purulent discharge; corneal ulcers; retinal hemorrhages; papilledema; peripapillary hemorrhages.

Clinical: Eczema; epistaxis; purpura; hematemesis; bloody diarrhea; otitis media.

Aldrich RA, et al. Pedigree demonstrating sex-linked recessive condition characterized by draining ears, eczematoid dermatitis and bloody diarrhea. *Pediatrics* 1954; 13:133.

Guss RB, McCulley JP. Abnormal immune responses in the ocular presentation of Wiskott-Aldrich syndrome. *Ann Ophthalmol* 1982; 14:1058-1060.

Podos SM, et al. Ophthalmic manifestations of the Wiskott-Aldrich syndrome. *Arch Ophthalmol* 1969; 82:322.

Rosenstein Y, et al. CD43, a molecule defective in Wiskott-Aldrich syndrome, binds ICAM-1. *Nature* 1991; 354:233.

Wiskott A. Familiar Angeborener Morbus Werlhofii? *Monatschr Kinderheilkd* 1937; 68:212.

Wolf-Hirschhorn Syndrome 1346

General: Partial deletion of the short arm of chromosome 4 (4p)

Ocular: exodeviation; nasolacrimal obstruction; foveal hypoplasia; upperlid coloboma; optic disk anomalies; microcornea; hypertelorism; nystagmus; chorioretinal coloboma

Clinical: Developmental delay; microcephaly; seizures; craniofacial anomalies; mental retardation; cardiac defects

Wu-Chen WY, Christiansen SP, Berry Sa, et al. Ophthalmic manifestations of Wolf-Hirchhorn syndrome. *J AAPOS*. 2004; 8: 345-348.

Wolf Syndrome (Monosomy 4 Partial Syndrome; Chromosome 4 Partial Deletion Syndrome; Hirschhorn-Cooper Syndrome) 1347

General: Partial deletion of chromosome 4 of the B group; short life expectancy; present from birth (see Cri-du-Chat Syndrome).

Ocular: Hypertelorism; antimongoloid slanting of palpebral fissures; ptosis; nystagmus;strabismus; iris coloboma; retinal coloboma.

Clinical: Microcephaly; mental retardation; seizures; ear malformations; hypospadias; beaked nose; broad nasal root; cleft lip and palate; hypotonia.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Wolf U, et al. Deletion on short arms of a B-chromosome without cri-du-chat syndrome. *Lancet* 1965; 1:769.

Woody-Ghadimi Syndrome (Ghadimi-Woody Syndrome; Hyperlysinemia, Persistent) 1348

General: Inherited probably autosomal dominant; affects both sexes; age of detection from infancy to early adulthood.

Ocular: Strabismus; ectopia lentis.

Clinical: Severe mental retardation; convulsions; laxity of joints; absence of secondary sex characteristics; hepatosplenomegaly; altered facial features.

Ghadimi H, et al. Hyperlysinemia associated with retardation. *N Engl J Med* 1965; 273:723-729.

Magalini SI, Scrascia E. *Dictionary of Medical Syndromes*, 2nd ed. Philadelphia: JB Lippincott, 1981.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

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Woody NC. Hyperlysinemia. *Proceedings of the American Pediatric Society VII Annual Meeting, Seattle, Washington*, 1964:33(abst).

Wrinkly Skin Syndrome 1349

General: Autosomal recessive; consanguinity; similar to pseudoxanthoma elasticum and Ehlers-Danlos syndrome; onset from birth.

Ocular: Myopia; chorioretinitis; partial optic atrophy; microphthalmia; blepharophimosis; cataract.

Clinical: Dry skin with many wrinkles, including the dorsal surfaces of hands and feet; many creases of palms and soles; loss of skin elasticity; no abnormal wrinkles or creases about the face; prominent venous pattern over the chest wall; mental retardation; dwarfism; kyphosis; delayed growth; small size at birth; retarded growth and development; microcephaly; craniofacial dysmorphism; skeletal anomalies.

Gazit E, et al. The wrinkly skin syndrome: a new heritable disorder of connective tissue. *Clin Genet* 1973; 4:186.

Geeraets WJ. *Ocular Syndromes*, 3rd ed. Philadelphia: Lea & Febiger, 1976.

Goodman RM, et al. Evolution of palmar skin creases in the Ehlers-Danlos syndrome. *Clin Genet* 1972; 3:67.

Kreuz FR, Wittwer BH. Del (2q) - cause of the wrinkly skin syndrome? *Clin Genet* 1993; 43:132-138.

X

X Chromosomal Deletion 1350

General: Deletion of proximal part of long arm of the X chromosome; deletion covers part of region Xq21.1-Xq21.31, the locus for choroideremia; congenital deafness; probable mental retardation.

Ocular: Choroideremia, translucent pigment epithelium; peripheral hyperpigmentation; diffuse choriocapillary layer and retinal pigment epithelium; decreased night vision; optic atrophy; excessive myopia; nystagmus.

Clinical: Congenital deafness; mental retardation; corpus callosum agenesia; cleft lip and palate; anhidrotic ectodermal dysplasia; agammaglobulinemia.

Bleeker-Wagemakers LM, et al. Close linkage between Norrie disease: a cloned DNA sequence from the proximal short arm and the centromere of the X-chromosome. *Hum Genet* 1987; 71:211-214.

Rosenberg T, et al. Choroideremia, congenital deafness and mental retardation in a family with an X chromosomal deletion. *Ophthalmic Paediatr Genet* 1987; 8:139-143.

Xanthism (Rufous Albinism) 1351

General: Autosomal recessive; occurs in blacks.

Ocular: Lack of color in iris.

Clinical: Bright copper-red color of skin and hair.

Barnicot NA. Human pigmentation. *Man* 1957; 57:114-120.

McKusick VA. *Mendelian Inheritance in Man; A Catalog of Human Genes and Genetic Disorders*, 12th ed. Baltimore: The Johns Hopkins University Press, 1998.

Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine, February 12, 2007. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Xeroderma Pigmentosum 1352

General: Rare autosomal recessive disorder characterized by extreme cutaneous photosensitivity; both sexes affected; onset in infancy or early childhood.

Ocular: Conjunctivitis; symblepharon; keratitis; corneal ulcer; blepharitis; uveitis; malignancies of conjunctiva, cornea, eyelids, and iris; ectropion; keratoconus; lid freckles; chronic conjunctival congestion; corneal opacification; bilateral pterygium; epibulbar and palpebral squamous cell corneal carcinoma.

Clinical: Neurologic abnormalities and cutaneous malignancies of ectodermal origin; speech disorders; spastic paralysis; convulsions; mental deficiency; gonadal hypoplasia; stunted growth; carcinoma of tongue.

Calugaru M, Barsu M. Exuberant epibulbar tumor penetrating into the orbit in xeroderma pigmentosum. *Graefes Arch Clin Exp Ophthalmol* 1992; 230:352-357.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Goyal JL, et al. Oculocutaneous manifestations in xeroderma pigmentosa. *Br J Ophthalmol* 1994; 78:295-297.

Newsome DA, et al. Repair of DNA in xeroderma pigmentosum conjunctiva. *Arch Ophthalmol* 1975; 93:660-662.

Sevel D. Xeroderma pigmentosum with ocular complications. *Br J Ophthalmol* 1963; 47:687-689.

Vivian AJ, et al. Ocular melanomas in xeroderma pigmentosum. *Br J Ophthalmol* 1993; 77:597-598.

X-Linked Cone Dysfunction Syndrome 1353

General: X-linked pattern of inheritance; stationary cone dysfunction

Ocular: Myopia; visual loss; color vision abnormality

Michaelides M, Johnson S, Bradshaw K,et al. X-linked cone dysfunction syndrome with myopia and protanopia. *Ophthalmology*. 2005; 112:1448-1454.

X-Linked Mental Retardation Syndrome (XLMR) 1354

General: X-linked mental retardation syndrome, of which many types may exist.

Ocular: Glaucoma may be found in some patients; optic atrophy has been noted.

Clinical: Short stature; small hands and feet; seizures; cleft palate; "coarse" facial appearance; brachydactyly.

Armfield K, et al. X-linked mental retardation syndrome with short stature, small hands and feet, seizures, cleft palate, and glaucoma is linked to Xq28. *Am J Med Genet* 1999;85:236-242.

Carpenter NJ, et al. X-linked mental retardation syndrome with characteristic "coarse" facial appearance, brachydactyly, and short stature maps to proximal Xq. *Am J Med Genet* 1999; 85:230-235.

XXXXX Syndrome (penta X Syndrome) 1355

General: Congenital syndrome due to aneuploidy.

Ocular: Epicanthal folds; hypertelorism; antimongoloid (upward slant) of palpebral fissures.

Clinical: Growth retardation, bilateral.

Martini G, et al. On the parental origin of the X's in a prenatally diagnosed 49, XXXXX syndrome. *Prenat Diagn* 1993; 13:763-766.

XXXXY Syndrome 1356

General: 49 chromosome anomaly; characterized by mental retardation; hypoplastic male genitalia; proximal radioulnar synostosis.

Ocular: Upward slanting of palpebral fissures; strabismus; hypertelorism.

Clinical: Microcephaly; mongoloid facies; high-arched palate; cubitus valgus; in-curving fifth fingers and toes; depressed nasal bridge; nasal speech; prominent lower lip; broad chin; round face configuration; small chest; depressed sternum; wide-spaced nipples; genu valgum; flat feet; no facial or pubic hair; small testes; poor scrotal development; girdle obesity; tremor; excessive dribbling; withdrawal; irritability; proximal radioulnar synostosis; vertebral anomalies; parkinsonism.

Hecht F. Observation on the natural history of 49 XXXXY individuals. *Am J Med Genet* 1982; 8:220.

Singh TH, Rajkowa S. 49 XXXXY chromosome anomaly: an unusual variant of Klinefelter's syndrome. *Br J Psychiatry* 1986; 148:209-210.

XYY Syndrome 1357

General: Y chromosome polysomy syndrome.

Ocular: Colobomata.

Clinical: Mild mental retardation; autism; impulsiveness; aggressive behavior; developmental motor and language delays; excessive height for age; frequent antisocial behavior.

Onwochei Be, et al. Ocular colobomata: major review. *Surv Ophthalmol* 2000; 45:175-194.

Von Gontard A, Hillig U. Ein Kind mit XYY-Syndrom im Erleben seiner Mutter. Ein Bericht nach Tagebuchaufzeichnungen. A child with XYY syndrome as experienced by his mother. A report of daily journal recordings. *Z Kinder Jugendpsychiatr* 1992; 20:46-53.

Y

Y Syndrome 1358

General: Etiology unknown.

Ocular: Partial V-pattern strabismus seen only in superior gaze; a partial A-syndrome with the deviation only occurring in down gaze can be considered an inverted Y-syndrome; occurs primarily with exodeviation.

Clinical: None.

Pau H. *Differential diagnosis of eye diseases*. New York: Thieme, 1987.

Yaws (Frambesia) 1359

General: Chronic infectious disease of childhood; causative agent is *Treponema pertenue;* incubation period 3 to 4 weeks; factors include poor hygiene, humidity, and scanty clothing; infectious cutaneous relapses occur during first 5 years of life.

Ocular: Conjunctivitis; lid granulomas; keratitis; iridocyclitis; lid ulceration; cornea leukomata; cicatricial ectropion.

Clinical: Skin lesion and relapsing secondary lesion of skin and bone; painful papules of soles of feet; crablike gait; fever; lymphadenopathy; nocturnal bone pain; periostitis; gummas of skin and long bones; hyperkeratoses of soles and palms; osteitis; periosteitis; hydrarthrosis.

Kasper DL, et al., eds. *Harrison's Principles of Internal Medicine*, 16th ed. New York: McGraw-Hill, 2005.

Duke-Elder S, MacFaul PA, eds. *System of Ophthalmology, vol. XIII.* St. Louis: CV Mosby, 1974.

Falabella R. Nonvenereal treponematoses: yaws, endemic syphilis, and pinta. *J Am Acad Dermatol* 1994; 31:1075.

Yellow Fever 1360

General: Acute infectious disease of short duration and extremely variable severity.

Ocular: Lid edema; orbital pain; subconjunctival, vitreal, and anterior chamber hemorrhages; mydriasis; optic neuritis; partial optic atrophy.

Clinical: Fever; headache; nausea; epistaxis; relative bradycardia; albuminuria; convulsions; tongue shows red margins with white furred center; copious hemorrhages; anuria; delirium; slow pulse; jaundice; "black vomit"; hematemesis; coma; death.

Andrei G, De Clercq E. Molecular approaches for the treatment of hemorrhagic fever virus infections. *Antiviral Res* 1993; 22:45-75.

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Yersiniosis 1361

General: Caused by gram-negative bacilli *Yersinia enterocolitica* or *Yersinia pseudotuberculosis.*

Ocular: Conjunctivitis; corneal ulcer; endophthalmitis; panophthalmitis; uveitis; retinal hemorrhages; cataract; hypopyon; Parinaud oculoglandular syndrome; iridocyclitis; panuveitis; disseminated chorioretinitis.

Clinical: Gastroenteritis; diarrhea; vomiting; fever; acute terminal ileitis; mesenteric adenitis; arthritis; erythema nodosum.

Bottone EJ. Atypical Yersinia enterocolitica: *Clinical* and epidemiological parameters. *J Clin Microbiol* 1978; 7: 562-567.

Chin GN, Noble RC. Ocular involvement Yersinia enterocolitica infection presenting as Parinaud's oculoglandular syndrome. *Am J Ophthalmol* 1977; 83:19.

Fraunfelder FT, Roy FH. *Current Ocular Therapy*, 5th ed. Philadelphia: WB Saunders, 2000.

Osusky R, Kain HL. Beidseitige Chorioretinitis nach Infektion mit Yersinia enterocolitica. *Fortschr Ophthalmol* 1991; 88:446-449.

Saari KM, et al. Ocular inflammation associated with Yersinia infection. *Am J Ophthalmol* 1980; 89:84-95.

Saebo A, Lassen J. Yersinia enterocolitica: an inducer of chronic inflammation. *Int J Tissue Reactions* 1994; 16:51-57.

Sonnenwirth AC, Weaver RE. Yersinia enterocolitica. *N Engl J Med* 1970; 283: 1468.

Young-Simpson Syndrome 1362

General: Rare congenital syndrome.

Ocular: Blepharophimosis.

Clinical: Congenital hypothyroidism; congenital heart defects; facial dysmorphism (microcephaly, bulbous nose, low-set ears, micrognathia); cryptorchism in males; hypotonia; mental retardation; postnatal growth retardation.

Bonthron DT, et al. Parental consanguinity in the blepharophimosis, heart defect, hypothyroidism, mental retardation syndrome (Young-Simpson syndrome). *J Med Genet* 1993; 30:255-256.

Makamura T, Noma S. A Japanese boy with Young-Simpson syndrome. *Paediatr Jpn* 1997; 39:472-474.

Masuno M, et al. Young-Simpson syndrome: further delineation of a distinct syndrome with congenital hypothyroidism, congenital heart defects, facial dysmorphism, and mental retardation. *J Med Genet* 1999; 84:8-11.

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Zellweger Syndrome (Cerebrohepatorenal Syndrome of Zellweger) 1363

General: Rare; congenital; lethal disease; prevalent in females; demyelination of cerebral white matter, spinal cord, and peripheral nerves; enzymatic defects cause myelin deficiency (see Smith-Lemli-Opitz Syndrome); severe multisystem disorder resulting from defective biogenesis of the peroxisome causes death within the first year.

Ocular: Hypertelorism; microphthalmia; nystagmus; glaucoma; hemimydriasis; corneal opacities; cataract; narrowing of retinal vessels; pigment irregularities and areas of depigmentation; retinal holes without detachment; tapetoretinal degeneration; irregular border-lined optic disks; gray-colored disks; hypoplastic supraorbital ridges; optic atrophy.

Clinical: Hypotony; hepatomegaly; albuminuria; mental retardation; failure to thrive; vomiting; seizures; low birth weight; jaundice; short stature; broad nose; hypospadias; cryptorchidism; septal defect; craniofacial dysmorphic features; high forehead; renal cyst; psychomotor retardation; hepatosplenomegaly; severe hearing impairment.

Fenichel GM. Cerebrohepatorenal syndrome (Zellweger syndrome). In: Fenichel GM, ed. *Clinical pediatric neurology*, 2nd ed. Philadelphia: WB Saunders, 1993:151.

Haddad R, et al. Cerebro-hepato-renal syndrome of Zellweger. *Arch Ophthalmol* 1976; 94:1927-1930.

Martinez M. Abnormal profiles of polyunsaturated fatty acids in the brain, liver, kidney and retina of patients with peroxisomal disorders. *Brain Res* 1992; 583: 171-182.

Stanescu B, Dralando L. Cerebro-hepato-renal (Zellweger's) syndrome. *Arch Ophthalmol* 1972; 87:590.

Zellweger H, et al. A familial syndrome of multiple congenital defects. *Bull Johns Hopkins Hosp* 1964; 114:402-414.

Zieve Syndrome (Hyperlipemia-Hemolytic Anemia-Icterus Syndrome) 1364

General: Unknown etiology; pancreatitis; prevalent in middle-aged males with history of recent alcohol intake; insidious onset.

Ocular: Moderate-to-marked icterus; cloudy cornea; corneal ulcers; retinal lipemia.

Clinical: Anorexia; nausea and vomiting; epigastric pain; diarrhea; weight loss; fatigue; weakness; jaundice; hepatomegaly.

Claude R, et al. Disorders of blood lipids in Zieve's syndrome. *Nouv Presse Med* 1974; 3:1439.

Zieve L. Jaundice, hyperlipemia and hemolytic anemia: a heretofore unrecognized syndrome associated with alcoholic fatty liver and cirrhosis. *Ann Intern Med* 1958; 48:471.

Zinsser-Engman-Cole Syndrome (Dyskeratosis Congenita with Pigmentation; Cole-Rauschkolb-Toomey Syndrome) 1365

General: Variant of Fanconi familial aplastic anemia; recessively inherited with male linkage; consanguinity; onset between ages 5 and 13 years.

Ocular: Ectropion; chronic blepharitis; obstruction of lacrimal puncta; conjunctival keratinization; bullous conjunctivitis; epiphora; nasolacrimal duct obstruction; loss of lashes; cataract; glaucoma; strabismus; abnormal fundi.

Clinical: Congenital dyskeratosis with pigmentation of "marble" configuration or "gun metal" appearance; atrophic areas and telangiectasis; dystrophy of nails; vesicular and bullous lesions of oral cavity followed by ulceration; mucosal atrophy; leukoplakia; aplastic anemia; defect of teeth; physical and mental development may be retarded; tufts of hairs on the limbs; keratinized basal cell; papillomas on the trunk.

Cole HN, et al. Dyskeratosis congenita with pigmentation, dystrophia, unguis and leukokeratosis oris. *Arch Dermatol Syphil* 1930; 21:71.

Drachtman RA, Alter BP. Dyskeratosis congenita. *Dermatol Clin* 1995; 13:33-39.

Engman MF. A unique case of reticular pigmentation of the skin with atrophy. *Arch Dermatol Syphil* 1926; 13:685.

Joshi RK, et al. Dyskeratosis congenita in a female. *Br J Dermatol* 1994;130:520-522.

Zinsser F. Atrophica Cutis Reticularis cum Pigmentatione, Dystrophia Unguinum et Leukoplakia Oris. *Ikonographia Dermat (Kyoto)* 1906;219-223.

Ziprkowski Syndrome 1366

General: X-linked recessive albinism.

Ocular: Ocular albinism.

Clinical: Albinism generalized.

Francois J. Albinism. *Ophthalmologica* 1979; 178: 19-31.

Zollinger-Ellison Syndrome (Polyglandular Adenomatosis Syndrome; Multiple Endocrine Adenomatosis Partial Syndrome) 1367

General: Autosomal dominant; more frequent in males (2:1); etiology is islet cell adenoma of pancreas secreting a gastrin-like material; onset third to fifth decade (see Werner Syndrome).

Ocular: Scotomata according to size and position of pituitary tumors; optic nerve atrophy; papilledema; bilateral extraocular muscle metastases.

Clinical: Enteritis and/or peptic ulcers; malignant or benign tumor of islet cell of pancreas; hypersecretion; vomiting; diarrhea; polyglandular adenomatosis; endocrine involvement.

Ballard HS, et al. Familial multiple endocrine adenoma-peptic ulcer complex. *Medicine (Baltimore)* 1964; 43:481.

Berg CL, Wolfe MM. Zollinger-Ellison syndrome. *Med Clin North Am* 1991; 75:903-921.

Liu GT, et al. Bilateral extraocular muscle metastases in Zollinger-Ellison syndrome. *Arch Ophthalmol* 1994; 112: 451-452.

Orloff SL, Debas HT. Advances in the management of patients with Zollinger-Ellison syndrome. *Surg Clin North Am* 1995; 75:511-524.

Zollinger RM, Ellison EH. Primary peptic ulcerations of jejunum associated with islet cell tumors of pancreas. *Am Surg* 1955; 42:709.

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