Visionary Philanthropy for Rare Inherited Conditions

Mandeep Singh, M.D., Ph.D., works at the cutting edge of research, using the very latest tools and knowledge of genetic medicine to explore, and hopefully someday solve, enduring problems in ophthalmology. Some of the inherited retinal conditions he researches are so rare that no other scientists in the world are studying them. The work is exacting and time-consuming, requiring the latest lab equipment, patience and, of course, funding to make it happen.

Of the resources at Singh's disposal, the most cherished is people — the sort of highly skilled, creative and detail-oriented young collaborators who will likely have their own labs someday, once they begin attracting the grant funding that fuels most of American medical research. These scientists are difficult to attract and retain, Singh says. In that regard, philanthropy is critical to his high-risk, high-reward explorations.

The Christopher and Christine Hekimian family is among Singh's most generous benefactors. Christopher Hekimian suffers from an extremely rare inherited genetic disorder that is slowly reducing his vision. "It's peering through a slowly shrinking doughnut hole — a small area of focus surrounded by a wide band of complete blindness," Hekimian says. "It's so rare that they told me I'd have to fund research myself."

For Singh, identifying the exact gene or genes responsible for Hekimian's condition is the first step to a possible cure. Once he finds those genes, Singh can focus on the biochemical mechanisms of the disease, opening avenues for potential drugs to treat the disease or possibly even stem cell approaches that hold the promise of a cure.

Hekimian's eye disease is just one among several genetic conditions that Singh is exploring simultaneously. To speed the scientist's work, Hekimian

has funded a research position in Singh's lab, the Joseph Albert Hekimian Fellow, a position currently held by **Kanza Aziz, M.D.**, whom Singh describes as "a star." The endowment was made in remembrance of Hekimian's son, Joseph, a promising engineering student with a deep love of science who died in 2016 at age 26. Without the support from the Joseph Albert Hekimian Research Fellowship, Singh says he likely would be unable to pursue research into Christopher Hekimian's condition.

Together, Singh and Aziz are working to improve knowledge of genetic eye diseases to serve more families, like the Hekimians, whose inherited conditions often span generations. There are hundreds of genes that might be responsible for any particular disease and a high likelihood that a combination of genes is at work, exponentially complicating the researchers' quest. In that regard, Aziz's contributions have greatly amplified Singh's capabilities in gene discovery and identification, which could help speed possible treatments down the road.

"The Hekimians' generosity has really catapulted our work to the next level," Singh says. "It gives us the security of getting the right people on board for the right length of time to see projects through to the end. I think Joseph Hekimian would be honored by what we're doing in his memory right now." ●

Above: Mandeep Singh

From left, Joseph Hekimian and Kanza Aziz, the Joseph Albert Hekimian Fellow