

Is your eye problem genetic?

AJIT JAIN

Many eye diseases are genetic, says Dr Madhavan Jagdeesen, Healthy Kid International Fellow at Hospital for Sick Children in Toronto. For example, retinoblastoma, a rare childhood eye cancer seen in children below 5 years of age.

"In ophthalmology," said Dr Jagdeesen, one of the five doctors from India on a one-year HKI fellowship to Canada, "15 to 20 percent of diseases are genetic — run in the families. So, it is very important to study the genetics of various eye diseases to learn the molecular wiring of diseases and develop diagnostic testing and treatment."

The Indian government supports the HKI exchange program for doctors, and has contributed approximately \$120,000.

Dr Jagdeesen, who did his specialization in ophthalmology from Madras Medical College and also has a PhD in ophthalmology with specialization in genetics and molecular biology from Sankara Nethralaya, came here in July last year and will return to India in December.

"When patients come to us and if we know the disease runs in the family, we assess and predict the risk of disease getting transmitted to the subsequent generations. The risk of transmission can be

assessed by looking at the pedigree of the family and confirmed by molecular diagnostic testing."

Dr Jagdeesen's PhD work was on retinoblastoma. He said the disease can be diagnosed if a child's photograph shows "loss of red reflex in the eye, or there's squinting of eyes."

"If the disease is not detected early it may require extensive chemo and rarely radiotherapy. Sometimes in spite of this treatment the child may lose vision or an eyeball. In far advanced stage of the disease, it may be life threatening," he added.

"During genetic counseling," he explained, "we explain to the family about



Dr Madhavan Jagdeesen

the disease occurrence, its progression and its transmission. If the parents opt to have second child, which most parents do, prenatal testing can be done to predict the risk of the newborn to have the same problem."

Retinal dystrophy is another genetic eye disease. It can affect any age group.

"Most of the patients with this disease start losing vision at young age and become legally blind in a short period. A severe form of retinal dystrophy called Lebers Congenital Amaurosis occurs in infants. The child is noticed to have low vision since birth and most of the times they are legally blind when found. This disease occurs due to defect in a number of genes. Till last year there was no treatment available to cure this condition. Recently, researchers have found that gene replacement therapy can cure the retinal dystrophy."

Genetic eye diseases are more prevalent in India due to high population and consanguineous marriages — marriage within families — in some states.

"On an average we see 15 to 20 patients with genetic eye diseases every day in our centre in India," Jagdeesen said. "In India, enormous improvement is required in the management of ocular and systemic genetic disorders. There is a special need for physicians to undergo training in genetics and human diseases. Further awareness among people is required to detect these diseases early. Understanding the prevalence of these diseases in the community, the government and major institutions in India have started to focus on improving the infrastructure and training to manage and continue research in this area."

