

It's in the genes

SNEC's Genetic Service offers hope for patients with inherited eye dystrophies.

Dystrophies in the eye are an inherited condition. However, for some, there are no symptoms at birth – they only surface from mid to late adulthood.

Patients with retinal dystrophies usually have night blindness and loss of peripheral vision. A macular dystrophy is when the central vision is affected – patients have difficulty reading or seeing faces, problems with colour vision, and are averse to bright lights.

In Singapore, the prevalence of dystrophies is one in 4,000 people. Although less common than other eye conditions, it can affect an individual drastically. For instance, a person with retinal dystrophy may lose his ability to navigate independently.

GENETIC TESTING

Although there is no treatment for eye dystrophies at the moment, there is a ray of hope with the launch of SNEC's Genetic Service helmed by Dr Chan Choi Mun, Senior Consultant, Medical Retina Department, SNEC.

The clinic runs once every two weeks. Patients, who are typically referred from other SNEC clinics, undergo a protocolised workup, which includes detailed imaging and electrophysiology. Interested patients can undergo genetic testing to look for the disease-causing gene mutation. "There was a patient who,



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in spite of seeing multiple other ophthalmologists, couldn't identify her gene mutation until she came to us. After that, she went back to Australia and found a PhD student to do a specific research and treatment trial just for her. This is an example of how we can give patients hope for the future," Dr Chan said.

CLOSING THE GAP

Dr Chan not only looks after the eye health of her patients, she also ensures they have access to resources to continue leading fulfilling lives. She refers her patients to the Singapore Association of the Visually Handicapped (SAVH) for white cane training or to Guide Dogs Singapore to assess if they are suitable to get one. In addition, Dr Chan encourages her patients to register as visually impaired for benefits, such as reduced bus fares; and to avoid misunderstandings in certain situations, such as when a patient accidentally bumps into others.



Vision of a person with peripheral vision loss

"In the past, these patients may feel marginalised as they are often told 'nothing can be done'. At our clinic, we make sure they get onto our database. Even if they don't see us for one to two years, we can contact them if there are any new developments. With the Genetic Service, patients are viewed holistically, and those who have fallen through the cracks are listened to and have their needs addressed.

In line with the Ministry of Health's increased emphasis on genetics and precision medicine, the Genetic Service team and its scope of work are set to expand. Genetic counsellors and geneticists are also expected to come onboard to offer patients a broader scope of services.

BENEFITS OF GENETIC TESTING INCLUDE:

- Able to confirm diagnosis and provide a more accurate prognosis
- Helpful for family planning
- With knowledge of his or her affected gene, a patient can potentially enter a research or therapeutic trial