

# Singapore team develops Asia's first genetic test that can prevent corneal blindness

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A team of eye doctors and scientists from Singapore have developed Asia's first genetic test for identifying patients with a type of eye disease that affects the cornea called corneal stromal dystrophy which can lead to blurring and loss of vision.

Called the POLARIS TGFBI (Transforming Growth Factor beta Induced) test, this genetic test is designed to aid in the diagnosis and management of patients with corneal stromal dystrophies. It can also identify family members carrying a TGFBI mutation who may also be at risk of developing corneal stromal dystrophy.

The test was developed as part of the PersonalizedOMIC Lattice for Advanced Research and Improving Stratification (POLARIS) programme, a national initiative with multiple partners funded by A\*STAR, to translate research findings from Singapore back to Singapore patients. Building on prior research performed by Singapore National Eye Centre (SNEC) and Singapore Eye Research Institute (SERI), the Genome Institute of Singapore (GIS) and Singapore General Hospital (SGH) developed the clinical test, which was then validated by the National University Hospital (NUH). Subsequently, clinical samples will be sent to NUH's Molecular Diagnosis Centre (MDC) for analysis.

This genetic test provides corneal stromal dystrophy patients with a personalized diagnosis of their TGFBI mutation, facilitating downstream clinical care in order to be able to prognosticate future disease progression and selection of mutation-specific therapies. Test results can

also assist in genetic counselling of families with affected individuals, and pre-surgical risk assessment of these family members before any [eye](#) surgery.

Patients with corneal stromal dystrophy will be advised by their eye doctors and counselled accordingly on the next steps. Individuals who may have a family history of the disease and wish to know the likelihood of inheriting the condition can take the test. Those who fall in the above categories and considering corneal surgery such as LASIK can discuss taking the test with their eye surgeon. The POLARIS™ TGFBI test is now available at SNEC if the eye doctor determines that it is appropriate for the patient.

"It has been our privilege, as POLARIS, to work with the best and brightest clinicians and scientists across Singapore, to bring research findings back to Singapore patients. We are united by a common passion, which is to improve patient outcomes and healthcare", said Prof Patrick Tan, Program Director, POLARIS.

"The development of such a test is a testament to the value of collaborative medical research conducted here in Singapore, where high quality and complex scientific bench research in genetics directly translates to the bedside, and our clinics, to actually help eye physicians diagnose, restore and maintain good vision in our patients, to prevent blindness, and to provide better a better quality of life in a cost-effective manner", said Prof Donald Tan, Medical Director, SNEC.

Speaking on the benefits of the POLARIS programme, Dr Benjamin Seet, Executive Director of the Biomedical Research Council, A\*STAR said: "This is an excellent example whereby we bring science out of the research lab and right to the point of patient care. The first POLARIS test screens for a genetic eye disease. Beyond this, our scientists are developing screening panels for common cancers seen in Singapore, that

will allow doctors to personalise treatments according to a patient's genetic information. This is the future of how medicine will be practised."

"Singapore General Hospital's Pathology department closely collaborates with research scientists and clinicians to translate research discoveries into clinical assays for better patient care. The development of this test, which helps clinicians to identify and address the problem earlier, is a very good example of how research and convergence of experts from various fields can improve patient care and clinical outcomes," said Prof Tan Puay Hoon, Head and Senior Consultant, Department of Pathology, SGH.

"We are privileged to be invited by the POLARIS project team to be involved in the lab setup and test validation for this national programme, and are proud to be working with such an enthusiastic group of people," said Assoc Prof Evelyn Koay, Centre Director, Molecular Diagnosis, Dept of Laboratory Medicine, National University Hospital

Provided by SingHealth

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