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Singapore scientists discover blindness genes

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Singapore: Scientists at Singapore Eye Research Institute and A*STAR's Genome Institute of Singapore have succeeded in identifying genes for central corneal thickness that may cause potentially blinding eye conditions. These eye conditions include glaucoma, as well as the progressive thinning of the cornea, which may eventually lead to a need for corneal transplantation.

The authors jointly led a multi-center study involving 55 hospitals and research centres around the world. They performed a meta-analysis on more than 20,000 individuals in European and Asian populations. Their findings were published in the prestigious science journal, *Nature Genetics*.

Central corneal thickness (CCT) is associated with potentially blinding eye conditions such as keratoconus, a condition where the cornea progressively thins and takes on a more conical shape that may eventually require transplantation. CCT has an estimated heritability up to 95 percent and may determine the severity of one's glaucoma and assist eye doctors in identifying patients with high risk for progression. In fact, it is one of the leading causes of corneal transplantation worldwide.

The Singapore team has had remarkable success identifying the most CCT-associated loci to date prior to this collaborative world-wide effort, by identifying six distinct genetic loci in two papers published in 2011 and 2012 via sample collections involving Singaporean Chinese, Indians, and Malays, as well as Beijing Chinese. However, none was found to be associated with common eye diseases like this study has now shown. Overall this new study identified a total of 27 associated loci, including six for the keratoconus.

These observations suggest that most of the CCT-associated loci identified from populations of European descent are shared with Asian populations. These findings show that Singapore is well placed globally in eye and genetics research in finding causes for sight threatening conditions. Eye doctors can in the future through genetic analysis better manage such patients, preventing regression of their conditions.