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Singapore scientists find genes associated with glaucoma

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Singapore scientists have identified three new genes associated with Primary Angle Closure Glaucoma (PACG), a leading cause of blindness in Chinese people. PACG affects 15 million people worldwide, 80% of whom live in Asia.

The discovery, published in the prestigious scientific journal, *Nature Genetics*, on 26 August 2012, was conducted collaboratively by scientists from the Singapore Eye Research Institute (SERI)/Singapore National Eye Centre (SNEC), Genome Institute of Singapore (GIS), National University of Singapore (NUS), National University Hospital's Department of Ophthalmology and Tan Tock Seng Hospital.

The team of scientists led an international consortium that carried out a genome-wide association study (GWAS) of 1,854 PACG cases and 9,608 controls of over five sample collections in Asia. They performed validation experiments in another 1,917 PACG cases and 8,943 controls collected from a further six sample collections from around the world. A total of 1,293 Singaporeans with PACG and 8,025 Singaporean controls were enrolled in this study. This work is the first to study PACG genetics using a genome-wide perspective.

This finding confirms the long-standing suspicion of Professor Aung Tin, the lead Principal Investigator of this project, who is Senior Consultant and Head of Glaucoma Service at SNEC, Deputy Executive Director at SERI, and Professor of Ophthalmology at NUS. Prof Aung has worked on PACG for over 10 years and believes from clinical

observations that the disease is strongly hereditary.

"This provides further evidence that genetic factors play a role in development of PACG," said Prof Aung. "It is a major achievement for our Singapore team leading the largest international consortium of doctors and scientists involved in glaucoma research. The results may lead to new insights into disease understanding and open the possibility of novel treatments in the future as well as the potential of early identification of people at risk of the disease."

Prof Wong Tien Yin, Executive Director at SERI and Provost's Chair Professor and Head, Department of Ophthalmology, NUHS, said, "This is a landmark finding, and may potentially change how we view PACG as a disease with genetic links. It highlights how a collective effort from scientists and clinicians and clinician-scientists can unravel diseases of major importance to Singapore. Because this disease is more common in Asians than in the Western populations, such studies will not be done in the US/Europe. This study has to be done in Asia as it is a disease with more implication for Asians. As such, Singapore has led the way forward."

Dr Khor Chiea Chuen, Principal Investigator, Human Genetics, at GIS added, "Modern genomics is a very powerful tool in dissecting the hereditable basis of common human diseases. It gives all of us a ray of hope, however far-fetched it may be, that one day we will be able to tailor treatments based on individual genetic profile."

"The information on genes involved in PACG has also opened up new and exciting research areas for us that we hope will culminate in new treatment modalities for angle closure glaucoma in the future," Said Dr Eranga Vithana, Associate Director, Basic and Experimental Sciences at SERI, and lead author of the paper.

Prof Janey Wiggs, Paul Austin Chandler Assoc. Professor of Ophthalmology, Harvard Medical School added, "This is a landmark study identifying three genes that contribute to angle-closure glaucoma, a form of glaucoma that is particularly common in Asians. These data are the first critical steps toward a better understanding of the underlying molecular events responsible for this blinding disease."

More information: The research findings described in the press release can be found in the 26 August 2012 advance online issue of *Nature Genetics* under the title "Genome-wide association analyses identify three new susceptibility loci for Primary Angle Closure Glaucoma".

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