UC San Diego Health

Researchers Identify Gene Variant Linked to Glaucoma

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An international team, led by researchers from the University of California, San Diego School of Medicine and the National Eye Institute, has discovered gene variants for glaucoma in a black population. The finding could lead to future treatments or a cure for this disease, which leads to blindness in two million Americans each year.

The study by Kang Zhang, MD, PhD, Director of the Institute for Genomic Medicine and professor of ophthalmology and human genetics at the Shiley Eye Center at UC San Diego and J. Fielding Hejtmancik, MD, PhD, medical officer and chief of the Ophthalmic Molecular Genetics Section at the National Eye Institute, National Institutes of Health, along with the Barbados Family Study Group and colleagues in the United States, China and Barbados, will be published in the early online edition of the *Proceedings of the National Academy of Science (PNAS)* the week of September 21.

κang Zhang, MD, PhD

Glaucoma is the leading cause of blindness among blacks, affecting close to five percent of the population. The researchers chose to conduct the study in the Afro-Caribbean population of Barbados, where the incidence of glaucoma is double that figure – nearly 10 percent of all residents of the island – and where there is a strong genetic predisposition.

Known as "the silent thief of sight," glaucoma is a neurodegenerative disease that causes the death of ganglion cells of the retina, resulting in gradual and irreversible loss of peripheral vision. Reducing intra-ocular pressure can slow the progression to blindness, but there is no cure or reversal for glaucoma.

"The cause and progression of glaucoma are poorly understood, although we know there is a strong genetic predisposition to the disease," said co-author Robert N. Weinreb, MD, Director of the Hamilton Glaucoma Center and Distinguished Professor of Ophthalmology at UC San Diego.

"We have now identified very common gene variants that have a dramatic impact on an individual's risk for developing glaucoma," Zhang added. "These gene variants are present in 40 percent of individuals with glaucoma in the Barbados population and explains nearly one-third of their genetic risk for the disease. This study should give us a better handle on earlier diagnoses and new therapies."

Looking at 249 patients with glaucoma and 128 control subjects, the research built on early studies which scanned the entire human genome. The scientists then homed in on a particular segment of the human genome, and finally localized the gene on chromosome 2.

"Once we understand the specific gene or protein structure that is altered in the disease, we are one step closer to developing gene or stem cell-based therapies to treat glaucoma," said Zhang. Identifying the gene variants can also provide a more accurate and earlier diagnosis, allowing early intervention to slow glaucoma's progression.

Additional contributors to the study include researchers at the University of Utah; Yale

University; Stony Brook University; University of the West Indies; Qingdao University, Qingdao, China; Sichuan Provincial People's Hospital, China; and West China Hospital, Sichuan University.

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