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Study links genes to common forms of glaucoma

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Results from the largest genetic study of glaucoma, a leading cause of blindness and vision loss worldwide, showed that two genetic variations are associated with primary open angle glaucoma (POAG), a common form of the disease. The identification of genes responsible for this disease is the first step toward the development of gene-based disease detection and treatment.

About 2.2 million people in the U.S. have glaucoma. POAG is often associated with increased eye pressure but about one-third of patients have normal pressure glaucoma (NPG). Currently, no curative treatments exist for NPG.

Researchers including lead author Janey Wiggs, M.D., Ph.D., and Lou Pasquale, M.D. Co-Directors of the Glaucoma the Harvard Glaucoma Center of Excellence, analyzed DNA sequences of 6,633 participants, half of whom had POAG. Participants were part of two NIH-funded studies: GLAUGEN (Glaucoma Genes and Environment) and NEIGHBOR (NEI Glaucoma Human genetics collaBORation), conducted at 12 sites in the United States. Dr. Pasquale is also Director of the Glaucoma Service at Mass. Eye and Ear.

The results, reported online in *PLoS Genetics* (April 26, 2012), found that two variations were associated with POAG, including NPG. These are the first variants commonly associated with NPG. One variant is in a gene located on chromosome 9 called CDKN2BAS whereas the other variant is in a region of chromosome 8 where it may affect the

expression of genes LRP12 or ZFPM2. These genes may interact with transforming growth factor beta (TGF-beta), a molecule that regulates cell growth and survival throughout the body. Previous studies have also implicated TGF-beta in glaucoma.

This study has provided important new insights into the disease pathogenesis and will make future studies focused on translating this information into the clinic possible. Ultimately we hope to prevent blindness caused by this very common eye disease," said lead author Dr. Wiggs.

"This study has identified an important molecular pathway in the development of POAG. Control of TGF-beta might lead to more effective therapies for this blinding disease," said Dr. Hemin Chin, associate director for Ophthalmic Genetics at the National Eye Institute.

Provided by Massachusetts Eye and Ear Infirmary

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