SOUTHWESTERN NEWS

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Gene variation could be responsible for age-related macular degeneration

DALLAS – March 10, 2005 – Half of all cases of age-related macular degeneration, the leading cause of blindness among the elderly, could be caused by a variation in a particular gene, according to UT Southwestern Medical Center researchers involved in a multicenter study.

The National Eye Institute study – which will appear in an upcoming edition of the journal *Science* and is available online – links a mutation in the gene *Complement Factor H* to an increased risk of age-related macular degeneration (AMD).

Macular degeneration is a complex disease that is the leading cause of blindness in Americans over the age of 50. By age 75, an estimated 30 percent of Americans have some manifestation of AMD.

The macula is an area in the center of the retina where light is focused and changed into nerve signals to compose an image in the brain. This central or "macular" vision enables us to read, drive and do things requiring fine, sharp, straight-ahead vision.

"We've identified a gene that is implicated in the pathogenesis of AMD," said Robert Ritter, a UT Southwestern research scientist involved in the *Science* study. "It provides a starting point for future investigations that will help us understand what takes place during the breakdown of the visual process."

Scientists have long suspected a genetic role in the disease but previously were only able to narrow the culprit gene's location to one region of a particular chromosome.

"We know that one of the most significant factors in determining who gets macular degeneration is family history," said Dr. Albert Edwards, the study's lead author and an assistant professor of ophthalmology at UT Southwestern when he conducted his research. "A positive family history can increase a person's chances of developing macular degeneration several fold compared to people in the general population."

In the study, researchers analyzed genetic data from more than 200 patients who were at high

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risk for developing AMD or who already had AMD in one or both eyes, and from more than 130 healthy participants without a known family history of the disease. The genetic mutation of *Complement Factor H* was present in half of those with AMD or at high risk for the disease.

Researchers from the UT Southwestern Eugene McDermott Center for Human Growth and Development provided genotyping, technical advice and assistance.

"This is an important study that gives us new insight into a disease that impacts a growing population of aging Americans," said Dr. Helen Hobbs, director of the center and chief of Clinical Genetics at UT Southwestern. "Genetic studies such as this provide the basis for clinicians to identify those at risk and may lead to better treatment options."

These findings may help researchers develop new preventive and therapeutic strategies for managing AMD.

"By finding genes, we can understand where the biological pathways are and the processes involved in the disease," said Dr. Edwards, who is now the president of the Institute for Retina Research at Presbyterian Hospital of Dallas. "Once we determine which genes are responsible for macular degeneration, we can screen the population and manipulate biological pathways to develop treatments."

Also involved in the study were researchers from Boston University School of Medicine and Sequenom, Inc.

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