Researchers Discover Gene Believed Responsible for Age-Related Macular Degeneration

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Researchers from Boston University School of Medicine (BUSM), UT Southwestern Medical Center at Dallas, and SEQUENOM, Inc., report the discovery of a genetic variation, that is the strongest known risk factor associated with age-related macular degeneration (AMD). Published in the March 10, 2005, online issue of the journal *Science*, the study entitled "Complement Factor H Polymorphism and Age-Related Macular Degeneration," details the discovery of the gene that may account for approximately fifty percent of the cases of AMD in the population.

"Considering that AMD is such a common and complex condition with multiple risk factors, we did not expect that we would identify a gene that is responsible for almost half of the cases," said author Lindsay A. Farrer, PhD, chief of the genetics program at BUSM, and a professor of medicine, neurology, genetics and genomics, epidemiology and biostatistics at BUSM.

The biological basis of this disorder, which is the most common cause of blindness in the elderly, is unclear. However, it was strongly suspected that genetics did play a role. The researchers tested single nucleotide polymorphisms (SNPs) for association of AMD in a region of 14 million base pairs on chromosome 1q25-31 where a gene for AMD had been previously localized in families with multiple persons affected with AMD.

Using two independent case-control populations, the researchers found that possession of at least one copy of histidine at position 402 of complement factor H (CFH) increased the risk of AMD almost three-fold.

“Given the rapid aging of the population, an estimated 3 million people will have complications of AMD by the year 2020. We hope our findings will create new avenues for developing preventative and therapeutic strategies for AMD,” added co-author and retina specialist Albert O. Edwards, MD, PhD, currently the President of the Institute for Retina Research at the Presbyterian Hospital of Dallas, Texas. Edwards conducted his research while on faculty at UT Southwestern.

“We are pleased to be part of this important collaboration with BU School of Medicine and UT Southwestern, and that the use of our MassARRAY® System helped contribute to the discovery of the genetic variations associated with AMD,” said Charles Cantor, PhD, SEQUENOMS’s chief scientific officer.

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About Boston University School of Medicine
Boston University School of Medicine is a leading academic and research institution, with an enrollment of nearly 630 students and nearly 2,400 full-time, part-time and volunteer faculty members. The School of Medicine is nationally known for its programs in Alzheimer’s disease, arthritis, cardiovascular disease, cancer, human genetics, pulmonary diseases and dermatology, among others. The School is affiliated with Boston Medical Center, its principal teaching hospital, and Boston Veterans Administration Medical Center. Along with Boston Medical Center, the School of Medicine is a partner in Boston HealthNet, a consumer-driven urban health network.

Source: Boston University