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Study offers new insight for understanding leading cause of blindness

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An international study has identified the number of genetic factors known to play a role in age-related macular degeneration (AMD), the worldwide leading cause of blindness in the elderly. This new discovery may help to further research into the biological processes that results in AMD and allow for the development of new therapeutics, including a personalized medicine approach for treating this debilitating disease.

AMD is a progressive ocular neurodegenerative disease affecting approximately 150 million people globally. The disease results in the loss of sharp, central vision by causing damage to the cells that sense light at the back of the eye. It is most common in adults over 50 and its development is believed to be influenced by a variety of factors including genetic pre-disposition and environmental and lifestyle factors such as tobacco use and diet. Currently no cure or preventive measure is available.

The International AMD Genomic Consortium, composed of 26 centers worldwide including Boston University Schools of Medicine and Public Health, collected and analyzed genetic data from more than 43,000 people to identify variations in genes associated with AMD. They found the number of loci (discrete genetic regions) involved in the development of AMD has expanded from 21 to 34, with a total of 52 variants discovered. These findings appear in the journal *Nature Genetics*.

"This new discovery is critical to furthering advances in AMD research

and development of new therapeutics as the variants associated with AMD risk and the genes that contain them are potential targets for novel drug targets," explained Lindsay A. Farrer, PhD, chief of the Biomedical Genetics division at Boston University School of Medicine and a co-leader of the study. "It is likely that the large number of genes implicated in our study collectively have roles in multiple pathways. Using the concept of personalized medicine, the goal would be to develop a drug to target a particular pathway for individuals having a discrete genetic profile," he added.

Provided by Boston University Medical Center