DUKE center for HUMAN GENETICS

Important AMD Research Update

Spring 2005

Gene Variant Found that Increases Risk of Age-Related Macular Degeneration

A research team at the Duke Center for Human Genetics (CHG) and the Duke University Eye Center (DUEC) has pinpointed the first major gene that contributes to an individual's risk for developing age-related macular degeneration (AMD). AMD, a chronic progressive disease that affects as many as 15 million people in the United States, is the leading cause of visual impairment and legal blindness in the elderly. Led by CHG director Margaret Pericak-Vance, PhD and the DUEC's Eric Postel, MD, researchers worked with partners from the Vanderbilt Center for Human Genetics Research, led by Jonathan Haines, PhD, to uncover genetic factors related to AMD.

The gene is called complement factor H (CFH). There are several different forms (variants) of the CFH gene, and every individual inherits one form from their mother and one from their father. Research estimates indicate that having one particular form of this gene explains approximately 43 percent of AMD cases. The Duke team identified the gene after screening 182 families and 495 other individuals affected by AMD.

The Duke researchers and their collaborators reported their findings in *Science* magazine (published online March 10, 2005, in *Science Express*). In the same issue of *Science*, two other research groups report similar findings about the CFH gene and AMD. It is unusual for three independent teams to find a gene at the same time for a disease as complex as AMD. Each group searched for AMD genes in different ways, and their combined findings mean the evidence for CFH's role in AMD is very convincing.

Is Testing Available for this Gene?

At this point, it is too early to offer genetic testing for CFH. While this finding is exciting and will lead to valuable knowledge about AMD, it does not yet have a practical use. There is still a lot to learn about the role this gene plays in AMD. If we, or others, continue to make positive discoveries pertaining to this gene, a clinical test will likely become available. In the event a genetic test is developed, we will notify you, our participants.

This finding helps us understand what causes AMD and could lead to new and more effective avenues for prevention and treatment. The advance may also help us to identify patients at the greatest risk for AMD before symptoms arise, when therapies and lifestyle changes might be most effective in slowing the disease progression.

Identifying a gene in a complex disease such as AMD is very exciting, but it will take many years of further study to identify a cure, including research into understanding how the CFH gene contributes to AMD.

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Published reports suggest that chronic inflammation plays a role in macular degeneration. In addition, other studies suggest that CFH aids in protecting blood vessels from inflammation and damage. Together, these findings suggest a role for CFH in development of AMD. Since we have shown that complement factor H is an important contributor to the disease, inflammation—or a lack of control of inflammation due to some difference in function of CFH—is likely to contribute to AMD.

Given that CFH is so important in AMD, accounting for over 40% of cases, further studies of CFH and the compounds with which it interacts might lead to a rapid increase in understanding the disease. That information, in turn, should allow scientists to advance on new treatments and preventive therapies.

Thank You! Your Participation Led to this Success!

We are indebted to all the individuals and family members who have so generously agreed to participate in this ongoing genetic research study for AMD. Each individual and, in turn, each family that participates, helps the pieces of this research puzzle fall into place. We look forward to continuing to work with everyone over the next few years, and look forward to additional discoveries. Together, we all move closer to our common goal of understanding why and how AMD occurs. Please feel free to contact us with any questions about the research at:

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How Else You Can Help

This groundbreaking AMD research was made possible through both federal grant support and gifts made by individual donors who provide us with the resources necessary to gather essential data from patients. Donors contribute to the purchase of cutting-edge equipment and the development of new technologies. They support innovative research.

In fact, individual gifts, large and small, provide an important source of non-government support. Even modest donations, when merged together, create a significant resource for the support of our research, teaching, and patient care.

To make a gift to AMD Genetics research at the Duke Center for Human Genetics and the Duke University Eye Center, please call (800) DUKE-CHG, or visit us online at www.chg.duke.edu/help. Gifts can be mailed to: Duke Center for Human Genetics/Duke University Eye Center AMD Genetics Research, DUMC Box 3445, Durham, NC 27710.

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