

New inherited eye disease discovered

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Scientists at the University of Iowa recently found a new inherited eye disease that affects the macula, which is part of the retina. In this image of the macula of an affected person, there are brown spots and cysts that cause blurring or blind spots in the person's vision. Credit: University of Iowa, 2009

University of Iowa researchers have found the existence of a new, rare inherited retinal disease. Now the search is on to find the genetic cause, which investigators hope will increase understanding of more common retinal diseases.

The findings appeared in the Nov. 9 issue of the *Archives of Ophthalmology*.

The macula, located within the retina, is an area of high-resolution central vision that is needed to read or drive, for example. This area is



damaged in more common retinal conditions such as macular degeneration and can be damaged by diabetes.

"It is rare to find a new inherited eye disease that affects the macula. We thought we had seen them all," said the study's lead author Vinit Mahajan, M.D., Ph.D., assistant professor of ophthalmology and visual sciences at the University of Iowa Roy J. and Lucille A. Carver College of Medicine.

"This newly found <u>retinal disease</u> causes abnormal <u>blood vessels</u> in the macula, and these vessels are prone to bleeding. This causes swelling or scars that 'black out' or blur parts of the field of vision," said Mahajan, who also is a retinal specialist with University of Iowa Hospitals and Clinics.

The finding came about when one person in a family in the United States sought care for <u>eye problems</u>. "If a doctor saw just one family member, they would probably call this macular degeneration. We knew there was something different, and we had to examine the rest of the family," Mahajan said.

The team assessed 20 extended family members who were not blind but had visual problems of different severities. Some family members also had areas of central <u>vision loss</u>, and some family members had strabismus, a disorder in which the eyes are not aligned.

The University of Iowa researchers have presented their findings at international meetings of retinal specialists in Arizona, Florida and London. The investigators are now working with researchers worldwide to determine if other people have this particular disease.

"Through our paper and by sharing pictures of what the affected <u>eye</u> looks like, we hope to find more people affected," Mahajan said. "We



also will work to find the gene that causes the condition. This information could be very useful in eventually preventing or treating this and other diseases that affect the macula."

The advanced genetics research capabilities at the University of Iowa Carver Family Center for Macular Degeneration increase the likelihood of finding a gene, Mahajan said.

More information: Archives of Ophthalmology paper online.

Source: University of Iowa (<u>news</u>: <u>web</u>)

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