

**PEOPLE STRUCK WITH THE BLINDING DISEASE AGE-RELATED MACULAR DEGENERATION ONCE FOUND LITTLE HELP. BUT NOW RESEARCH IS LEADING TO A BETTER UNDERSTANDING OF THE DISEASE AND TO THE DEVELOPMENT OF IMPROVED TREATMENTS.**

**SOME OF THE LATEST ADVANCES INVOLVE THE STUDY OF THOSE SPECIAL CELL INHABITANTS KNOWN AS GENES THAT CONTROL EVERY TRAIT AND CHEMICAL PROCESS IN THE BODY. RECENTLY, SCIENTISTS DISCOVERED THAT VARIATIONS IN ONE SPECIFIC GENE MAY BE COMMON TO AS MANY AS HALF OF THOSE WITH AGE-RELATED MACULAR DEGENERATION. THERAPIES THAT TARGET THIS CULPRIT GENE AND PREVENT OR SLOW VISION LOSS MAY FOLLOW.**

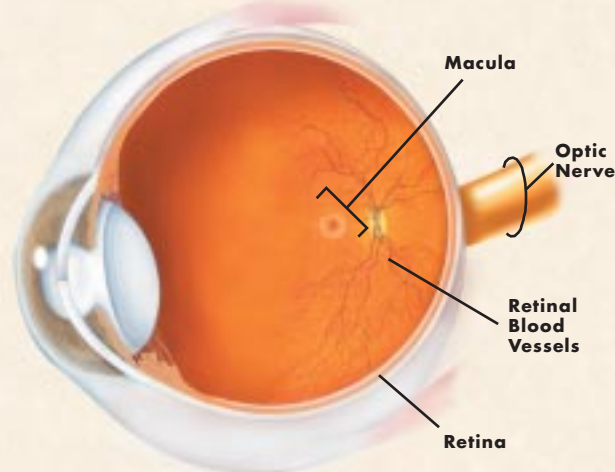
### **AGE-RELATED MACULAR DEGENERATION AND GENES**

While navigating the mall's crowd, your elderly aunt looks right at you and then detours into the food court. A cold, hard snub, you think.

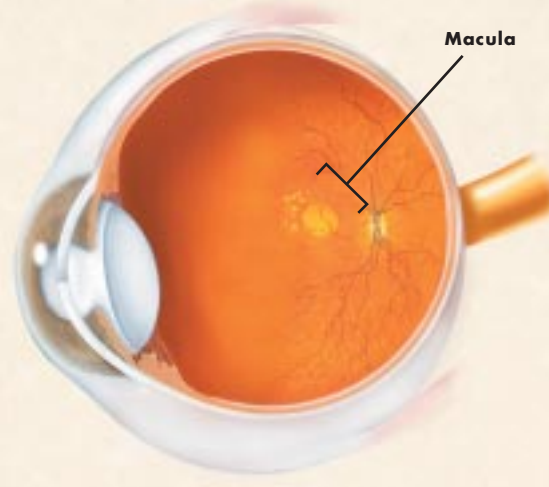
In reality, the slight may stem from something more troubling: age-related macular degeneration. A leading cause of vision loss in older Americans, this blinding disease, also known as AMD, robs the sight of some 1.8 million people age 40 and over in the United States. Those with the disorder may notice that the center or straight-ahead part of their vision becomes blurry. Details fade. Straight lines appear wavy. Shapes distort. Some experience actual blind spots. It can be hard to recognize faces and socialize, read, write, sew, drive, watch TV, and take part in many other pleasures of independent living.

Once there was little aid for those with AMD. But now, thanks to basic science research, options are available that can slow the vision loss in some people with the disorder, and even more help is on the way. Some of the latest advances are arising from the study of genes, including the discovery that variations in one specific gene may be com-

#### **A. Normal eye (oblique cross section)**



#### **B. Age-related macular degeneration**



**▲ OUR ABILITY TO SEE RELIES ON THE RETINA, A PAPER-THIN TISSUE THAT LINES THE BACK OF THE EYE AND SENDS VISUAL SIGNALS VIA THE OPTIC NERVE TO THE BRAIN. AGE-RELATED MACULAR DEGENERATION IS THOUGHT TO HARM VISION AS THE RESULT OF DAMAGE TO THE MACULA, A GROUP OF CELLS IN A TINY AREA OF THE RETINA THAT HELP PRODUCE CENTRAL, FINE VISION. ONE FORM, KNOWN AS DRY AGE-RELATED MACULAR DEGENERATION, IS THOUGHT TO INVOLVE A SLOW BREAKDOWN OF CELLS IN THE MACULA AND GRADUAL VISION LOSS. IN ANOTHER, SEVERE FORM, TERMED WET AGE-RELATED MACULAR DEGENERATION, ABNORMAL BLOOD VESSELS GROW RAPIDLY IN THE EYE AND LEAK, SWIFTLY DAMAGING THE MACULA AND VISION. SCIENTISTS HOPE GENETIC STUDIES AND RESEARCH INTO THE MECHANISM OF INFLAMMATION WILL LEAD TO NEW TREATMENTS FOR AMD.**

IMAGE BY LYDIA KIBLUK.

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mon to about half of those with AMD. The new work is leading to:

- Improved understanding of the roots of vision loss.
- Possible gene-targeted therapies.

AMD is thought to harm sight by damaging cells in the eye's macula, a tiny area made up of millions of light-sensing cells that help produce central vision (see image). No therapy can restore these cells and bring back lost vision, but some available treatments can sometimes slow the progression of the disease. For example, taking certain nutrients under the advice of a physician may help guard cells in the macula, and thus vision. Scientists found that a high dose of the mineral zinc and antioxidant nutrients, which are known to help maintain healthy cells and tissues, can sometimes reduce the risk of a severe stage of AMD and its accompanying vision loss in those who do not yet have advanced AMD or those who

have severe AMD in only one eye. Other available techniques can sometimes slow advanced forms of the disorder by targeting abnormal blood vessels. In certain severe cases these vessels grow and leak inside the fragile eye, rapidly damaging the macula. None of the treatments, however, is broadly effective.

New research suggests that more options may arise from the study of genes. Our genes and the proteins they produce play a key role in how we function. And variations in their make-up can help trigger disease. Several recent studies report that harboring a variation of a gene called complement factor H increases the risk that a person could develop AMD, possibly accounting for as many as 50 percent of cases. The complement gene produces a protein that is thought to regulate a specific inflammation response, which is designed to attack infections. In AMD, researchers think that the complement gene variation

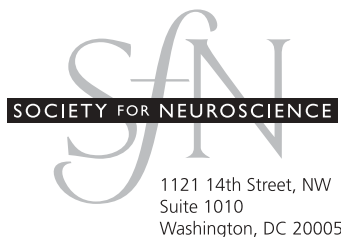
may produce an irregular protein that does not appropriately control this response, resulting in damage to cells in the macula. Work also indicates that abnormal deposits found in the eyes of those with AMD contain components of this complement response system, which further backs the idea that it is involved in AMD.

Currently, scientists are examining ways to adjust the complement system and treat AMD. Tests of a protein that targets the complement system, termed vaccinia virus complement control protein, are underway in a mouse model of AMD.

This line of study could also help improve the treatment of additional disorders where inflammation may play a role, including spinal cord injury, which impairs the movement of numerous Americans.

With continued study, researchers may soon improve the treatment of a wide array of people. In the meantime, cut your aunt some slack.

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