



Hereditry and Age-Related Macular Degeneration

Age-related macular degeneration (AMD) typically affects individuals over 50 years old. Scientific evidence shows that genes may play a role in the development of nearly three out of four cases of this devastating eye disease.

Several genes are believed to be strongly associated with the risk of developing AMD:

- **Factor H and Factor B genes** are responsible for proteins that help regulate inflammation in the part of the immune system that attacks diseased and damaged cells. According to study results published in 2006 by Columbia University, 74 percent of AMD patients carry certain variants in one or both of these genes, and these may significantly increase their risk of developing it.
- **PLEKHA1** – a gene located on chromosome 10; researchers believe it may increase the risk of developing AMD. Like Factors H and B, PLEKHA1 appears to be involved in the cellular processes related to inflammation.
- **LOC387715** – A certain variation of this gene appears to increase the risk of developing AMD. This risk is further heightened if a person with this gene variation also smokes.
- **HTRA1** – Scientists have identified a link between a mutation in this gene and the development of AMD. Specifically, the HTRA1 mutation is thought to be associated with the formation of drusen (yellow deposits of waste products under the retina that are often a sign of dry AMD), and may also promote the growth of fragile new blood vessels typical of wet AMD.

- **Complement C3** – Researchers have found that a variant in this gene increases the risk of developing the wet and dry forms of AMD. This gene plays an important role in the immune system, leading scientists to believe that inflammation is a vital part of the AMD disease process.

Other gene candidates are being studied to determine their role in AMD. While there is definitely a strong genetic component to this disease, it is highly likely that its development is due to a combination of multiple factors including gene mutations or variations and environmental factors such as sunlight exposure, diet and smoking.

This information should not in any way substitute for the advice of a qualified healthcare professional and is not intended to constitute medical advice. For further information, contact Macular Degeneration Research, a program of the American Health Assistance Foundation, at 22512 Gateway Center Drive, Clarksburg, Maryland 20871, call 1-800-437-2423, or visit our website at www.ahaf.org

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