ARMS2 gene

age-related maculopathy susceptibility 2

Normal Function

The *ARMS2* gene provides instructions for making a protein whose function is unknown. Studies suggest that the ARMS2 protein is found primarily in the placenta and in the specialized light-sensing tissue in the back of the eye (the retina). However, it is unclear what role, if any, the protein plays in early development or normal vision.

Health Conditions Related to Genetic Changes

Age-related macular degeneration

The *ARMS2* gene is located on the long (q) arm of chromosome 10 in a region known as 10q26. This region has been strongly associated with the risk of developing age-related macular degeneration, a common cause of vision loss in older adults. Researchers have identified several variations (polymorphisms) in and near the *ARMS2* gene that may explain the association between the 10q26 region and age-related macular degeneration. The best-studied of these variations, known as rs10490924, alters a single protein building block (amino acid) in the ARMS2 protein. Another common variation, a complex change that deletes a segment of the *ARMS2* gene and inserts new genetic material, may also contribute to disease risk.

It is unclear how polymorphisms in the *ARMS2* gene might be related to age-related macular degeneration. In the 10q26 region, the *ARMS2* gene is located next to a gene called *HTRA1*; changes in this gene have also been studied as a risk factor for the disease. Because the two genes are so close together, it is difficult to tell whether changes in one gene or the other, or possibly changes in both genes, account for the increased disease risk. Age-related macular degeneration is a complex condition that likely results from a combination of multiple genetic and environmental factors.

Other Names for This Gene

- age-related maculopathy susceptibility protein 2
- ARMD8
- ARMS2_HUMAN
- LOC387715

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ARMS2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=387715[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ARMS2%5BTIAB%5D%29+OR+%28LOC387715%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ARMS2 GENE; ARMS2 (https://omim.org/entry/611313)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/387715)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ARMS2[gene])

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Genomic Location

The *ARMS2* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromosome/10/).

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