CFHR5 gene

complement factor H related 5

Normal Function

The *CFHR5* gene provides instructions for making a protein called complement factor H-related 5. The precise function of this protein is unknown. However, its structure is similar to that of a protein called complement factor H (which is produced from the *CFH* gene). This similarity provides clues to the probable function of complement factor H-related 5.

Complement factor H regulates a part of the body's immune response known as the complement system. The complement system is a group of proteins that work together to destroy foreign invaders (such as bacteria and viruses), trigger inflammation, and remove debris from cells and tissues. This system must be carefully regulated so it targets only unwanted materials and does not damage the body's healthy cells. Complement factor H helps to protect healthy cells by preventing the complement system from being turned on (activated) when it is not needed. Studies suggest that complement factor H-related 5 also plays a role in controlling the complement system.

Health Conditions Related to Genetic Changes

Atypical hemolytic-uremic syndrome

MedlinePlus Genetics provides information about Atypical hemolytic-uremic syndrome

C3 glomerulopathy

Several mutations in the *CFHR5* gene have been found to cause a rare form of kidney disease called C3 glomerulopathy. This disorder damages the kidneys and can lead to end-stage renal disease (ESRD), a life-threatening condition that prevents the kidneys from filtering fluids and waste products from the body effectively.

The most common *CFHR5* gene mutation has been identified in people from the Mediterranean island of Cyprus. This genetic change abnormally copies (duplicates) regions of the *CFHR5* gene known as exons 2 and 3. The duplication alters the structure and function of complement factor H-related 5, preventing it from regulating the complement system effectively. As a result, the complement system becomes overactive, which damages structures called glomeruli in the kidneys. These structures

are clusters of tiny blood vessels that help filter waste products from the blood. Damage to glomeruli prevents the kidneys from filtering waste products normally and can lead to ESRD.

Several other changes involving the *CFHR5* gene do not cause C3 glomerulopathy directly but appear to increase the likelihood of developing the disorder. It is unclear how variations in this gene affect the regulation of the complement system, and researchers are still working to determine how these genetic changes contribute to disease risk.

Age-related macular degeneration

MedlinePlus Genetics provides information about Age-related macular degeneration

Other Names for This Gene

- CFHL5
- complement factor H-related 5
- complement factor H-related protein 5
- factor H-related protein 5
- FHR-5
- FHR5
- FHR5_HUMAN
- FLJ10549
- MGC133240

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CFHR5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=81494[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CFHR5%5BTIAB%5D%29+OR+%28%28complement+factor+H-related+protein+5%5BTIAB%5D%29+OR+%28factor+H-related+protein+5%5BTIAB%5D%29+OR+%28FHR-5%5BTIAB%5D%29+OR+%28FHR5%5BTIAB%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

- COMPLEMENT FACTOR H-RELATED 5; CFHR5 (https://omim.org/entry/608593)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/81494)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CFHR5[gene])

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Genomic Location

The *CFHR5* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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