CFI gene

complement factor I

Normal Function

The *CFI* gene provides instructions for making a protein called complement factor I. This protein helps regulate 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 attack the body's healthy cells. Complement factor I and several related proteins protect healthy cells by preventing activation of the complement system when it is not needed.

Health Conditions Related to Genetic Changes

Complement factor I deficiency

At least 10 mutations in the *CFI* gene have been identified in people with complement factor I deficiency, a disorder characterized by immune system dysfunction. The mutations result in abnormal, nonfunctional, or absent complement factor I.

The lack (deficiency) of functional complement factor I protein allows uncontrolled activation of the complement system. The unregulated activity of the complement system decreases blood levels of another complement protein called C3, reducing the immune system's ability to fight infections. In addition, the immune system may malfunction and attack its own tissues, resulting in autoimmune disorders.

Age-related macular degeneration

MedlinePlus Genetics provides information about Age-related macular degeneration

Atypical hemolytic-uremic syndrome

MedlinePlus Genetics provides information about Atypical hemolytic-uremic syndrome

C3 glomerulopathy

MedlinePlus Genetics provides information about C3 glomerulopathy

Other disorders

Mutations in the *CFI* gene have also been found in people with glomerulonephritis with isolated C3 deposits. This condition, which may also occur in people with complement factor I deficiency, is characterized by kidney malfunction that can be serious or life-threatening. The *CFI* gene mutations identified in this disorder result in an abnormal or nonfunctional version of complement factor I. The defective protein allows uncontrolled activation of the complement system. The overactive complement system attacks certain kidney cells, which damages the kidneys and leads to a loss of protein in the urine (proteinuria).

A common variation (polymorphism) in the *CFI* gene has also been associated with age-related macular degeneration (AMD). AMD is a leading cause of vision loss among older adults. It is characterized by damage to the retina and a loss of sharp vision (visual acuity). Researchers suggest that the *CFI* gene variation that has been associated with AMD changes the way the gene is activated (expressed). It is unclear how this change is related to the development of AMD. A combination of genetic and environmental factors likely determines the risk of developing this complex eye disorder.

Other Names for This Gene

- AHUS3
- C3b-INA
- C3b-inactivator
- C3B/C4B inactivator
- C3BINA
- CFAI_HUMAN
- complement component I
- complement control protein factor I
- complement factor I heavy chain
- complement factor I preproprotein
- FI
- IF
- KAF
- Konglutinogen-activating factor
- light chain of factor I

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CFI (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3426[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CFI%5BTIAB%5D%29+OR+%28complement+factor+I%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

- COMPLEMENT FACTOR I; CFI (https://omim.org/entry/217030)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3426)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CFI[gene])

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Genomic Location

The *CFI* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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