

Complement component 8 deficiency

Description

Complement component 8 deficiency is a disorder that causes the immune system to malfunction, resulting in a form of immunodeficiency. Immunodeficiencies are conditions in which the immune system is not able to protect the body effectively from foreign invaders such as bacteria. People with complement component 8 deficiency have a significantly increased risk of recurrent bacterial infections, particularly by a bacterium called *Neisseria meningitidis*. Infection by this bacterium causes inflammation of the membranes surrounding the brain and spinal cord (meningitis). Although meningitis can be life-threatening, individuals with complement component 8 deficiency are less likely to die from the infection than people in the general population who contract it.

The severity of complement component 8 deficiency varies widely. While some people with this condition experience one or more infections, others do not have any health problems related to the disorder.

There are two types of complement component 8 deficiency, types I and II, classified by their genetic cause. The two types have the same signs and symptoms.

Frequency

Complement component 8 deficiency is a rare disorder, although its prevalence is unknown. Type I occurs in several populations, particularly in people with Hispanic, Japanese, or African Caribbean heritage, whereas type II primarily occurs in people of Northern European descent.

Causes

Complement component 8 deficiency is caused by mutations in the *C8A* or *C8B* gene. *C8A* gene mutations underlie type I, and *C8B* gene mutations cause type II. These genes provide instructions for making pieces of a protein complex called complement component 8. This complex is composed of an alpha subunit, produced from the *C8A* gene, a beta subunit, produced from the *C8B* gene, and a gamma subunit, produced from another gene called *C8G*.

Complement component 8 aids in 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, trigger inflammation, and remove debris from cells and

tissues. Complement component 8 combines with several other complement proteins to form the membrane attack complex (MAC), which inserts itself in the outer membrane of bacterial cells. This complex creates a hole (pore) in the membrane, which kills the bacterium.

Mutations in either *C8A* or *C8B* lead to a shortage of the respective subunit, which impairs formation of complement component 8. Without complement component 8, membrane attack complexes cannot form, which impairs the immune response, particularly against *Neisseria meningitidis*. This impaired immune response leaves affected individuals prone to recurrent episodes of meningitis.

[Learn more about the genes associated with Complement component 8 deficiency](#)

- C8A
- C8B

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- C8 deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Type I complement component 8 deficiency (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151081/>)
- Genetic Testing Registry: Type II complement component 8 deficiency (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151080/>)

Genetic and Rare Diseases Information Center

- Complement component 8 deficiency, type I (<https://rarediseases.info.nih.gov/diseases/10626/index>)
- Complement component 8 deficiency, type II (<https://rarediseases.info.nih.gov/diseases/10625/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Catalog of Genes and Diseases from OMIM

- COMPLEMENT COMPONENT 8 DEFICIENCY, TYPE II; C8D2 (<https://omim.org/entry/613789>)
- COMPLEMENT COMPONENT 8 DEFICIENCY, TYPE I; C8D1 (<https://omim.org/entry/613790>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28complement+component+8+deficiency%5BTIAB%5D%29+OR+%28C8+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

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