

C8B gene

complement C8 beta chain

Normal Function

The *C8B* gene provides instructions for making one piece, the beta subunit, of a protein complex called complement component 8. To form this complex, the beta subunit interacts with another piece made up of the alpha subunit (produced from the *C8A* gene) and the gamma subunit (produced from the *C8G* gene), which are linked.

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. This part of the immune response appears to be especially important for fighting infection by bacteria in the *Neisseria* genus.

Health Conditions Related to Genetic Changes

Complement component 8 deficiency

Several mutations in the *C8B* gene cause complement component 8 deficiency type II. This condition is an immune system disorder, known as an immunodeficiency, 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 developing recurrent infections, particularly by *Neisseria meningitidis*, which causes meningitis, a serious condition that involves inflammation of the membranes surrounding the brain and spinal cord.

C8B gene mutations involved in complement component 8 deficiency are most often of a type called a C to T transition, in which a DNA building block (nucleotide) called cytosine (C) is changed to the nucleotide thymine (T). Most commonly, this change occurs in a region of the gene called exon 9, but it can occur in other regions. These mutations alter the sequence of protein building blocks, resulting in an abnormally short protein that, if produced, is likely broken down quickly. Other types of mutations can occur in the *C8B* gene; they also lead to an abnormally short protein that is quickly broken down. The resulting shortage of this protein impairs formation of complement

component 8. Deficiency of this component prevents formation of membrane attack complexes. Without this part of the immune response, affected individuals are prone to infection, particularly by *Neisseria* bacteria.

Other Names for This Gene

- C82
- complement component 8 subunit beta
- complement component 8, beta polypeptide
- complement component C8 beta chain isoform 1 preproprotein
- complement component C8 beta chain isoform 2
- complement component C8 beta chain isoform 3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of C8B ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=732\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=732[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28C8B%5BTIAB%5D%29+OR+%28complement+component+8,+beta+polypeptide%5BTIAB%5D%29%29+OR+%28%28C82%5BTIAB%5D%29+OR+%28complement+component+8+subunit+beta%5BTIAB%5D%29+OR+%28complement+component+C8+beta+chain+isoform+1+preproprotein%5BTIAB%5D%29+OR+%28complement+component+C8+beta+chain+isoform+2%5BTIAB%5D%29+OR+%28complement+component+C8+beta+chain+isoform+3%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%29>)

Catalog of Genes and Diseases from OMIM

- COMPLEMENT COMPONENT 8, BETA SUBUNIT; C8B (<https://omim.org/entry/120960>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/732>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=C8B\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=C8B[gene]))

References

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Genomic Location

The *C8B* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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