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.
**Chromosomal Location**

Cytogenetic Location: 1p32.2, which is the short (p) arm of chromosome 1 at position 32.2

Molecular Location: base pairs 56,929,207 to 56,974,383 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

**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**

**Educational Resources**

- Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): The Terminal Complement Proteins Polymerize to Form Pores in Membranes that can Kill Certain Pathogens
  https://www.ncbi.nlm.nih.gov/books/NBK27100/#_A186_

- Molecular Biology of the Cell (fourth edition, 2002): Complement Activation Targets Pathogens for Phagocytosis or Lysis
  https://www.ncbi.nlm.nih.gov/books/NBK26846/#_A4679_
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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+OR+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28%28%28Genetic+Phenomena%5BMH%5D%29+AND+english+%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- COMPLEMENT COMPONENT 8, BETA SUBUNIT
  http://omim.org/entry/120960

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=C8B%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:732

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P07358

Sources for This Summary


- OMIM: COMPLEMENT COMPONENT 8, BETA SUBUNIT
  http://omim.org/entry/120960


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26074922
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4443744/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15565265

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/7594510

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