MCCC2 gene
methylcrotonoyl-CoA carboxylase 2

Normal Function

The *MCCC2* gene provides instructions for making one part (the beta subunit) of an enzyme called 3-methylcrotonoyl-CoA carboxylase or 3-MCC. Beta subunits join with larger alpha subunits made from the *MCCC1* gene; six of these pairings together form a functioning enzyme.

The 3-MCC enzyme is found in mitochondria, which are the energy-producing centers inside cells. This enzyme plays a critical role in breaking down proteins obtained from the diet. Specifically, it is responsible for the fourth step in the breakdown of leucine, an amino acid that is a building block of many proteins. This step converts a molecule called 3-methylcrotonyl-CoA to a molecule called 3-methylglutaconyl-CoA. Additional chemical reactions convert 3-methylglutaconyl-CoA into molecules that are later used for energy.

Health Conditions Related to Genetic Changes

3-methylcrotonyl-CoA carboxylase deficiency

More than 40 mutations in the *MCCC2* gene have been identified in people with 3-methylcrotonyl-CoA carboxylase deficiency (also known as 3-MCC deficiency). Most of these mutations change single amino acids in 3-MCC, which severely reduces the activity of the enzyme. Other mutations prevent the production of any functional enzyme. As a result, leucine cannot be broken down properly, and byproducts of leucine processing build up to toxic levels in the body. These toxic substances can damage the brain, causing the characteristic signs and symptoms of 3-MCC deficiency.
**Chromosomal Location**

Cytogenetic Location: 5q13.2, which is the long (q) arm of chromosome 5 at position 13.2

Molecular Location: base pairs 71,587,340 to 71,658,706 on chromosome 5 (Homo sapiens Updated Annotation Release 109.20191205, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- 3-methylcrotonyl-CoA carboxylase non-biotin-containing subunit
- MCCase subunit beta
- MCCB
- MCCB_HUMAN
- methylcrotonoyl-CoA carboxylase 2 (beta)
- non-biotin containing subunit of 3-methylcrotonyl-CoA carboxylase

**Additional Information & Resources**

**Educational Resources**

- Basic Neurochemistry (sixth edition, 1999): Major pathways of branched-chain amino acid metabolism
  https://www.ncbi.nlm.nih.gov/books/NBK20436/figure/A3097/

**Scientific Articles on PubMed**

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28MCCC2%5BTIAB%5D%29+OR+%28MCCB%5BTIAB%5D%29+OR+%283-methylcrotonyl-CoA+carboxylase%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- 3-METHYLCROTONYL-CoA CARBOXYLASE 2
  http://omim.org/entry/609014

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MCCC2%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:64087

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/Q9HCC0

Sources for This Summary

  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC199271/


  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1235267/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11406611

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16835865

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