

MCCC2 gene

methylecrotonyl-CoA carboxylase subunit 2

Normal Function

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

The 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 food. 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-methylecrotonyl-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-methylecrotonyl-CoA carboxylase deficiency

Many variants (also called mutations) in the *MCCC2* gene have been identified in people with 3-methylecrotonyl-CoA carboxylase deficiency (also called MCC deficiency). MCC deficiency is an inherited disorder in which the body is unable to process certain proteins properly. Most of these variants change single amino acids in MCC, which severely reduces the activity of the enzyme. Other variants prevent the production of any functional enzyme. As a result, leucine cannot be broken down properly, and byproducts of leucine processing can build up in the body. Some people with these genetic changes will show signs and symptoms of MCC deficiency.

Other Names for This Gene

- 3-methylecrotonyl-CoA carboxylase 2
- 3-methylecrotonyl-CoA carboxylase non-biotin-containing subunit

- 3-methylcrotonyl-CoA carboxylase, beta
- MCCase subunit beta
- MCCB
- MCCCbeta
- MCCCβ
- methylcrotonoyl-CoA carboxylase 2
- methylcrotonoyl-CoA carboxylase beta
- methylcrotonoyl-Coenzyme A carboxylase 2 (beta)

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MCCC2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=64087\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=64087[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28MCCC2%5BTIAB%5D%29+OR+%28%28MCCB%5BTIAB%5D%29+OR+%283-methylcrotonyl-CoA+carboxylase%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+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- 3-METHYLCROTONYL-CoA CARBOXYLASE 2; MCCC2 (<https://omim.org/entry/609014>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/64087>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MCCC2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MCCC2[gene]))

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Genomic Location

The *MCCC2* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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