MCCC1 gene

methylcrotonyl-CoA carboxylase subunit 1

Normal Function

The *MCCC1* gene provides instructions for making one part (the alpha subunit) of an enzyme called 3-methylcrotonoyl-CoA carboxylase or MCC. These alpha subunits join with smaller beta subunits made from the *MCCC2* gene; six of these pairings together form a functioning enzyme. The alpha subunit also includes a region for binding to the B vitamin biotin, which is required for the enzyme to function.

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-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

Many variants (also called mutations) in the *MCCC1* gene have been identified in people with 3-methylcrotonyl-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, but a few lead to the production of an abnormally short version of the enzyme. Variants in the *MCCC1* gene severely reduce or eliminate the activity of MCC. 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-methylcrotonyl-CoA carboxylase 1
- 3-methylcrotonyl-CoA carboxylase alpha
- 3-methylcrotonyl-CoA carboxylase biotin-containing subunit

- MCCA
- MCCase subunit alpha
- MCCCalpha
- MCCCα
- methylcrotonoyl-CoA carboxylase 1
- methylcrotonoyl-CoA carboxylase 1 alpha

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MCCC1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=56922[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MCCC1%5BTIAB%5D%29+OR+%28%28MCCA%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)

Catalog of Genes and Diseases from OMIM

- 3-METHYLCROTONYL-CoA CARBOXYLASE 1; MCCC1 (https://omim.org/entry/609010)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/56922)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MCCC1[gene])

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Genomic Location

The *MCCC1* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

Last updated March 7, 2024