MCEE gene

methylmalonyl-CoA epimerase

Normal Function

The *MCEE* gene provides instructions for making an enzyme called methylmalonyl CoA epimerase, which converts one form of the molecule methylmalonyl CoA to another. Specifically, the enzyme converts D-methylmalonyl CoA to L-methylmalonyl CoA. This conversion takes place within the pathway that converts the molecule propionyl-CoA to succinyl-CoA. This pathway is important in the breakdown of certain protein building blocks (amino acids), specific fats (lipids), and cholesterol.

Health Conditions Related to Genetic Changes

Methylmalonic acidemia

At least three mutations in the *MCEE* gene have been found to cause methylmalonic acidemia, a condition characterized by feeding difficulties, developmental delay, and long-term health problems. These mutations are thought to result in the production of a methylmalonyl CoA epimerase enzyme with little or no function. People with methylmalonic acidemia caused by mutations in the *MCEE* gene typically have milder signs and symptoms than people with the condition caused by mutations in other genes. The features may be milder because there is an alternate pathway for the conversion of propionyl-CoA to succinyl-CoA that does not involve methylmalonyl CoA epimerase, so some succinyl-Co is produced even when there are mutations in the *MCEE* gene. This alternate pathway cannot compensate for the breakdown of certain molecules that occurs in the regular pathway, so people with *MCEE* gene mutations still have a buildup of the byproducts of some amino acids and certain fats. As a result, these toxic compounds build up in the body's organs and tissues, causing the signs and symptoms of methylmalonic acidemia.

Other Names for This Gene

- DL-methylmalonyl-CoA racemase
- GLOD2
- glyoxalase domain containing 2
- MCEE_HUMAN
- methylmalonyl CoA epimerase

- methylmalonyl-CoA epimerase, mitochondrial
- methylmalonyl-CoA epimerase, mitochondrial precursor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MCEE (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=84693[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MCEE%5BTIAB%5D%29+OR+%28methylmalonyl+CoA+epimerase%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

- METHYLMALONYL-CoA EPIMERASE; MCEE (https://omim.org/entry/608419)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/84693)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MCEE[gene])

References

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- Dobson CM, Gradinger A, Longo N, Wu X, Leclerc D, Lerner-Ellis J, Lemieux M, Belair C, Watkins D, Rosenblatt DS, Gravel RA. Homozygous nonsense mutation inthe MCEE gene and siRNA suppression of methylmalonyl-CoA epimerase expression: anovel cause of mild methylmalonic aciduria. Mol Genet Metab. 2006Aug;88(4):327-33. doi: 10.1016/j.ymgme.2006.03.009. Epub 2006 May 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16697227)
- Gradinger AB, Belair C, Worgan LC, Li CD, Lavallee J, Roquis D, Watkins D, Rosenblatt DS. Atypical methylmalonic aciduria: frequency of mutations in themethylmalonyl CoA epimerase gene (MCEE). Hum Mutat. 2007 Oct;28(10):1045.

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Genomic Location

The *MCEE* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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