ACSF3 gene

acyl-CoA synthetase family member 3

Normal Function

The *ACSF3* gene provides instructions for making an enzyme involved in the formation (synthesis) of fatty acids, which are building blocks used to make fats (lipids). The ACSF3 enzyme performs a chemical reaction that converts malonic acid to malonyl-CoA, which is the first step of fatty acid synthesis. Based on this activity, the enzyme is classified as a malonyl-CoA synthetase. The ACSF3 enzyme also converts methylmalonic acid to methylmalonyl-CoA, making it a methylmalonyl-CoA synthetase as well.

Fatty acid synthesis occurs through two pathways, one of which takes place in cellular structures called mitochondria. Mitochondria convert the energy from food into a form that cells can use, and fatty acid synthesis in these structures is thought to be important for their proper functioning. The ACSF3 enzyme is found only in mitochondria and is involved in mitochondrial fatty acid synthesis.

Health Conditions Related to Genetic Changes

Combined malonic and methylmalonic aciduria

About a dozen mutations in the *ACSF3* gene have been found in people with combined malonic and methylmalonic aciduria (CMAMMA), a condition characterized by elevated levels of chemicals known as malonic acid and methylmalonic acid in the body. This condition can cause development and growth problems beginning in childhood or neurological problems beginning in adulthood.

Most *ACSF3* gene mutations involved in CMAMMA change single protein building blocks (amino acids) in the ACSF3 enzyme. The altered enzyme likely has little or no function. Because the enzyme cannot convert malonic and methylmalonic acids, they build up in the body. Damage to organs and tissues caused by accumulation of malonic and methylmalonic acids may be responsible for the signs and symptoms of CMAMMA, although the mechanisms are unclear.

Other Names for This Gene

- ACSF3_HUMAN

- acyl-CoA synthetase family member 3, mitochondrial
- acyl-CoA synthetase family member 3, mitochondrial precursor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACSF3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=197322[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ACSF3%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ACYL-CoA SYNTHETASE FAMILY, MEMBER 3; ACSF3 (https://omim.org/entry/614245)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/197322)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACSF3[gene])

References

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Genomic Location

The *ACSF3* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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