



## 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

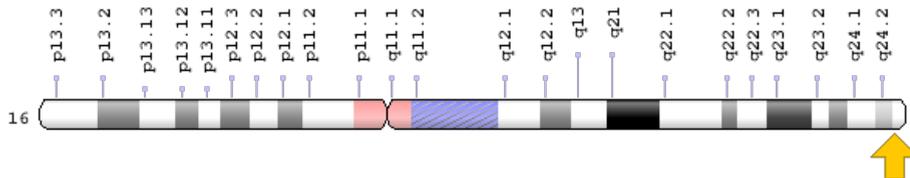
About a dozen mutations in the *ACSF3* gene have been found in people with combined malonic and methylmalonic aciduria (CMAMMA), a condition characterized by elevated levels of chemicals known as malonic acid and methylmalonic acid in the body. This condition can cause development and growth problems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

## Chromosomal Location

Cytogenetic Location: 16q24.3, which is the long (q) arm of chromosome 16 at position 24.3

Molecular Location: base pairs 89,093,809 to 89,160,556 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## 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

### Genetic Testing Registry

- GTR: Genetic tests for ACSF3  
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=197322%5Bgeneid%5D>

### Scientific Articles on PubMed

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

### OMIM

- ACYL-CoA SYNTHETASE FAMILY, MEMBER 3  
<http://omim.org/entry/614245>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ACSF3.html](http://atlasgeneticsoncology.org/Genes/GC_ACSF3.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ACSF3%5Bgene%5D>
- HGNC Gene Family: Acyl-CoA synthetase family  
<http://www.genenames.org/cgi-bin/genefamilies/set/40>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=27288](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=27288)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/197322>
- UniProt  
<http://www.uniprot.org/uniprot/Q4G176>

## **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/ACSF3>

Reviewed: January 2013  
Published: February 14, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services