ACADVL gene

acyl-CoA dehydrogenase very long chain

Normal Function

The *ACADVL* gene provides instructions for making an enzyme called very long-chain acyl-CoA dehydrogenase (VLCAD). This enzyme functions within mitochondria, the energy-producing centers in cells. Very long-chain acyl-CoA dehydrogenase is essential for fatty acid oxidation, which is the multistep process that breaks down (metabolizes) fats and converts them to energy.

Very long-chain acyl-CoA dehydrogenase is required to metabolize a group of fats called very long-chain fatty acids. These fatty acids are found in food and body fat. Fatty acids are a major source of energy for the heart and muscles. During periods without food (fasting), fatty acids are also an important energy source for the liver and other tissues.

Health Conditions Related to Genetic Changes

Very long-chain acyl-CoA dehydrogenase deficiency

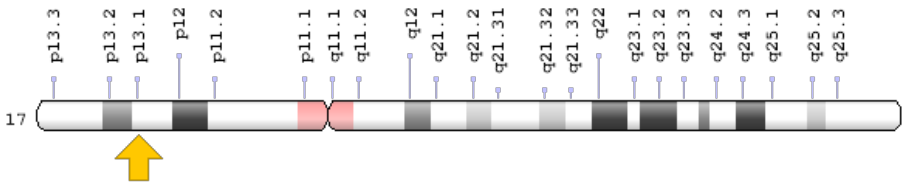
More than 100 mutations in the *ACADVL* gene have been found to cause very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. Many of these mutations change single protein building blocks (amino acids) in the VLCAD enzyme. Other mutations delete part of the *ACADVL* gene or create a premature stop signal in the instructions for making VLCAD. These mutations lead to a change in the enzyme's structure, severely reducing or eliminating its activity. As a result, very little functional enzyme is produced.

With a shortage (deficiency) of functional VLCAD enzyme, very-long chain fatty acids are not metabolized properly. As a result, these fats are not converted to energy, which can lead to signs and symptoms of this disorder such as the lack of energy (lethargy) and low blood sugar (hypoglycemia). Very long-chain fatty acids or partially metabolized fatty acids may build up in tissues and damage the heart, liver, and muscles. This abnormal buildup causes the other signs and symptoms of VLCAD deficiency.

Chromosomal Location

Cytogenetic Location: 17p13.1, which is the short (p) arm of chromosome 17 at position 13.1

Molecular Location: base pairs 7,217,125 to 7,225,267 on chromosome 17 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ACAD6
- ACADV_HUMAN
- acyl-CoA dehydrogenase, very long chain
- acyl-coenzyme A dehydrogenase, very long chain
- LCACD
- VLCAD

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Utilization of Fatty Acids as Fuel Requires Three Stages of Processing
  https://www.ncbi.nlm.nih.gov/books/NBK22581/

Clinical Information from GeneReviews

- Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK6816

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ACADVL%5BTIAB%5D%29+OR+%28%28LCACD%5BTIAB%5D%29+OR+%28VLCAD%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+2520+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN
  http://omim.org/entry/609575

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ACADVL.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ACADVL%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:92
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:37
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/37
- UniProt
  https://www.uniprot.org/uniprot/P49748

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