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 break down 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

Variants (also known as mutations) in the *ACADVL* gene have been found to cause very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. Many of these variants change single protein building blocks (amino acids) in the VLCAD enzyme. Other variants delete part of the *ACADVL* gene or create a premature stop signal in the instructions for making VLCAD. These variants 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 broken down 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 glucose (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.

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

Tests Listed in the Genetic Testing Registry

- Tests of ACADVL (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=37[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; ACADVL (https://omim.org/entry/609575)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/37)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACADVL[gene])

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Genomic Location

The *ACADVL* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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