

ACADS gene

acyl-CoA dehydrogenase short chain

Normal Function

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

SCAD is required to metabolize a group of fats called short-chain fatty acids. These fatty acids are found in some foods and are also produced when larger fatty acids are metabolized. 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

Short-chain acyl-CoA dehydrogenase deficiency

More than 55 mutations in the *ACADS* gene have been found to cause short-chain acyl-CoA dehydrogenase (SCAD) deficiency. Almost all of these mutations change single protein building blocks (amino acids) in the SCAD enzyme. These mutations prevent the enzyme from properly metabolizing short-chain fatty acids. As a result, these fats are not converted into energy, which can lead to the characteristic signs and symptoms of this disorder, including lack of energy (lethargy), low blood glucose (hypoglycemia), poor muscle tone (hypotonia), and weakness.

Researchers have also identified two common variations (polymorphisms) in the *ACADS* gene that each change one amino acid in the SCAD enzyme. Unlike other changes in the *ACADS* gene, these polymorphisms do not cause SCAD deficiency but may increase a person's risk of developing this disorder. One of these polymorphisms replaces the amino acid arginine with the amino acid tryptophan at protein position 147 (written as Arg147Trp or R147W). The other polymorphism switches the amino acid glycine with the amino acid serine at protein position 185 (written as Gly185Ser or G185S). Other genetic and environmental factors likely influence the risk of developing SCAD deficiency when a person carries either of these polymorphisms.

Other Names for This Gene

- ACAD3
- ACADS_HUMAN
- acyl-CoA dehydrogenase, C-2 to C-3 short chain
- acyl-Coenzyme A dehydrogenase, C-2 to C-3 short chain precursor
- Butyryl dehydrogenase
- Butyryl-CoA dehydrogenase
- SCAD
- Unsaturated acyl-CoA reductase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACADS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=35\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=35[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ACADS%5BTIAB%5D%29+OR+%28short-chain+acyl-CoA+dehydrogenase%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- ACYL-CoA DEHYDROGENASE, SHORT-CHAIN; ACADS (<https://omim.org/entry/606885>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/35>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ACADS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ACADS[gene]))

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Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14568186>)

Genomic Location

The *ACADS* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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