



ACADSB gene

acyl-CoA dehydrogenase, short/branched chain

Normal Function

The *ACADSB* gene provides instructions for making an enzyme called 2-methylbutyryl-CoA dehydrogenase, which plays an important role in processing proteins. Normally, the body breaks down proteins from food into smaller parts called amino acids. Amino acids can be further processed to provide energy for the body. In cells throughout the body, 2-methylbutyryl-CoA dehydrogenase is found within specialized structures called mitochondria. Mitochondria convert energy from food to a form that cells can use.

The 2-methylbutyryl-CoA dehydrogenase enzyme helps break down a particular amino acid called isoleucine. Specifically, this enzyme helps with the third step of the process, performing a chemical reaction that converts a molecule called 2-methylbutyryl-CoA to another molecule, tiglyl-CoA. Additional chemical reactions convert tiglyl-CoA into molecules that are used for energy.

Health Conditions Related to Genetic Changes

2-methylbutyryl-CoA dehydrogenase deficiency

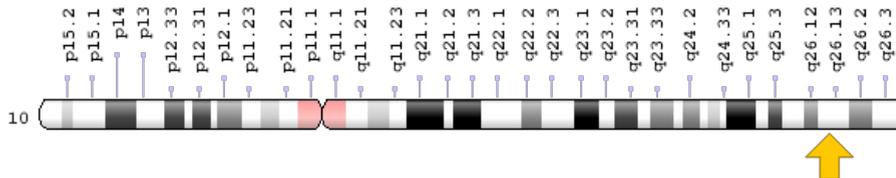
Researchers have identified more than 10 *ACADSB* gene mutations in people with 2-methylbutyryl-CoA dehydrogenase deficiency. While most people with this condition have no related health problems, some have reduced energy (lethargy), muscle weakness, seizures, developmental delays, or other health problems.

Many of the *ACADSB* gene mutations replace one of the amino acids in the 2-methylbutyryl-CoA dehydrogenase enzyme with an incorrect amino acid. Other mutations lead to an abnormally small version of this enzyme that is missing several amino acids. As a result of these mutations, 2-methylbutyryl-CoA dehydrogenase has little or no activity. With a shortage (deficiency) of normal enzyme activity, the body is unable to break down isoleucine properly. Researchers speculate that some features of this disorder, such as lethargy and muscle weakness, occur because isoleucine is not converted to energy. In addition, impairment of 2-methylbutyryl-CoA dehydrogenase may allow the buildup of toxic compounds, which can lead to serious health problems.

Chromosomal Location

Cytogenetic Location: 10q26.13, which is the long (q) arm of chromosome 10 at position 26.13

Molecular Location: base pairs 123,008,913 to 123,058,290 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 2-MEBCAD
- 2-methyl branched chain acyl-CoA dehydrogenase
- ACAD7
- ACDSB_HUMAN
- SBCAD
- short/branched chain acyl-CoA dehydrogenase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Proteins are Built from a Repertoire of 20 Amino Acids
<https://www.ncbi.nlm.nih.gov/books/NBK22379/>
- Structural Genomics Consortium
<http://www.thesgc.org/structures/details?pdbid=2JIF>

Genetic Testing Registry

- GTR: Genetic tests for ACADSB
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=36%5Bgeneid%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ACADSB%5BALL%5D%29+OR+%28%282-methyl+branched+chain+acyl-CoA+dehydrogenase%5BTIAB%5D%29+OR+%28SBCAD%5BALL%5D%29+OR+%28short/branched+chain+acyl-CoA+dehydrogenase%5BTIAB%5D%29+OR+%282-methylbutyryl-CoA+dehydrogenase%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22Iast+3600+days%22%5Bdp%5D>

OMIM

- ACYL-CoA DEHYDROGENASE, SHORT/BRANCHED CHAIN
<http://omim.org/entry/600301>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ACADSB%5Bgene%5D>
- HGNC Gene Family: Acyl-CoA dehydrogenase family
<http://www.genenames.org/cgi-bin/genefamilies/set/974>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=91
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/36>
- UniProt
<http://www.uniprot.org/uniprot/P45954>

Sources for This Summary

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