ACADSB gene

acyl-CoA dehydrogenase short/branched chain

Normal Function

The *ACADSB* gene provides instructions for making an enzyme called short/branched chain acyl-CoA dehydrogenase (SBCAD, also known as 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, SBCAD is found within specialized structures called mitochondria. Mitochondria convert energy from food into a form that cells can use.

The SBCAD 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. Through similar chemical reactions, the SBCAD enzyme also aids in the breakdown of other amino acids.

Health Conditions Related to Genetic Changes

Short/branched chain acyl-CoA dehydrogenase deficiency

Researchers have identified more than 10 *ACADSB* gene mutations in people with short/branched chain acyl-CoA dehydrogenase (SBCAD) 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 SBCAD 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, SBCAD 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 SBCAD may allow the buildup of toxic compounds, which can lead to serious health problems.

Other Names for This Gene

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACADSB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=36[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ACYL-CoA DEHYDROGENASE, SHORT/BRANCHED CHAIN; ACADSB (https://omim.org/entry/600301)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/36)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACADSB[gene])

References

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Genomic Location

The *ACADSB* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromosome/10/).

Last updated February 1, 2017