Short-chain acyl-CoA dehydrogenase deficiency

Description

Short-chain acyl-CoA dehydrogenase (SCAD) deficiency is a condition that prevents the body from converting certain fats into energy, especially during periods without food (fasting).

Signs and symptoms of SCAD deficiency may appear during infancy or early childhood and can include vomiting, low blood glucose (hypoglycemia), a lack of energy (lethargy), poor feeding, and failure to gain weight and grow at the expected rate (failure to thrive). Other features of this disorder may include poor muscle tone (hypotonia), seizures, developmental delay, and a small head size (microcephaly).

The symptoms of SCAD deficiency may be triggered by fasting or illnesses such as viral infections. This disorder is sometimes mistaken for Reye syndrome, a severe condition that may develop in children while they appear to be recovering from viral infections such as chicken pox or flu. Most cases of Reye syndrome are associated with the use of aspirin during these viral infections.

In some people with SCAD deficiency, signs and symptoms do not appear until adulthood. These individuals are more likely to have problems related to muscle weakness and wasting.

The severity of this condition varies widely, even among members of the same family. Some individuals are diagnosed with SCAD deficiency based on laboratory testing but never develop any symptoms of the condition.

Frequency

This disorder is thought to affect approximately 1 in 35,000 to 50,000 newborns.

Causes

Mutations in the *ACADS* gene cause SCAD deficiency. This gene provides instructions for making an enzyme called short-chain acyl-CoA dehydrogenase, which is required to break down (metabolize) a group of fats called short-chain fatty acids. Fatty acids are a major source of energy for the heart and muscles. During periods of fasting, fatty acids are also an important energy source for the liver and other tissues.

Mutations in the *ACADS* gene lead to a shortage (deficiency) of the SCAD enzyme within cells. Without sufficient amounts of this enzyme, short-chain fatty acids are not metabolized properly. As a result, these fats are not converted into energy, which can lead to the signs and symptoms of this disorder, such as lethargy, hypoglycemia, and muscle weakness. It remains unclear why some people with SCAD deficiency never develop any symptoms.

Learn more about the gene associated with Short-chain acyl-CoA dehydrogenase deficiency

- ACADS

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- ACADS deficiency
- Deficiency of butyryl-CoA dehydrogenase
- Lipid-storage myopathy secondary to short-chain acyl-coa dehydrogenase deficiency
- SCAD deficiency
- SCADH deficiency
- Short-chain acyl-coenzyme A dehydrogenase deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Deficiency of butyryl-CoA dehydrogenase (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342783/)

Genetic and Rare Diseases Information Center

- Short chain acyl-CoA dehydrogenase deficiency (https://rarediseases.info.nih.gov/diseases/4822/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- ACYL-CoA DEHYDROGENASE, SHORT-CHAIN, DEFICIENCY OF; ACADSD (https://omim.org/entry/201470)

Scientific Articles on PubMed

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

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