Holocarboxylase synthetase deficiency

Description

Holocarboxylase synthetase deficiency is an inherited disorder in which the body is unable to use the vitamin biotin effectively. This disorder is classified as a multiple carboxylase deficiency, which is a group of disorders characterized by impaired activity of certain enzymes that depend on biotin.

The signs and symptoms of holocarboxylase synthetase deficiency typically appear within the first few months of life, but the age of onset varies. Affected infants often have difficulty feeding, breathing problems, a skin rash, hair loss (alopecia), and a lack of energy (lethargy). Immediate treatment and lifelong management with biotin supplements may prevent many of these complications. If left untreated, the disorder can lead to delayed development, seizures, and coma. These medical problems may be life-threatening in some cases.

Frequency

The exact incidence of this condition is unknown, but it is estimated to affect 1 in 87,000 people.

Causes

Mutations in the *HLCS* gene cause holocarboxylase synthetase deficiency. The *HLCS* gene provides instructions for making an enzyme called holocarboxylase synthetase. This enzyme is important for the effective use of biotin, a B vitamin found in foods such as liver, egg yolks, and milk. Holocarboxylase synthetase attaches biotin to certain enzymes that are essential for the normal production and breakdown of proteins, fats, and carbohydrates in the body. Mutations in the *HLCS* gene reduce the enzyme's ability to attach biotin to these enzymes, preventing them from processing nutrients properly and disrupting many cellular functions. These defects lead to the serious medical problems associated with holocarboxylase synthetase deficiency.

Learn more about the gene associated with Holocarboxylase synthetase deficiency

- HLCS

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

- Biotin-(propionyl-CoA-carboxylase) ligase deficiency
- Biotin-(propionyl-coenzyme A-carboxylase) ligase deficiency
- Early-onset biotin-responsive multiple carboxylase deficiency
- Early-onset combined carboxylase deficiency
- HLCS deficiency
- Infantile multiple carboxylase deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Holocarboxylase synthetase deficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268581/)

Genetic and Rare Diseases Information Center

- Holocarboxylase synthetase deficiency (https://rarediseases.info.nih.gov/diseases/2721/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Holocarboxylase synthetase deficiency%22)

Catalog of Genes and Diseases from OMIM

- HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (https://omim.org/entry/253270)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28holocarboxylase+synthetase+deficiency%5BALL%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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