Hyperlysinemia

Description

Hyperlysinemia is an inherited condition characterized by elevated blood levels of the amino acid lysine, a building block of most proteins. Hyperlysinemia is caused by the shortage (deficiency) of the enzyme that breaks down lysine. Hyperlysinemia typically causes no health problems, and most people with elevated lysine levels are unaware that they have this condition. Rarely, people with hyperlysinemia have intellectual disability or behavioral problems. It is not clear whether these problems are due to hyperlysinemia or another cause.

Frequency

The incidence of hyperlysinemia is unknown.

Causes

Mutations in the *AASS* gene cause hyperlysinemia. The *AASS* gene provides instructions for making an enzyme called aminoadipic semialdehyde synthase. This enzyme performs two functions in the breakdown of lysine. First, the enzyme breaks down lysine to a molecule called saccharopine. It then breaks down saccharopine to a molecule called alpha-aminoadipate semialdehyde.

Mutations in the *AASS* gene that impair the breakdown of lysine result in elevated levels of lysine in the blood and urine. These increased levels of lysine do not appear to have any negative effects on the body.

When mutations in the *AASS* gene impair the breakdown of saccharopine, this molecule builds up in blood and urine. This buildup is sometimes referred to as saccharopinuria, which is considered to be a variant of hyperlysinemia. It is unclear if saccharopinuria causes any symptoms.

Learn more about the gene associated with Hyperlysinemia

- AASS

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

- Alpha-aminoadipic semialdehyde deficiency disease
- Familial hyperlysinemia
- Lysine alpha-ketoglutarate reductase deficiency disease
- Saccharopine dehydrogenase deficiency disease
- Saccharopinuria

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hyperlysinemia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268553/)
- Genetic Testing Registry: Saccharopinuria (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268556/)

Genetic and Rare Diseases Information Center

- Hyperlysinemia (https://rarediseases.info.nih.gov/diseases/2828/index)
- Saccharopinuria (https://rarediseases.info.nih.gov/diseases/314/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- SACCHAROPINURIA (https://omim.org/entry/268700)
- HYPERLYSINEMIA, TYPE I (https://omim.org/entry/238700)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28hyperlysinemia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+day s%22%5Bdp%5D)

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