Hyperlysinemia

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.

**Inheritance Pattern**

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

**Diagnosis & Management**

**Genetic Testing Information**

• What is genetic testing?  
  /primer/testing/genetictesting

• Genetic Testing Registry: Hyperlysinemia  

• Genetic Testing Registry: Saccharopinuria  

**Additional Information & Resources**

**Health Information from MedlinePlus**

• Health Topic: Amino Acid Metabolism Disorders  
  https://medlineplus.gov/aminoacidmetabolismdisorders.html

• Health Topic: Newborn Screening  
  https://medlineplus.gov/newbornscreening.html

**Genetic and Rare Diseases Information Center**

• Hyperlysinemia  
  https://rarediseases.info.nih.gov/diseases/2828/hyperlysinemia

• Saccharopinuria  
  https://rarediseases.info.nih.gov/diseases/314/saccharopinuria

**Educational Resources**

• MalaCards: hyperlysinemia, type i  
  http://www.malacards.org/card/hyperlysinemia_type_i

• MalaCards: saccharopinuria  
  http://www.malacards.org/card/saccharopinuria

• Orphanet: Hyperlysinemia  
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2203

• Orphanet: Saccharopinuria  
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3124
Patient Support and Advocacy Resources

- Metabolic Support UK
  https://www.metabolicsupportuk.org/
- University of Kansas Medical Center Resource List: Metabolic Conditions
  http://www.kumc.edu/gec/support/metaboli.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28hyperlysinemia%5BTIAB%5D%29+AND+english%5BLa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- HYPERLYSINEMIA, TYPE I
  http://omim.org/entry/238700
- SACCHAROPINURIA
  http://omim.org/entry/268700

Medical Genetics Database from MedGen

- Hyperlysinemia
- Saccharopinuria

Sources for This Summary

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