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
  https://www.malacards.org/card/hyperlysinemia_type_i
• MalaCards: saccharopinuria
  https://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
  %5D%29+AND+english%5BIa%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

• Markovitz PJ, Chuang DT, Cox RP. Familial hyperlysinemias. Purification and characterization
  of the bifunctional aminoadipic semialdehyde synthase with lysine-ketoglutarate reductase and
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/6434529

• Sacksteder KA, Biery BJ, Morrell JC, Goodman BK, Geisbrecht BV, Cox RP, Gould SJ, Geraghty
  MT. Identification of the alpha-aminoadipic semialdehyde synthase gene, which is defective in
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10775527
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1378037/

• Saudubray JM, Rabier D. Biomarkers identified in inborn errors for lysine, arginine, and ornithine. J
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17513445

Reprinted from Genetics Home Reference: