Cystinuria

Cystinuria is a condition characterized by the buildup of the amino acid cystine, a building block of most proteins, in the kidneys and bladder. As the kidneys filter blood to create urine, cystine is normally absorbed back into the bloodstream. People with cystinuria cannot properly reabsorb cystine into their bloodstream, so the amino acid accumulates in their urine.

As urine becomes more concentrated in the kidneys, the excess cystine forms crystals. Larger crystals become stones that may lodge in the kidneys or in the bladder. Sometimes cystine crystals combine with calcium molecules in the kidneys to form large stones. These crystals and stones can create blockages in the urinary tract and reduce the ability of the kidneys to eliminate waste through urine. The stones also provide sites where bacteria may cause infections.

Frequency

Cystinuria affects approximately 1 in 10,000 people.

Causes

Mutations in the \( SLC3A1 \) or \( SLC7A9 \) gene cause cystinuria. The \( SLC3A1 \) and \( SLC7A9 \) genes provide instructions for making the two parts (subunits) of a protein complex that is primarily found in the kidneys. Normally this protein complex controls the reabsorption of certain amino acids, including cystine, into the blood from the filtered fluid that will become urine. Mutations in either the \( SLC3A1 \) gene or \( SLC7A9 \) gene disrupt the ability of the protein complex to reabsorb amino acids, which causes the amino acids to become concentrated in the urine. As the levels of cystine in the urine increase, the crystals typical of cystinuria form. The other amino acids that are reabsorbed by the protein complex do not create crystals when they accumulate in the urine.

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

- CSNU
Diagnosis & Management

Genetic Testing Information

- What is genetic testing? https://primer/testing/genetictesting

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov https://clinicaltrials.gov/ct2/results?cond=%22cystinuria%22

Other Diagnosis and Management Resources


Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Cystinuria (image) https://medlineplus.gov/ency/imagepages/17046.htm

Genetic and Rare Diseases Information Center

- Cystinuria https://rarediseases.info.nih.gov/diseases/6237/cystinuria

Additional NIH Resources

Educational Resources

• MalaCards: cystinuria
  https://www.malacards.org/card/cystinuria

• Merck Manual Consumer Version
  https://www.merckmanuals.com/home/children-s-health-issues/congenital-kidney-
tubular-disorders/cystinuria

• Orphanet: Cystinuria
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=214

Patient Support and Advocacy Resources

• Metabolic Support UK
  https://www.metabolicsupportuk.org/

• National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/cystinuria/

• University of Kansas Medical Center Resource List
  http://www.kumc.edu/gec/support/cystinur.html

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Cystinuria%5BMAJR%5D%29+AND+%28cystinuria%5BTIAB%5D%29+NOT+%28gene%5BTIAB%5D%29+AND+
  english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days
  %22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

• CYSTINURIA
  http://omim.org/entry/220100

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22281707

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15602663

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15021200

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