SLC7A9 gene
solute carrier family 7 member 9

Normal Function

The SLC7A9 gene provides instructions for producing one part (subunit) of a protein made primarily in the kidneys. This subunit joins with another protein subunit, produced from the SLC3A1 gene, to form a transporter protein complex. During the process of urine formation in the kidneys, this protein complex absorbs particular protein building blocks (amino acids) back into the blood. In particular, the amino acids cystine, ornithine, arginine, and lysine are absorbed back into the blood through this mechanism.

Health Conditions Related to Genetic Changes

Cystinuria

At least 95 mutations in the SLC7A9 gene have been found to cause cystinuria. Many of these mutations alter a single DNA building block (nucleotide) or insert or delete a small number of nucleotides in the SLC7A9 gene. These changes lead to an abnormally functioning transporter protein complex, which causes certain amino acids to become concentrated in the urine. Cystine is the only amino acid that forms crystals and stones in the bladder or kidneys, leading to the signs and symptoms of cystinuria.

Chromosomal Location

Cytogenetic Location: 19q13.11, which is the long (q) arm of chromosome 19 at position 13.11

Molecular Location: base pairs 32,830,511 to 32,870,957 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)
Other Names for This Gene

• b0,+AT
• BAT1_HUMAN
• CSNU3
• solute carrier family 7 (amino acid transporter light chain, bo,+ system), member 9
• solute carrier family 7 (glycoprotein-associated amino acid transporter light chain, bo,+ system), member 9
• solute carrier family 7, member 9

Additional Information & Resources

Educational Resources

• National Institute of Diabetes and Digestive and Kidney Diseases: Your Kidneys and How They Work
  https://www.niddk.nih.gov/health-information/kidney-disease/kidneys-how-they-work

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC7A9%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D %29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last +1800+days%22+AND+human%5Bmh%5D+AND+%22last+1800+days%22

Catalog of Genes and Diseases from OMIM

• SOLUTE CARRIER FAMILY 7, MEMBER 9
  http://omim.org/entry/604144

Research Resources

• Atlas of Genetics and Cytogenetics in Oncology and Haematology
• ClinVar
  https://www.ncbi.nlm.nih.govclinvar?term=SLC7A9%5Bgene%5D
• HGNC Gene Symbol Report
• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:11136
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12239244

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12167606

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11157794

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15635077
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735913/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15499206

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

- OMIM: SOLUTE CARRIER FAMILY 7, MEMBER 9
  http://omim.org/entry/604144

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14531788

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