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.

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

Tests Listed in the Genetic Testing Registry

- Tests of SLC7A9 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=11136[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 7 (CATIONIC AMINO ACID TRANSPORTER, y+ SYSTEM), MEMBER 9; SLC7A9 (https://omim.org/entry/604144)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/11136)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC7A9[gene])

References

- Dello Strologo L, Pras E, Pontesilli C, Beccia E, Ricci-Barbini V, de SanctisL, Ponzone A, Gallucci M, Bisceglia L, Zelante L, Jimenez-Vidal M, Font M,Zorzano A, Rousaud F, Nunes V, Gasparini P, Palacin M, Rizzoni G. Comparisonbetween SLC3A1 and SLC7A9 cystinuria patients and carriers: a need for a newclassification. J Am Soc Nephrol. 2002 Oct;13(10):2547-53. doi:10.1097/01.asn.0000029586.17680.e5. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12239244)
- Fernandez E, Carrascal M, Rousaud F, Abian J, Zorzano A, Palacin M, ChillaronJ. rBAT-b(0,+)AT heterodimer is the main apical reabsorption system for cystinein the kidney. Am J Physiol Renal Physiol. 2002 Sep;283(3):F540-8. doi:10.1152/ajprenal.00071.2002. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12167606)
- Font MA, Feliubadalo L, Estivill X, Nunes V, Golomb E, Kreiss Y, Pras E,Bisceglia L, d'Adamo AP, Zelante L, Gasparini P, Bassi MT, George AL Jr, ManzoniM, Riboni M, Ballabio A, Borsani G, Reig N, Fernandez E, Zorzano A, Bertran J,Palacin M; International Cystinuria Consortium. Functional analysis of mutationsin SLC7A9, and genotype-phenotype correlation in non-Type I cystinuria. Hum MolGenet. 2001 Feb 15;10(4):305-16. doi: 10.1093/hmg/10.4.305. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11157794)
- Font-Llitjos M, Jimenez-Vidal M, Bisceglia L, Di Perna M, de Sanctis L,Rousaud F, Zelante L, Palacin M, Nunes V. New insights into cystinuria: 40 newmutations, genotype-phenotype correlation, and digenic inheritance causingpartial phenotype. J Med Genet. 2005 Jan;42(1):58-68. doi:10.1136/jmg.2004.022244. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15635077) or Free article on PubMed

Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735913/)

- Goodyer P. The molecular basis of cystinuria. Nephron Exp Nephrol.2004;98(2):e45-9. doi: 10.1159/000080255. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15499206)
- Langman CB. The molecular basis of kidney stones. Curr Opin Pediatr. 2004Apr;16(2):188-93. doi: 10.1097/00008480-200404000-00013. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15021200)
- Schmidt C, Vester U, Wagner CA, Lahme S, Hesse A, Hoyer P, Lang F, Zerres K, Eggermann T; Arbeitsgemeinschaft fur Padiatrische Nephrologie. Significantcontribution of genomic rearrangements in SLC3A1 and SLC7A9 to the etiology ofcystinuria. Kidney Int. 2003 Nov;64(5):1564-72. doi:10.1046/j.1523-1755.2003.00250.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14531788)

Genomic Location

The *SLC7A9* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

Last updated January 1, 2009