

CTNS gene

cystinosis, lysosomal cystine transporter

Normal Function

The *CTNS* gene provides instructions for making a protein called cystinosis. This protein is located in the membrane of lysosomes, which are compartments in the cell that digest and recycle materials. Proteins digested inside lysosomes are broken down into smaller building blocks, called amino acids. The amino acids are then moved out of lysosomes by transport proteins. Cystinosis is a transport protein that specifically moves the amino acid cystine out of the lysosome.

Health Conditions Related to Genetic Changes

Cystinosis

More than 80 different mutations that are responsible for causing cystinosis have been identified in the *CTNS* gene. The most common mutation is a deletion of a large part of the *CTNS* gene (sometimes referred to as the 57-kb deletion), resulting in the complete loss of cystinosis. This deletion is responsible for approximately 50 percent of cystinosis cases in people of European descent. Other mutations result in the production of an abnormally short protein that cannot carry out its normal transport function. Mutations that change very small regions of the *CTNS* gene may allow the transporter protein to retain some of its usual activity, resulting in a milder form of cystinosis.

Other Names for This Gene

- CTNS-LSB
- CTNS_HUMAN
- Cystinosis
- PQLC4

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CTNS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1497\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1497[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28CTNS%5BTIAB%5D%29+OR+%28Cystinosis%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%22%29>)

Catalog of Genes and Diseases from OMIM

- CYSTINOSIN; CTNS (<https://omim.org/entry/606272>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1497>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CTNS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CTNS[gene]))

References

- Anikster Y, Shotelersuk V, Gahl WA. CTNS mutations in patients withcystinosis. Hum Mutat. 1999;14(6):454-8. doi:10.1002/(SICI)1098-1004(199912)14:63.O.CO;2-H. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10571941>)
- Bendavid C, Kleta R, Long R, Ouspenskaia M, Muenke M, Haddad BR, Gahl WA. FISHdiagnosis of the common 57-kb deletion in CTNS causing cystinosis. Hum Genet.2004 Nov;115(6):510-4. doi: 10.1007/s00439-004-1170-2. Epub 2004 Sep 9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15365816>)
- Haq MR, Kalatzis V, Gubler MC, Town MM, Antignac C, Van't Hoff WG, Woolf AS.Immunolocalization of cystinosin, the protein defective in cystinosis. J Am SocNephrol. 2002 Aug;13(8):2046-51. doi: 10.1097/01.asn.0000022012.95680.ba. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12138135>)
- Kalatzis V, Antignac C. Cystinosis: from gene to disease. Nephrol DialTransplant. 2002 Nov;17(11):1883-6. doi: 10.1093/ndt/17.11.1883. No abstractavailable. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12401840>)
- Kalatzis V, Nevo N, Cherqui S, Gasnier B, Antignac C. Molecular pathogenesisof cystinosis: effect of CTNS mutations on the transport activity and subcellularlocalization of cystinosin. Hum Mol Genet. 2004 Jul 1;13(13):1361-71. doi: 10.1093/hmg/ddh152. Epub 2004 May 5. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15128704>)
- Servais A, Moriniere V, Grunfeld JP, Noel LH, Goujon JM, Chadeaux-Vekemans B, Antignac C. Late-onset nephropathic cystinosis: clinical presentation, outcome,and genotyping. Clin J Am Soc Nephrol. 2008 Jan;3(1):27-35. doi:10.2215/CJN.01740407. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18178779>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2390982/>)

- Touchman JW, Anikster Y, Dietrich NL, Maduro VV, McDowell G, Shotelersuk V, Bouffard GG, Beckstrom-Sternberg SM, Gahl WA, Green ED. The genomic region encompassing the nephropathic cystinosis gene (CTNS): complete sequencing of a 200-kb segment and discovery of a novel gene within the common cystinosis-causing deletion. *Genome Res.* 2000 Feb;10(2):165-73. doi: 10.1101/gr.10.2.165. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10673275>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC310836/>)
- Town M, Jean G, Cherqui S, Attard M, Forestier L, Whitmore SA, Callen DF, Gribouval O, Broyer M, Bates GP, van't Hoff W, Antignac C. A novel gene encoding an integral membrane protein is mutated in nephropathic cystinosis. *Nat Genet.* 1998 Apr;18(4):319-24. doi: 10.1038/ng0498-319. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9537412>)
- Wamelink MM, Struys EA, Jansen EE, Levtchenko EN, Zijlstra FS, Engelke U, Blom HJ, Jakobs C, Wevers RA. Sedoheptulokinase deficiency due to a 57-kb deletion in cystinosis patients causes urinary accumulation of sedoheptulose: elucidation of the CARKL gene. *Hum Mutat.* 2008 Apr;29(4):532-6. doi: 10.1002/humu.20685. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18186520>)

Genomic Location

The *CTNS* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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