SLC6A8 gene

solute carrier family 6 member 8

Normal Function

The *SLC6A8* gene provides instructions for making a protein called sodium- and chloride-dependent creatine transporter 1. This protein transports the compound creatine into cells. Creatine is needed for the body to store and use energy properly.

Health Conditions Related to Genetic Changes

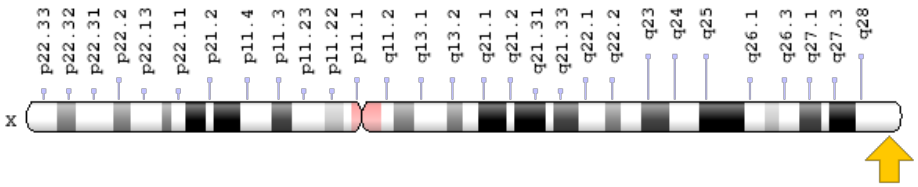
X-linked creatine deficiency

At least 80 mutations in the *SLC6A8* gene have been identified in people with X-linked creatine deficiency, a disorder that causes intellectual disability, behavioral problems, seizures, and muscle weakness. *SLC6A8* gene mutations impair the ability of the transporter protein to bring creatine into cells, resulting in a creatine shortage (deficiency). The effects of creatine deficiency are most severe in organs and tissues that require large amounts of energy, especially the brain.

Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 153,687,926 to 153,696,593 on the X chromosome (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- creatine transporter 1
- creatine transporter SLC6A8
- CRT

- CRTR
- CT1
- MGC87396
- SC6A8_HUMAN
- sodium- and chloride-dependent creatine transporter 1
- solute carrier family 6 (neurotransmitter transporter), member 8
- solute carrier family 6 (neurotransmitter transporter, creatine), member 8

Additional Information & Resources

Clinical Information from GeneReviews

- Creatine Deficiency Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK3794

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC6A8%5BTIAB%5D%29+OR+%28%28CRTR%5BTIAB%5D%29+OR+%28creatine+transporter+1%5BTIAB%5D%29+OR+%28creatine+transporter+SLC6A8%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, CREATINE), MEMBER 8
  http://omim.org/entry/300036

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC6A8%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11055
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6535
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6535
- UniProt
  https://www.uniprot.org/uniprot/P48029

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