SLC30A10 gene

solute carrier family 30 member 10

Normal Function

The *SLC30A10* gene provides instructions for making a protein that transports the element manganese across cell membranes. Manganese is important for many cellular functions, but large amounts are toxic, particularly to brain and liver cells. Excess amounts of the element are normally removed from the body through bile, which is a fluid produced in the liver that is important for digestion and the removal of waste materials.

The SLC30A10 protein is found in the membranes surrounding liver cells and nerve cells in the brain, as well as in the membranes of structures within these cells. It protects cells from high concentrations of manganese by removing manganese when levels become elevated. In the liver, the SLC30A10 protein transports manganese out of cells into bile so that the element can be removed from the body.

Health Conditions Related to Genetic Changes

Hypermanganesemia with dystonia

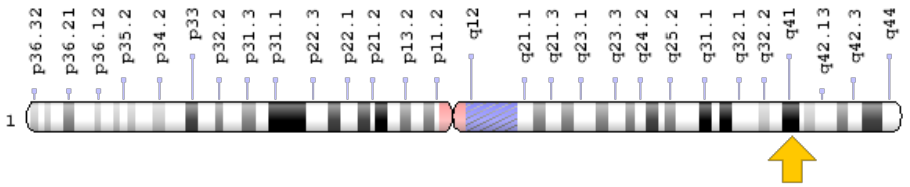
More than a dozen mutations in the *SLC30A10* gene have been identified in people with hypermanganesemia with dystonia, polycythemia, and cirrhosis (HMDPC, also known as hypermanganesemia with dystonia 1). This inherited disorder is characterized by high levels of manganese (hypermanganesemia) in the blood, brain, and liver. The disorder causes movement problems, such as involuntary tensing of the muscles (dystonia); an increased number of red blood cells (polycythemia); and liver abnormalities, including irreversible liver disease (cirrhosis).

Mutations in the *SLC30A10* gene impair the transport of manganese out of the cell, allowing the element to build up in brain and liver cells. Manganese accumulates in a region of the brain that helps control movement, damaging nerve cells and leading to the movement problems characteristic of HMDPC. Damage caused by a buildup of manganese in the liver causes the characteristic liver problems. High levels of manganese help increase the production of red blood cells, so excess amounts of this element may underlie polycythemia.

Chromosomal Location

Cytogenetic Location: 1q41, which is the long (q) arm of chromosome 1 at position 41

Molecular Location: base pairs 219,910,395 to 219,959,098 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DKFZp547M236
- HMDPC
- solute carrier family 30, member 10
- zinc transporter 10
- ZnT-10
- ZNT10
- ZNT10_HUMAN
- ZRC1

Additional Information & Resources

Educational Resources

- Agency for Toxic Substances and Disease Registry: Manganese
  https://www.atsdr.cdc.gov/substances/toxsubstance.asp?toxid=23
- Oregon State University Linus Pauling Institute: Manganese
  https://lpi.oregonstate.edu/mic/minerals/manganese

Clinical Information from GeneReviews

- Dystonia/Parkinsonism, Hypermanganesemia, Polycythemia, and Chronic Liver Disease
  https://www.ncbi.nlm.nih.gov/books/NBK100241

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC30A10%5BTIAB%5D%29+OR+%28%28ZRC1%5BTIAB%5D%29+OR+%28ZNT10%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+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 30 (ZINC TRANSPORTER), MEMBER 10
  http://omim.org/entry/611146

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC30A10.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC30A10%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:25355
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:55532
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/55532
- UniProt
  https://www.uniprot.org/uniprot/Q6XR72

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