SLC20A2 gene

solute carrier family 20 member 2

Normal Function

The *SLC20A2* gene provides instructions for making a protein called sodium-dependent phosphate transporter 2 (PiT-2). This protein is highly active in nerve cells (neurons) in the brain. It plays a major role in regulating the levels of a molecule called phosphate in cells (phosphate homeostasis). Specifically, the PiT-2 protein uses positively charged sodium atoms (ions) to transport phosphate in out and of cell membranes. Phosphate is needed for many cellular functions including the breakdown of substances (metabolic processes), signaling between cells, and the production of DNA building blocks (nucleic acids) and fats.

Health Conditions Related to Genetic Changes

Primary familial brain calcification

More than 60 *SLC20A2* gene mutations have been found to cause primary familial brain calcification. This condition is characterized by abnormal deposits of calcium (calcification) in the brain and movement and psychiatric problems. Most of the mutations that cause primary familial brain calcification change single protein building blocks (amino acids) in the PiT-2 protein and severely impair its ability to transport phosphate into cells. As a result, phosphate levels in the bloodstream rise. The excess phosphate combines with calcium and forms deposits within blood vessels in the brain.

Although the *SLC20A2* gene is active throughout the body, its activity is highest in structures deep within the brain that help start and control movement (basal ganglia) and in other brain regions that are involved in primary familial brain calcification, which may explain why the effects of these mutations are limited to these regions.

Other Names for This Gene

- gibbon ape leukemia virus receptor 2
- GLVR-2
- GLVR2
- MLVAR
- murine leukemia virus, amphotropic, receptor for

- PIT-2
- PIT2
- S20A2_HUMAN
- sodium-dependent phosphate transporter 2
- solute carrier family 20 (phosphate transporter), member 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC20A2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6575[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC20A2%5BTIAB%5D%29+OR+%28%28PIT-2%5BTIAB%5D%29+OR+%28PIT2%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 20 (PHOSPHATE TRANSPORTER), MEMBER 2; SLC20A2 (https://omim.org/entry/158378)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6575)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC20A2[gene])

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Genomic Location

The *SLC20A2* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

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