VPS13A gene
vacuolar protein sorting 13 homolog A

Normal Function

The VPS13A gene provides instructions for producing a protein called chorein. Chorein is found in various tissues throughout the body. The function of this protein is unknown. Some researchers believe that chorein plays a role in the movement of proteins within cells.

Health Conditions Related to Genetic Changes

Chorea-acanthocytosis

Approximately 75 mutations in the VPS13A gene have been found to cause chorea-acanthocytosis. Most of these mutations insert or delete one or more building blocks of DNA (nucleotides) in the VPS13A gene. These changes in the gene lead to the production of an abnormally small, nonfunctional version of chorein. Many of the mutations are specific to single families, although people with chorea-acanthocytosis who are French-Canadian or Japanese tend to have a mutation that is specific to their population. It is unclear why mutations in the VPS13A gene affect only the brain and red blood cells, causing the signs and symptoms of chorea-acanthocytosis.

Chromosomal Location

Cytogenetic Location: 9q21.2, which is the long (q) arm of chromosome 9 at position 21.2

Molecular Location: base pairs 77,177,353 to 77,421,537 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

- CHAC
- CHOREIN
- FLJ42030
- KIAA0986
- vacuolar protein sorting 13 homolog A (S. cerevisiae)
- vacuolar protein sorting 13A
- VP13A_HUMAN

Additional Information & Resources

GeneReviews
- Chorea-Acanthocytosis
  https://www.ncbi.nlm.nih.gov/books/NBK1387

Scientific Articles on PubMed
- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28VPS13A%5BTIAB%5D%29+OR+%28chorein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29%29+AND+english%5BLa%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D

OMIM
- VACUOLAR PROTEIN SORTING 13, YEAST, HOMOLOG OF, A
  http://omim.org/entry/605978

Research Resources
- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_VPS13A.html
- ClinVar
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1908
- NCBI Gene
- UniProt
  http://www.uniprot.org/uniprot/Q96RL7
Sources for This Summary


- OMIM: VACUOLAR PROTEIN SORTING 13, YEAST, HOMOLOG OF, A http://omim.org/entry/605978


Reviewed: May 2008
Published: March 27, 2018

Lister Hill National Center for Biomedical Communications
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