

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.

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

Tests Listed in the Genetic Testing Registry

- Tests of VPS13A ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23230\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23230[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28VPS13A%5BTIAB%5D%29+OR+%28chorein%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+2160+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- VACUOLAR PROTEIN SORTING 13 HOMOLOG A; VPS13A (<https://omim.org/entry/605978>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/23230>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=VPS13A\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=VPS13A[gene]))

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Genomic Location

The *VPS13A* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

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