

## 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>)

### 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]))

## **References**

- Dobson-Stone C, Danek A, Rampoldi L, Hardie RJ, Chalmers RM, Wood NW, Bohlega S, Dotti MT, Federico A, Shizuka M, Tanaka M, Watanabe M, Ikeda Y, Brin M, Goldfarb LG, Karp BI, Mohiddin S, Fananapazir L, Storch A, Fryer AE, Maddison P, Sibon I, Trevisol-Bittencourt PC, Singer C, Caballero IR, Aasly JO, Schmierer K, Dengler R, Hiersemenzel LP, Zeviani M, Meiner V, Lossos A, Johnson S, Mercado FC, Sorrentino G, Dupre N, Rouleau GA, Volkmann J, Arpa J, Lees A, Geraud G, Chouinard S, Nemeth A, Monaco AP. Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. *Eur J Hum Genet.* 2002 Nov;10(11):773-81. doi: 10.1038/sj.ejhg.5200866. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12404112>)
- Dobson-Stone C, Velayos-Baeza A, Jansen A, Andermann F, Dubeau F, Robert F, Summers A, Lang AE, Chouinard S, Danek A, Andermann E, Monaco AP. Identification of a VPS13A founder mutation in French Canadian families with chorea-acanthocytosis. *Neurogenetics.* 2005 Sep;6(3):151-8. doi:10.1007/s10048-005-0220-9. Epub 2005 Sep 28. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15918062>)
- Rampoldi L, Dobson-Stone C, Rubio JP, Danek A, Chalmers RM, Wood NW, Verellen C, Ferrer X, Malandrini A, Fabrizi GM, Brown R, Vance J, Pericak-Vance M, Rudolf G, Carre S, Alonso E, Manfredi M, Nemeth AH, Monaco AP. A conserved sorting-associated protein is mutant in chorea-acanthocytosis. *Nat Genet.* 2001 Jun;28(2):119-20. doi: 10.1038/88821. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11381253>)

- Ueno S, Maruki Y, Nakamura M, Tomemori Y, Kamae K, Tanabe H, Yamashita Y, Matsuda S, Kaneko S, Sano A. The gene encoding a newly discovered protein, chorein, is mutated in chorea-acanthocytosis. *Nat Genet.* 2001 Jun;28(2):121-2. doi: 10.1038/88825. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11381254>)
- Velayos-Baeza A, Vettori A, Copley RR, Dobson-Stone C, Monaco AP. Analysis of the human VPS13 gene family. *Genomics.* 2004 Sep;84(3):536-49. doi:10.1016/j.ygeno.2004.04.012. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15498460>)

## Genomic Location

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

**Last updated May 1, 2008**