VPS13B gene

vacuolar protein sorting 13 homolog B

Normal Function

Researchers are still working to determine the exact role of the *VPS13B* gene (also called the *COH1* gene) in the human body. Studies show that the protein produced from this gene is a part of the Golgi apparatus, which is a cell structure in which newly produced proteins are modified so they can carry out their functions. In particular, the VPS13B protein is involved in a modification called glycosylation, which is the attachment of sugar molecules to proteins. The VPS13B protein also appears to be involved in the sorting and transporting of proteins inside the cell.

Studies suggest several functions for the VPS13B protein in the body. The protein appears to play an important role in the normal growth and development of nerve cells (neurons). It may also be involved in the growth and development of adipocytes, which are cells that store fats for energy, and may play a role in the storage and distribution of fats in the body.

Health Conditions Related to Genetic Changes

Cohen syndrome

More than 150 different mutations in the *VPS13B* gene have been found in individuals with Cohen syndrome. Individuals with this condition typically have intellectual disability, eye problems, and distinctive facial features. Another common feature of the condition is obesity, particularly around the torso but not the arms and legs (truncal obesity). Most of the mutations that cause Cohen syndrome result in a premature stop signal in the instructions for making the VPS13B protein. Researchers believe that this genetic change leads to the production of an abnormally short, nonfunctional version of the protein. Studies suggest that a loss of this protein's function disrupts the organization of the Golgi apparatus and impairs normal glycosylation. However, it is not known how the absence of functional VPS13B protein or these cellular changes lead to the signs and symptoms of Cohen syndrome. Researchers speculate that problems with neuron development underlie intellectual disability, eye problems and other features of Cohen syndrome. Abnormal fat storage may cause truncal obesity in people with the condition.

In the Finnish population, 75 percent of individuals with Cohen syndrome have a mutation in both copies of the *VPS13B* gene that deletes two DNA building blocks (base

pairs). This mutation is sometimes written as 3348_3349delCT. The deletion causes a premature stop signal in the instructions for making the VPS13B protein.

Two common mutations occur in the Old Order Amish population. The first mutation adds one base pair and is sometimes written as 9258_9259insT. This mutation creates a premature stop signal in the instructions for making the VPS13B protein. The second mutation changes a single protein building block (amino acid) in the VPS13B protein. Specifically, this mutation replaces the amino acid isoleucine with the amino acid threonine at position 2820 (written as Ile2820Thr or I2820T). Outside the Finnish and Amish populations, nearly all mutations in the *VPS13B* gene occur in only one or a small number of families.

Other Names for This Gene

- CHS1
- COH1
- Cohen syndrome 1
- DKFZp313I0811
- KIAA0532
- vacuolar protein sorting 13 homolog B (yeast)
- vacuolar protein sorting 13B
- VP13B_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of VPS13B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=157680[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28VPS13B%5BTIAB%5D%29+OR+%28COH1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28streptococcus%29+OR+%28streptococcal%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- VACUOLAR PROTEIN SORTING 13 HOMOLOG B; VPS13B (https://omim.org/entry/607817)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/157680)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=VPS13B[gene])

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Genomic Location

The *VPS13B* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

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