MYO5B gene

myosin VB

Normal Function

The *MYO5B* gene provides instructions for making a protein called myosin Vb. This protein is one of a group of proteins with similar structures called myosins, which are involved in cell movement and the transport of materials within and between cells. Myosin Vb helps to determine the position of various components within cells (cell polarity). Myosin Vb also plays a role in moving components from the cell membrane to the interior of the cell for recycling.

Health Conditions Related to Genetic Changes

Microvillus inclusion disease

More than 40 mutations in the *MYO5B* gene have been found to cause microvillus inclusion disease. This condition is characterized by chronic, life-threatening diarrhea beginning in infancy. The *MYO5B* gene mutations that cause this condition result in a decrease or absence of myosin Vb function. In cells that line the small intestine (enterocytes), a lack of myosin Vb function changes the cell polarity. As a result, enterocytes cannot properly form structures called microvilli, which normally project like small fingers from the surface of the cells and absorb nutrients and fluids from food as it passes through the intestine. Inside affected enterocytes, small clumps of abnormal microvilli mix with misplaced digestive proteins to form microvillus inclusions, which contribute to the dysfunction of enterocytes. Disorganized enterocytes with poorly formed microvilli reduce the intestine's ability to take in nutrients and fluids. The inability to absorb nutrients and fluids during digestion leads to severe diarrhea, malnutrition, and dehydration in individuals with microvillus inclusion disease.

Other Names for This Gene

- KIAA1119
- MYO5B variant protein
- myosin-Vb
- unconventional myosin-Vb

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MYO5B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4645[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MYO5B%5BTIAB%5D%29+OR+%28myosin+VB%5BTIAB%5D%29%29+OR+%28myosin-Vb%5BTIAB%5D%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

- MYOSIN VB; MYO5B (https://omim.org/entry/606540)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4645)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MYO5B[gene])

References

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- Thoeni CE, Vogel GF, Tancevski I, Geley S, Lechner S, Pfaller K, Hess MW,Muller T, Janecke AR, Avitzur Y, Muise A, Cutz E, Huber LA. Microvillus inclusiondisease: loss of Myosin vb disrupts intracellular traffic and cell polarity.Traffic. 2014 Jan;15(1):22-42. doi: 10.1111/tra.12131. Epub 2013 Nov 19. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24138727)
- van der Velde KJ, Dhekne HS, Swertz MA, Sirigu S, Ropars V, Vinke PC, RengawT, van den Akker PC, Rings EH, Houdusse A, van Ijzendoorn SC. An overview andonline registry of microvillus inclusion disease patients and their MYO5Bmutations. Hum Mutat. 2013 Dec;34(12):1597-605. doi: 10.1002/humu.22440. Epub2013 Oct 16. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24014347)

Genomic Location

The *MYO5B* gene is found on chromosome 18 (https://medlineplus.gov/genetics/chromosome/18/).

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