



MYO5A gene

myosin VA

Normal Function

The *MYO5A* gene provides instructions for making a protein called myosin Va, which is part of a group of proteins called unconventional myosins. These proteins, which have similar structures, each play a role in transporting molecules within cells. Myosins interact with actin, a protein that is important for cell movement and shape. Researchers believe that myosins use long filaments of actin as tracks along which to transport other molecules.

Myosin Va is found in pigment-producing cells called melanocytes, where it helps transport structures called melanosomes. These structures produce a pigment called melanin, which is the substance that gives skin, hair, and eyes their color (pigmentation). Myosin Va interacts with proteins produced from the *MLPH* and *RAB27A* genes to form a complex that transports melanosomes to the outer edges of melanocytes. From there, the melanosomes are transferred to other types of cells, where they provide the pigment needed for normal hair, skin, and eye coloring.

Myosin Va also plays an important role in nerve cells (neurons) in the brain. Studies suggest that myosin Va transports various proteins and other molecules within neurons. It is also involved in the release of certain substances from these cells (exocytosis). The movement of these materials appears to be critical for normal brain function.

Health Conditions Related to Genetic Changes

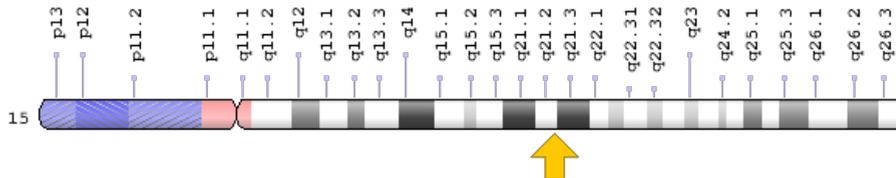
Griscelli syndrome

At least two mutations in the *MYO5A* gene have been found in people with Griscelli syndrome. These mutations cause a form of the condition designated type 1, which is characterized by unusually light (hypopigmented) skin, silvery-gray hair, and neurological abnormalities resulting in delayed development, intellectual disability, and seizures. The known *MYO5A* gene mutations prevent the production of functional myosin Va. Because the nonfunctional protein cannot form a complex with the proteins made from the *MLPH* and *RAB27A* genes, melanosomes cannot be transported to the edges of melanocytes. Instead, these structures clump near the center of melanocytes, trapping melanin within these cells and preventing normal pigmentation of skin and hair. A loss of myosin Va in neurons disrupts the transport of proteins and other molecules within and out of these cells, which likely causes the neurological problems found in Griscelli syndrome type 1.

Chromosomal Location

Cytogenetic Location: 15q21.2, which is the long (q) arm of chromosome 15 at position 21.2

Molecular Location: base pairs 52,307,283 to 52,529,050 on chromosome 15 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- dilute myosin heavy chain, non-muscle
- GS1
- MYH12
- MYO5
- MYO5A_HUMAN
- myosin-12
- myosin-Va
- myosin VA (heavy chain 12, myoxin)
- myosin, heavy polypeptide kinase
- myoxin
- MYR12
- unconventional myosin-Va

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Unconventional Myosins
<https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1804>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MYO5A%5BTIAB%5D%29+OR+%28myosin+VA%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- MYOSIN VA
<http://omim.org/entry/160777>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MYO5A.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MYO5A%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:7602
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:4644>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4644>
- UniProt
<https://www.uniprot.org/uniprot/Q9Y4I1>

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<https://ghr.nlm.nih.gov/gene/MYO5A>

Reviewed: September 2013

Published: May 14, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services