

## 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.

### **Other Names for This Gene**

- dilute myosin heavy chain, non-muscle
- GS1
- MYH12
- MYO5
- MYO5A\_HUMAN
- myosin VA (heavy chain 12, myoxin)
- myosin, heavy polypeptide kinase
- myosin-12
- myosin-Va
- myoxin
- MYR12
- unconventional myosin-Va

### **Additional Information & Resources**

#### Tests Listed in the Genetic Testing Registry

- Tests of MYO5A ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4644\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4644[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

#### Catalog of Genes and Diseases from OMIM

- MYOSIN VA; MYO5A (<https://omim.org/entry/160777>)

#### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4644>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MYO5A\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MYO5A[gene]))

## References

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## Genomic Location

The *MYO5A* gene is found on chromosome 15 (<https://medlineplus.gov/genetics/chromosome/15/>).

**Last updated September 1, 2013**