MYH9 gene

myosin heavy chain 9

Normal Function

The *MYH9* gene provides instructions for making a protein called myosin-9. This protein is one part (subunit) of the myosin IIA protein.

There are three forms of myosin II, called myosin IIA, myosin IIB and myosin IIC. They play roles in cell movement (cell motility); maintenance of cell shape; and cytokinesis, which is the step in cell division when the fluid surrounding the nucleus (the cytoplasm) divides to form two separate cells. While some cells use more than one type of myosin II, certain blood cells such as platelets and white blood cells (leukocytes) use only myosin IIA.

Each type of myosin II protein consists of two heavy chains and four light chains. The heavy chains each have two parts: a head region and a tail region. The head region interacts with actin, a protein that is important for cell movement and shape. The long tail region interacts with other proteins, including the tail regions of other myosin proteins.

Health Conditions Related to Genetic Changes

MYH9-related disorder

More than 45 mutations in the *MYH9* gene have been found to cause *MYH9*-related disorder. This disorder is characterized by bleeding problems, hearing loss, kidney (renal) disease, and clouding of the lens of the eyes (cataracts). Most of the mutations that cause this condition change single protein building blocks (amino acids) in the myosin-9 protein. Mutations that are located near the head of the myosin protein tend to lead to a more severe disorder than mutations that are located toward the tail of the protein. Recurring mutations involving the amino acid arginine at position 702 in the protein tend to result in many problems, including a severely reduced amount of platelets (thrombocytopenia), early-onset renal disease, and hearing loss in infancy.

Mutations in the *MYH9* gene lead to the production of a nonfunctional protein. A nonfunctional myosin-9 protein cannot properly interact with other subunits to form myosin IIA. Platelets, which only express myosin IIA, are most affected by a lack of functional myosin-9, accounting for the thrombocytopenia seen in all individuals with

MYH9-related disorder.

Nonsyndromic hearing loss

MedlinePlus Genetics provides information about Nonsyndromic hearing loss

Other Names for This Gene

- cellular myosin heavy chain, type A
- MYH9_HUMAN
- myosin heavy chain, non-muscle IIa
- myosin, heavy chain 9, non-muscle
- myosin-9
- NMHC-II-A
- NMMHC II-a
- NMMHC-A
- NMMHC-IIA
- NMMHCA
- non-muscle myosin heavy chain A
- non-muscle myosin heavy chain IIa
- nonmuscle myosin heavy chain II-A

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MYH9 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4627[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MYH9%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MYOSIN, HEAVY CHAIN 9, NONMUSCLE; MYH9 (https://omim.org/entry/160775)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4627)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MYH9[gene])

References

- Althaus K, Greinacher A. MYH9-related platelet disorders. Semin Thromb Hemost. 2009 Mar;35(2):189-203. doi: 10.1055/s-0029-1220327. Epub 2009 Apr 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19408192)
- Kunishima S, Saito H. Advances in the understanding of MYH9 disorders. CurrOpin Hematol. 2010 Sep;17(5):405-10. doi: 10.1097/MOH.0b013e32833c069c. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20601875)
- Sekine T, Konno M, Sasaki S, Moritani S, Miura T, Wong WS, Nishio H,Nishiguchi T, Ohuchi MY, Tsuchiya S, Matsuyama T, Kanegane H, Ida K, Miura K,Harita Y, Hattori M, Horita S, Igarashi T, Saito H, Kunishima S. Patients withEpstein-Fechtner syndromes owing to MYH9 R702 mutations develop progressiveproteinuric renal disease. Kidney Int. 2010 Jul;78(2):207-14. doi:10.1038/ki.2010.21. Epub 2010 Mar 3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20200500)

Genomic Location

The *MYH9* gene is found on chromosome 22 (https://medlineplus.gov/genetics/chromosome/22/).

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