MYH11 gene

myosin heavy chain 11

Normal Function

The *MYH11* gene provides instructions for making a protein called smooth muscle myosin heavy chain 11. It belongs to a group of proteins called myosins, which are involved in cell movement and the transport of materials within and between cells. Thick filaments made of myosin, along with thin filaments of another protein called actin, are the primary components of muscle fibers and are important for muscle tensing (contraction). Smooth muscle myosin heavy chain 11 forms part of a myosin protein complex found in smooth muscles. Smooth muscles are the muscles that line the internal organs of the body, including the blood vessels, stomach, and intestines; as part of their normal function in the body, these muscles contract and relax involuntarily.

Each myosin protein complex consists of two pairs of light chains, which regulate the complex and are produced from several other genes, and two heavy chains such as that produced from the *MYH11* gene. The heavy chains each have two parts: a head region and a tail region. The head region interacts with actin and includes a segment that attaches (binds) to ATP. ATP is a molecule that supplies energy for the cells' activities, including muscle contraction. The long tail region of the myosin heavy chain interacts with other proteins, including the tail regions of other myosins, enabling them to form thick filaments.

Health Conditions Related to Genetic Changes

Core binding factor acute myeloid leukemia

Rearrangements of genetic material involving the *MYH11* gene are involved in a form of blood cancer known as acute myeloid leukemia (AML). The most common of these rearrangements is an inversion of a region of chromosome 16 (written as inv(16)). An inversion involves breakage of the chromosome in two places; the resulting piece of DNA is reversed and reinserted into the chromosome. Less commonly, a rearrangement known as a translocation occurs between the two copies of chromosome 16 (written as t(16;16)). In this translocation, pieces of DNA from each copy of the chromosome break off and are interchanged. Both types of genetic rearrangement result in the fusion of two genes found on chromosome 16, *CBFB* and *MYH11*. These rearrangements are associated with 5 to 8 percent of AML cases in adults. AML associated with either inv(16) or t(16;16) is classified as core binding factor AML (CBF-

AML).

The protein produced from the normal *CBFB* gene interacts with another protein called RUNX1 to form a complex called core binding factor (CBF). This complex attaches to specific areas of DNA and turns on genes that are involved in the development of blood cells. The protein produced from the fusion gene, CBFβ-MYH11, can still bind to RUNX1; however, the function of CBF is impaired. The presence of CBFβ-MYH11 may block binding of CBF to DNA, impairing its ability to control gene activity. Alternatively, the MYH11 portion of the fusion protein may interact with other proteins that prevent the complex from controlling gene activity. This change in gene activity blocks the maturation (differentiation) of blood cells and leads to the production of abnormal, immature white blood cells called myeloid blasts. While inv(16) and t(16;16) are important for leukemia development, one or more additional genetic changes are typically needed for the myeloid blasts to develop into cancerous leukemia cells.

Familial thoracic aortic aneurysm and dissection

MedlinePlus Genetics provides information about Familial thoracic aortic aneurysm and dissection

Intestinal pseudo-obstruction

MedlinePlus Genetics provides information about Intestinal pseudo-obstruction

Megacystis-microcolon-intestinal hypoperistalsis syndrome

MedlinePlus Genetics provides information about Megacystis-microcolon-intestinal hypoperistalsis syndrome

Other Names for This Gene

- AAT4
- FAA4
- MYH11_HUMAN
- myosin heavy chain, smooth muscle isoform
- myosin, heavy chain 11, smooth muscle
- myosin, heavy polypeptide 11, smooth muscle
- myosin-11
- myosin-11 isoform SM1A
- myosin-11 isoform SM1B
- myosin-11 isoform SM2A
- myosin-11 isoform SM2B
- SMHC
- SMMHC

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MYH11 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4629[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MYH11%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+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MYOSIN, HEAVY CHAIN 11, SMOOTH MUSCLE; MYH11 (https://omim.org/entry/160745)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4629)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MYH11[gene])

References

- Durst KL, Lutterbach B, Kummalue T, Friedman AD, Hiebert SW. The inv(16)fusion protein associates with corepressors via a smooth muscle myosinheavy-chain domain. Mol Cell Biol. 2003 Jan;23(2):607-19. doi:10.1128/MCB.23.2.607-619.2003. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12509458) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC151524/)
- Eghtedar A, Borthakur G, Ravandi F, Jabbour E, Cortes J, Pierce S, KantarjianH, Garcia-Manero G. Characteristics of translocation (16;16)(p13;q22) acutemyeloid leukemia. Am J Hematol. 2012 Mar;87(3):317-8. doi: 10.1002/ajh.22258.Epub 2012 Jan 7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22228403) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4221258/)
- Goyama S, Mulloy JC. Molecular pathogenesis of core binding factor leukemia: current knowledge and future prospects. Int J Hematol. 2011 Aug;94(2):126-133.doi: 10.1007/s12185-011-0858-z. Epub 2011 May 3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21537931)
- Shigesada K, van de Sluis B, Liu PP. Mechanism of leukemogenesis by theinv(16) chimeric gene CBFB/PEBP2B-MHY11. Oncogene. 2004 May 24;23(24):4297-307. doi: 10.1038/sj.onc.1207748. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15156186)

Genomic Location

The *MYH11* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

Last updated November 1, 2013