MYH7 gene
myosin heavy chain 7

Normal Function

The MYH7 gene provides instructions for making a protein known as the beta (β)-myosin heavy chain. This protein is found in heart (cardiac) muscle and in type I skeletal muscle fibers. (Skeletal muscle are the muscles used for movement.) Type I fibers, which are also known as slow-twitch fibers, are one of two types of fibers that make up skeletal muscles. Type I fibers are the primary component of skeletal muscles that are resistant to fatigue. For example, muscles involved in posture, such as the neck muscles that hold the head steady, are made predominantly of type I fibers.

In cardiac and skeletal muscle cells, the β-myosin heavy chain forms part of a larger protein called type II myosin. Each type II myosin protein consists of two heavy chains (produced from the MYH7 gene) and two pairs of regulatory light chains (produced from several other genes). The heavy chains each have two parts: a head region and a tail region. The head region, called the motor domain, interacts with a protein called actin, which is important for cell movement and shape. The long tail region interacts with other proteins, including the tail regions of other myosin proteins.

Type II myosin generates the mechanical force that is needed for muscles to contract. It is integral to muscle cell structures called sarcomeres, which are the basic units of muscle contraction. Sarcomeres are composed of thick filaments made up of type II myosin and thin filaments made up of actin. The overlapping thick and thin filaments attach to each other and release, which allows the filaments to move relative to one another so that muscles can contract. In the heart, regular contractions of cardiac muscle pump blood to the rest of the body. The coordinated contraction and relaxation of skeletal muscles allow the body to move.

Health Conditions Related to Genetic Changes
Familial hypertrophic cardiomyopathy

Mutations in the MYH7 gene are a common cause of familial hypertrophic cardiomyopathy, accounting for up to 35 percent of all cases. This condition is characterized by thickening (hypertrophy) of the cardiac muscle. Although some people with familial hypertrophic cardiomyopathy have no obvious health effects, all affected individuals have an increased risk of heart failure and sudden death.

Most MYH7 gene mutations that cause familial hypertrophic cardiomyopathy change single protein building blocks (amino acids) in the β-myosin heavy chain protein. The altered protein is likely incorporated into the thick filament, but it may not function
properly. It is unclear how MYH7 gene mutations lead to the features of familial hypertrophic cardiomyopathy.

Laing distal myopathy

At least six mutations in the MYH7 gene have been found to cause Laing distal myopathy. This condition causes progressive muscle weakness, particularly affecting the arms and legs. Most of the mutations result in changes in the tail region of the β-myosin heavy chain. Some of these mutations change single amino acids, while others delete a single amino acid from the heavy chain. Changes in the MYH7 gene probably disrupt the normal function of type II myosin in muscle cells. Specifically, researchers suspect that mutations alter the structure of the tail region of the β-myosin heavy chain. The altered tail region may be unable to interact with other proteins, including the tail regions of other myosin proteins. It is unclear how these changes in the structure and function of myosin lead to progressive muscle weakness in people with Laing distal myopathy.

Left ventricular noncompaction

At least 30 MYH7 gene mutations have been found to cause left ventricular noncompaction, which occurs when the lower left chamber of the heart (left ventricle) does not develop correctly. The heart muscle is weakened and cannot pump blood efficiently. These cardiac abnormalities can result in a wide range of outcomes from a complete lack of symptoms to sudden cardiac death. Other signs and symptoms include an irregular heart rhythm (arrhythmia), shortness of breath (dyspnea), and heart failure.

It is unclear how MYH7 gene mutations cause left ventricular noncompaction. During normal development before birth, cardiac muscle gets compacted, becoming smooth and firm. MYH7 gene mutations likely lead to changes in this process, resulting in a left ventricular cardiac muscle that is not compacted but is thick and spongy. This abnormal cardiac muscle is weak and cannot contract effectively, causing the varied signs and symptoms of left ventricular noncompaction.

Myosin storage myopathy

At least six mutations in the MYH7 gene are involved in myosin storage myopathy. This condition causes muscle weakness and is characterized by the formation of protein clumps, which include type II myosin, within type I skeletal muscle fibers. The MYH7 gene mutations that cause myosin storage myopathy change amino acids in the tail region of cardiac β-myosin heavy chain. Researchers suggest that these mutations impair the proper formation of thick filaments. The abnormal proteins accumulate in type I skeletal muscle fibers, forming the protein clumps characteristic of the disorder. It is unclear how the gene mutations lead to muscle weakness in people with myosin storage myopathy.
Congenital fiber-type disproportion

Familial dilated cardiomyopathy

Familial restrictive cardiomyopathy

**Chromosomal Location**

Cytogenetic Location: 14q11.2, which is the long (q) arm of chromosome 14 at position 11.2

Molecular Location: base pairs 23,412,738 to 23,435,686 on chromosome 14 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- beta-myosin heavy chain
- MGC138376
- MGC138378
- MPD1
- MYH7_HUMAN
- MyHC-beta
- myhc-slow
- MYHCB
- myosin heavy chain (AA 1-96)
- Myosin heavy chain 7
- Myosin heavy chain, cardiac muscle beta isoform
- Myosin, cardiac, heavy chain, beta
- myosin, heavy chain 7, cardiac muscle, beta
- myosin, heavy polypeptide 7, cardiac muscle, beta
• SPMD
• SPMM

**Additional Information & Resources**

**Educational Resources**

  https://www.ncbi.nlm.nih.gov/books/NBK26888/#A3042

  https://www.ncbi.nlm.nih.gov/books/NBK26888/#A3065

  https://www.ncbi.nlm.nih.gov/books/NBK21724/

- Neuromuscular Disease Center, Washington University: Myosin and associated muscle proteins
  https://neuromuscular.wustl.edu/mother/myosin.htm

  https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1791

**Clinical Information from GeneReviews**

- Dilated Cardiomyopathy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1309

- Hypertrophic Cardiomyopathy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1768

- Laing Distal Myopathy
  https://www.ncbi.nlm.nih.gov/books/NBK1433

**Scientific Articles on PubMed**

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28MYH7%5BTIAB%5D%29+AND+%28%2BGenes%5BMH%5D%29+OR+%2BGenetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

**Catalog of Genes and Diseases from OMIM**

- CARDIOMYOPATHY, DILATED, 1S
  http://omim.org/entry/613426

- MYOSIN, HEAVY CHAIN 7, CARDIAC MUSCLE, BETA
  http://omim.org/entry/160760
Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MYH7.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MYH7%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4625
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P12883

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19336582
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2669361/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12601548
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Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1182058/

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