MYOT gene

myotilin

Normal Function

The *MYOT* gene provides instructions for making a protein called myotilin. Myotilin is found in heart (cardiac) muscle and muscles used for movement (skeletal muscle). Within muscle fibers, myotilin proteins are found in structures called sarcomeres, which are necessary for muscles to tense (contract). Myotilin attaches (binds) to other proteins to help form sarcomeres. Myotilin is also involved in linking neighboring sarcomeres to each another to form myofibrils, the basic unit of muscle fibers. The connection of sarcomeres to each other and the formation of myofibrils are essential for maintaining muscle fiber strength during repeated cycles of contraction and relaxation.

Health Conditions Related to Genetic Changes

Myofibrillar myopathy

At least five mutations in the *MYOT* gene have been found to cause myofibrillar myopathy. Most of these mutations are located in an area of the gene known as exon 2. *MYOT* gene mutations that cause myofibrillar myopathy change single protein building blocks (amino acids) in myotilin. Mutated myotilin proteins cluster together with other muscle proteins in the sarcomere to form clumps (aggregates). The aggregates prevent these proteins from functioning normally. A dysfunctional myotilin protein cannot properly bind with other proteins, preventing the formation of sarcomeres and myofibrils. *MYOT* gene mutations that cause myofibrillar myopathy impair the function of muscle fibers, causing weakness and the other features of this condition.

Limb-girdle muscular dystrophy

MedlinePlus Genetics provides information about Limb-girdle muscular dystrophy

Other Names for This Gene

- MYOTI_HUMAN
- TTID

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MYOT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9499[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MYOT%5BTIAB%5D%29+OR+%28myotilin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%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

- MYOTILIN; MYOT (https://omim.org/entry/604103)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/9499)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MYOT[gene])

References

- Ferrer I, Olive M. Molecular pathology of myofibrillar myopathies. Expert RevMol Med. 2008 Sep 3;10:e25. doi: 10.1017/S1462399408000793. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18764962)
- Schroder R, Schoser B. Myofibrillar myopathies: a clinical and myopathologicalguide. Brain Pathol. 2009 Jul;19(3):483-92. doi:10.1111/j.1750-3639.2009.00289.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19563540)
- Selcen D, Engel AG. Mutations in myotilin cause myofibrillar myopathy.Neurology. 2004 Apr 27;62(8):1363-71. doi: 10.1212/01.wnl.0000123576.74801.75.Erratum In: Neurology. 2004 Jul 27;63(2):405. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15111675)

Genomic Location

The *MYOT* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

Last updated April 1, 2011