Myosin storage myopathy

Description

Myosin storage myopathy is a condition that causes muscle weakness (myopathy) that does not worsen or worsens very slowly over time. This condition is characterized by the formation of protein clumps, which contain a protein called myosin, within certain muscle fibers. The signs and symptoms of myosin storage myopathy usually become noticeable in childhood, although they can occur later. Because of muscle weakness, affected individuals may start walking later than usual and have a waddling gait, trouble climbing stairs, and difficulty lifting the arms above shoulder level. Muscle weakness also causes some affected individuals to have trouble breathing.

Frequency

Myosin storage myopathy is a rare condition. Its prevalence is unknown.

Causes

Mutations in the *MYH7* gene cause myosin storage myopathy. The *MYH7* gene provides instructions for making a protein known as the cardiac beta (β)-myosin heavy chain. This protein is found in heart (cardiac) muscle and in type I skeletal muscle fibers, one of two types of fibers that make up the muscles that the body uses for movement. Cardiac β-myosin heavy chain is the major component of the thick filament in muscle cell structures called sarcomeres. Sarcomeres, which are made up of thick and thin filaments, are the basic units of muscle contraction. 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.

Mutations in the *MYH7* gene lead to the production of an altered cardiac β-myosin heavy chain protein, which is thought to be less able to form thick filaments. The altered proteins accumulate in type I skeletal muscle fibers, forming the protein clumps characteristic of the disorder. It is unclear how these changes lead to muscle weakness in people with myosin storage myopathy.

Learn more about the gene associated with Myosin storage myopathy

- MYH7

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Autosomal dominant hyaline body myopathy

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Myosin storage myopathy (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842160/)

Genetic and Rare Diseases Information Center

- Hyaline body myopathy (https://rarediseases.info.nih.gov/diseases/7148/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Myosin storage myopathy%22)

Catalog of Genes and Diseases from OMIM

- CONGENITAL MYOPATHY 7A, MYOSIN STORAGE, AUTOSOMAL DOMINANT; CMYP7A (https://omim.org/entry/608358)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28myosin+storage+myopathy%5BTIAB%5D%29+OR+%28autosomal+dominant+hyaline+body+myopathy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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