

Laing distal myopathy

Description

Laing distal myopathy is a condition that affects skeletal muscles, which are muscles that the body uses for movement. This disorder causes progressive muscle weakness that appears in childhood. The first sign of Laing distal myopathy is usually weakness in certain muscles in the feet and ankles. This weakness leads to tightening of the Achilles tendon (the band that connects the heel of the foot to the calf muscles), an inability to lift the first (big) toe, and a high-stepping walk. Months to years later, muscle weakness develops in the hands and wrists. Weakness in these muscles makes it difficult to lift the fingers, particularly the third and fourth fingers. Many affected people also experience hand tremors.

In addition to muscle weakness in the hands and feet, Laing distal myopathy causes weakness in several muscles of the neck and face. A decade or more after the onset of symptoms, mild weakness also spreads to muscles in the legs, hips, and shoulders. Laing distal myopathy progresses very gradually, and most affected people remain mobile throughout life. Life expectancy is normal in people with this condition.

Frequency

Although Laing distal myopathy is thought to be rare, its prevalence is unknown. Several families with the condition have been identified worldwide.

Causes

Mutations in the *MYH7* gene cause Laing distal myopathy. The *MYH7* gene provides instructions for making a protein that is found in heart (cardiac) muscle and in type I skeletal muscle fibers. 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 protein produced from the *MYH7* gene forms part of a larger protein called type II myosin. This type of myosin generates the mechanical force that is needed for muscles to 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.

It is unknown how mutations in the *MYH7* gene cause progressive muscle weakness in people with Laing distal myopathy. Researchers have proposed that these mutations alter the structure of myosin in skeletal muscles, which prevents it from interacting with other proteins. The abnormal myosin gradually impairs the function of type I skeletal muscle fibers.

In most people with Laing distal myopathy, the signs and symptoms of the disorder are limited to weakness of skeletal muscles. Because myosin made with the MYH7 protein is also found in cardiac muscle, it is unclear why heart problems are not a typical feature of this condition.

[Learn more about the gene associated with Laing distal 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.

In most cases, an affected person inherits the mutation from one affected parent. A small percentage of cases result from new mutations in the gene. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- Distal myopathy 1
- Laing early-onset distal myopathy
- MPD1

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: MYH7-related skeletal myopathy (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C4552004/>)

Genetic and Rare Diseases Information Center

- Laing early-onset distal myopathy (<https://rarediseases.info.nih.gov/diseases/10769/index>)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- MYOPATHY, DISTAL, 1; MPD1 (<https://omim.org/entry/160500>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28laing+AND+distal+myopathy%5BTIAB%5D%29+OR+%28laing+AND+early-onset+AND+distal+myopathy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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