DYSF gene

dysferlin

Normal Function

The *DYSF* gene provides instructions for making a protein called dysferlin. This protein is found in the thin membrane that surrounds muscle fibers, which is called the sarcolemma. Dysferlin is thought to play an important role in repairing the sarcolemma when it becomes damaged due to muscle strain. Researchers suggest that dysferlin may also be involved in transporting materials within muscle fibers. Dysferlin may also work together with calcium to regulate cell signaling pathways.

Health Conditions Related to Genetic Changes

Limb-girdle muscular dystrophy

Variants (also called mutations) in the *DYSF* gene can cause a type of limb-girdle muscular dystrophy called LGMD R2. Limb-girdle muscular dystrophy is characterized by weakness (myopathy) and wasting (atrophy) of the muscles that are close to the center of the body (proximal muscles), such as the muscles of the shoulders, hips, thighs, and upper arms.

The *DYSF* gene variants that lead to limb-girdle muscular dystrophy cause cells to produce a version of the protein that does not function properly. The altered protein interferes with muscle repair, which likely leads to the myopathy and atrophy seen in people with LGMD R2.

Miyoshi myopathy

Variants in the *DYSF* gene can also cause Miyoshi myopathy, a muscle disorder that is characterized by myopathy and atrophy of the muscles that are away from the center of the body (distal muscles), particularly those in the lower legs. Many of the *DYSF* gene variants that are associated with Miyoshi myopathy lead to the substitution of one protein building block (amino acid) for another in the dysferlin protein. The altered protein does not function properly, which interferes with the cell's ability to repair damage to the sarcolemma. This impairs muscle function and leads to the myopathy and atrophy seen in people with Miyoshi myopathy. Recently, researchers have suggested that LGMD R2 and Miyoshi myopathy are not distinct disorders but are part of the same disease spectrum.

Other disorders

DYSF gene variants also cause a condition called distal myopathy with anterior tibial onset. This condition is characterized by myopathy that is most apparent in the muscles of the lower legs. The first muscle affected is typically the tibialis anterior, which is located at the front of the lower leg and helps to flex the foot. The myopathy associated with this condition worsens over time.

Muscle problems caused by changes in the *DYSF* gene are called dysferlinopathies. Some researchers have proposed that the conditions caused by variants in the *DYSF* gene are not separate disorders but are actually one disorder. These researchers believe that the term "dysferlinopathy" should be used to refer to all of the disorders that are caused by variants in the *DYSF* gene.

Other Names for This Gene

- FER1L1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DYSF (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8291[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DYSF%5BTIAB%5D%29+OR+%28dysferlin%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

- DYSFERLIN; DYSF (https://omim.org/entry/603009)
- MYOPATHY, DISTAL, WITH ANTERIOR TIBIAL ONSET; DMAT (https://omim.org/entry/606768)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8291)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DYSF[gene])

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Genomic Location

The *DYSF* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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