

TPM2 gene

tropomyosin 2

Normal Function

The *TPM2* gene provides instructions for making a protein called beta (β)-tropomyosin, which is part of the tropomyosin protein family. Tropomyosin proteins regulate the tensing of muscle fibers (muscle contraction) by controlling the binding of two muscle proteins, myosin and actin. In non-muscle cells, tropomyosin proteins play a role in controlling cell shape.

β -tropomyosin is found primarily in skeletal muscles, which are the muscles used for movement. This protein helps regulate muscle contraction by interacting with other muscle proteins, particularly myosin and actin. These interactions are essential for stabilizing and maintaining structures called sarcomeres within muscle cells. Sarcomeres are the basic units of muscle contraction; they are made of proteins that generate the mechanical force needed for muscles to contract.

Health Conditions Related to Genetic Changes

Cap myopathy

At least three *TPM2* gene mutations have been identified in people with cap myopathy, a disorder that leads to muscle weakness (myopathy) and poor muscle tone (hypotonia). These mutations delete or duplicate genetic material in the *TPM2* gene or replace single protein building blocks (amino acids) in the β -tropomyosin protein sequence. The specific effects of these *TPM2* gene mutations are unclear, but researchers suggest they may interfere with normal actin-myosin binding, impairing muscle contraction and resulting in the muscle weakness that occurs in cap myopathy.

Distal arthrogryposis type 1

At least three mutations in the *TPM2* gene have been found to cause distal arthrogryposis type 1, a disorder characterized by joint deformities (contractures) in the hands and feet. It is unclear how these mutations lead to contractures in people with distal arthrogryposis type 1, or why the joint problems are typically limited to the hands and feet. However, researchers speculate that contractures may be related to problems with muscle contraction that limit the movement of joints before birth.

Congenital fiber-type disproportion

MedlinePlus Genetics provides information about Congenital fiber-type disproportion

Nemaline myopathy

MedlinePlus Genetics provides information about Nemaline myopathy

Sheldon-Hall syndrome

At least six *TPM2* gene mutations have been identified in people with Sheldon-Hall syndrome, a muscle and skeletal disorder similar to distal arthrogryposis type 1 (described above) that impairs joint movement in the hands and feet. Mutations in the *TPM2* gene may alter the structure of β -tropomyosin and disrupt the protein's normal function in controlling muscle contractions, resulting in the contractures and other muscle and skeletal abnormalities associated with this condition.

Other Names for This Gene

- beta-tropomyosin
- NEM4
- TMSB
- TPM2_HUMAN
- tropomyosin 2 (beta)
- tropomyosin beta chain
- tropomyosin, skeletal muscle beta
- tropomyosin-2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TPM2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7169\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7169[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TPM2%5BTIAB%5D%29+OR+%28tropomyosin+2%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+3600+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- TROPOMYOSIN 2; TPM2 (<https://omim.org/entry/190990>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7169>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=TPM2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=TPM2[gene]))

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Genomic Location

The *TPM2* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

Last updated June 1, 2015