TNNI2 gene

troponin I2, fast skeletal type

Normal Function

The *TNNI2* gene provides instructions for making one form of a protein called troponin I. The troponin I protein produced from the *TNNI2* gene is found in skeletal muscles, which are the muscles used for movement. Troponin I is one of three proteins that make up the troponin complex in muscle cells. The troponin complex, along with calcium, helps regulate muscle tensing (contraction).

The troponin complex is part of a structure called the sarcomere, which is the basic unit of muscle contraction. Sarcomeres contain thick and thin filaments. The overlapping thick and thin filaments attach (bind) to each other and release, which allows the filaments to move relative to one another so that muscles can contract.

When calcium levels are low, the troponin complex blocks the binding between the thick and thin filaments that is needed for muscle contraction. An increase in calcium levels causes structural changes in the troponin complex, which exposes the binding sites and allows the thick and thin filaments to interact, leading to muscle contraction.

Health Conditions Related to Genetic Changes

Sheldon-Hall syndrome

At least eight *TNNI2* gene mutations have been identified in people with Sheldon-Hall syndrome. This disorder affects muscle and skeletal development before birth and is characterized by joint deformities (contractures) that restrict movement in the hands and feet. Researchers suggest that the *TNNI2* gene mutations that cause Sheldon-Hall syndrome may prevent the troponin complex from blocking thick and thin filament binding to control muscle contractions, resulting in the contractures and other muscle and skeletal abnormalities associated with Sheldon-Hall syndrome.

Other Names for This Gene

- AMCD2B
- DA2B
- FSSV
- fsTnI

- troponin I fast twitch 2
- troponin I type 2 (skeletal, fast)
- troponin I, fast skeletal muscle
- troponin I, fast skeletal muscle isoform 1
- troponin I, fast skeletal muscle isoform 2
- troponin I, fast-twitch isoform
- troponin I, fast-twitch skeletal muscle isoform
- troponin I, skeletal, fast

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TNNI2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7136[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TNNI2%5BTIAB%5D%29+OR+%28%28DA2B%5BTIAB%5D%29+OR+%28FSSV%5BTIAB%5D%29+OR+%28fsTnI%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+3240+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TROPONIN I, FAST-TWITCH SKELETAL MUSCLE ISOFORM; TNNI2 (https://omim.org/entry/191043)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7136)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TNNI2[gene])

References

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- Toydemir RM, Bamshad MJ. Sheldon-Hall syndrome. Orphanet J Rare Dis. 2009 Mar23;4:11. doi: 10.1186/1750-1172-4-11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19309503) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2663550/)

Genomic Location

The *TNNI2* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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