TNNT2 gene

troponin T2, cardiac type

Normal Function

The *TNNT2* gene provides instructions for making a protein called cardiac troponin T, which is found solely in the heart (cardiac) muscle. Cardiac troponin T is one of three proteins that make up the troponin protein complex in cardiac muscle cells. The troponin complex is part of a structure called the sarcomere, which is the basic unit of muscle contraction. Sarcomeres are made up of 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. The troponin complex, along with calcium, helps regulate contraction of cardiac muscle.

For the heart to beat normally, cardiac muscle must contract and relax in a coordinated way. Cardiac troponin T helps coordinate contraction of the heart muscle. When calcium levels are low, the troponin complex binds to the thin filament in sarcomeres, which blocks the interaction 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 allows the thick and thin filaments to interact, leading to contraction of the heart muscle.

Health Conditions Related to Genetic Changes

Familial hypertrophic cardiomyopathy

Mutations in the *TNNT2* gene can cause familial hypertrophic cardiomyopathy, a condition characterized by thickening (hypertrophy) of the cardiac muscle. *TNNT2* gene mutations are found in approximately 5 percent of individuals with this condition. Although some people with hypertrophic cardiomyopathy have no obvious health effects, all affected individuals have an increased risk of heart failure and sudden death.

Most *TNNT2* gene mutations in familial hypertrophic cardiomyopathy change single protein building blocks (amino acids) in the cardiac troponin T protein. The altered protein is likely incorporated into the troponin complex, but it may not function properly. However, it is unclear how the gene mutations lead to the features of familial hypertrophic cardiomyopathy.

Familial dilated cardiomyopathy

Mutations in the *TNNT2* gene have been found in people with a heart condition called familial dilated cardiomyopathy. The role *TNNT2* gene mutations play in this disorder is unclear. Familial dilated cardiomyopathy is a condition that weakens and enlarges the heart, preventing it from pumping blood efficiently. Familial dilated cardiomyopathy increases the risk of heart failure and premature death.

Familial restrictive cardiomyopathy

MedlinePlus Genetics provides information about Familial restrictive cardiomyopathy

Left ventricular noncompaction

Mutations in the *TNNT2* gene have been found in people with a heart condition called left ventricular noncompaction. However, the role *TNNT2* gene mutations play in this disorder is unclear. Left ventricular noncompaction occurs when the lower left chamber of the heart (left ventricle) does not develop correctly. The heart muscle is weakened and cannot pump blood efficiently, often leading to heart failure. Abnormal heart rhythms (arrhythmias) can also occur in individuals with left ventricular noncompaction.

Other Names for This Gene

- cardiac muscle troponin T
- cTnT
- LVNC6
- RCM3
- TNNT2_HUMAN
- TnTC
- troponin T type 2 (cardiac)
- troponin T, cardiac muscle
- troponin T2, cardiac

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TNNT2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7139[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TNNT2%5BTIAB%5D%29+OR+%28troponin+T+type+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+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CARDIOMYOPATHY, DILATED, 1D; CMD1D (https://omim.org/entry/601494)
- TROPONIN T2, CARDIAC; TNNT2 (https://omim.org/entry/191045)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7139)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TNNT2[gene])

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Genomic Location

The *TNNT2* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

Last updated February 1, 2013