TNNT2 Gene

Subjects: Genetics & Heredity
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Troponin T2, cardiac type: The TNNT2 gene provides instructions for making a protein called cardiac troponin T, which is found solely in the heart (cardiac) muscle.

genes

1. 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.

2. Health Conditions Related to Genetic Changes

2.1. 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.

2.2. Other disorders

Mutations in the *TNNT2* gene have been found in people with other heart conditions, including dilated cardiomyopathy and left ventricular noncompaction. However, the role *TNNT2* gene mutations play in either disorder is unclear. Dilated cardiomyopathy is a condition that weakens and enlarges the heart, preventing it from pumping blood efficiently. Dilated cardiomyopathy increases the risk of heart failure and premature death. 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.

Familial dilated cardiomyopathy

Familial restrictive cardiomyopathy

Left ventricular noncompaction

3. 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

References

1. Bashyam MD, Savithri GR, Kumar MS, Narasimhan C, Nallari P. Molecular geneticsof familial hypertrophic cardiomyopathy (FHC). J Hum Genet. 2003;48(2):55-64.Review.
2. Gomes AV, Barnes JA, Harada K, Potter JD. Role of troponin T in disease. MolCell Biochem. 2004 Aug;263(1-2):115-29. doi: 10.1023/B:MCBI.0000041853.20588.a0. Review.
3. Hershberger RE, Pinto JR, Parks SB, Kushner JD, Li D, Ludwigsen S, Cowan J,Morales A, Parvatiyar MS, Potter JD. Clinical and functional characterization of TNNT2 mutations identified in

patients with dilated cardiomyopathy. CircCardiovasc Genet. 2009 Aug;2(4):306-13. doi: 10.1161/CIRCGENETICS.108.846733.

4. Keren A, Syrris P, McKenna WJ. Hypertrophic cardiomyopathy: the geneticdeterminants of clinical disease expression. Nat Clin Pract Cardiovasc Med. 2008 Mar;5(3):158-68. doi: 10.1038/ncpcardio1110.in: Nat Clin Pract Cardiovasc Med. 2008 Nov;5(11):747.
5. Klaassen S, Probst S, Oechslin E, Gerull B, Krings G, Schuler P, Greutmann M, Hürlimann D, Yegitbasi M, Pons L, Gramlich M, Drenckhahn JD, Heuser A, Berger F, Jenni R, Thierfelder L. Mutations in sarcomere protein genes in left ventricular noncompaction. Circulation. 2008 Jun 3;117(22):2893-901. doi:10.1161/CIRCULATIONAHA.107.746164.
6. Luedde M, Ehlermann P, Weichenhan D, Will R, Zeller R, Rupp S, Müller A, SteenH, Ivandic BT, Ulmer HE, Kern M, Katus HA, Frey N. Severe familial leftventricular non-compaction cardiomyopathy due to a novel troponin T (TNNT2)mutation. Cardiovasc Res. 2010 Jun 1;86(3):452-60. doi: 10.1093/cvr/cvq009.
7. Marston SB. How do mutations in contractile proteins cause the primaryfamilial cardiomyopathies? J Cardiovasc Transl Res. 2011 Jun;4(3):245-55. doi:10.1007/s12265-011-9266-2.
8. Otten E, Lekanne Dit Deprez RH, Weiss MM, van Slegtenhorst M, Joosten M, vander Smagt JJ, de Jonge N, Kerstjens-Frederikse WS, Roofthooft MT, Balk AH, vanden Berg MP, Ruiter JS, van Tintelen JP. Recurrent and founder mutations in theNetherlands: mutation p.K217del in troponin T2, causing dilated cardiomyopathy.Neth Heart J. 2010 Oct;18(10):478-85.
9. Rodríguez JE, McCudden CR, Willis MS. Familial hypertrophic cardiomyopathy:basic concepts and future molecular diagnostics. Clin Biochem. 2009Jun;42(9):755-65. doi: 10.1016/j.clinbiochem.2009.01.020.Review.

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