TRPM4 gene

transient receptor potential cation channel subfamily M member 4

Normal Function

The *TRPM4* gene provides instructions for making a protein called transient receptor potential cation channel subfamily M member 4 (TRPM4). This protein acts as a channel, opening and closing at specific times to control the flow of positively charged atoms (cations) into and out of cells. The TRPM4 channel is embedded in the outer membrane of cells throughout the body, but it is abundant in heart (cardiac) cells and plays key roles in these cells' ability to generate and transmit electrical signals. TRPM4 channels play a major role in signaling the start of each heartbeat, coordinating the contractions of the upper and lower chambers of the heart, and maintaining a normal heart rhythm.

In addition to regulating electric signaling in the heart, the TRPM4 channel is important for the normal functioning of the immune system, the nervous system, the kidneys, and the pancreas.

Health Conditions Related to Genetic Changes

Progressive familial heart block

A few mutations in the *TRPM4* gene have been found to cause progressive familial heart block. This condition alters the normal beating of the heart and can lead to fainting (syncope) or sudden cardiac arrest and death. The *TRPM4* gene mutations change single protein building blocks (amino acids) in the TRPM4 channel. The altered channels have increased activity at the cell membrane, which likely alters the cell's ability to generate electrical signals because of the increased cation flow. Cardiac cells with these altered channels have difficulty producing and transmitting electrical signals that coordinate normal heartbeats. Interruption of this signaling causes heart block. Death of these impaired cardiac cells over time can lead to a buildup of scar tissue (fibrosis), worsening the heart block.

Brugada syndrome

MedlinePlus Genetics provides information about Brugada syndrome

Other Names for This Gene

- calcium-activated non-selective cation channel 1
- hTRPM4
- long transient receptor potential channel 4
- LTrpC4
- melastatin-4
- transient receptor potential cation channel, subfamily M, member 4
- TRPM4_HUMAN
- TRPM4B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TRPM4 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54795[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TRPM4%5BTIAB%5D%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

- TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY M, MEMBER 4; TRPM4 (https://omim.org/entry/606936)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/54795)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TRPM4[gene])

References

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Genomic Location

The *TRPM4* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

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