



TRPM6 gene

transient receptor potential cation channel subfamily M member 6

Normal Function

The *TRPM6* gene provides instructions for making a protein that acts as a channel, which allows charged atoms (ions) of magnesium (Mg^{2+}) to flow into cells; the channel may also allow small amounts of calcium ions (Ca^{2+}) to pass through cells. Magnesium is involved in many cell processes, including production of cellular energy, maintenance of DNA building blocks (nucleotides), protein production, and cell growth and death.

Additionally, Mg^{2+} is needed for the production of a substance called parathyroid hormone that regulates blood calcium levels. Magnesium and calcium are also required for the normal functioning of nerve cells that control muscle movement (motor neurons).

The TRPM6 channel is embedded in the membrane of epithelial cells that line the large intestine, structures in the kidneys known as distal convoluted tubules, the lungs, and the testes in males. When the body needs additional Mg^{2+} , the TRPM6 channel allows it to be absorbed in the intestine and filtered from the fluids that pass through the kidneys by the distal convoluted tubules. When the body has sufficient or too much Mg^{2+} , the TRPM6 channel does not filter out the Mg^{2+} from fluids but allows the ion to be released from the kidney cells into the urine. The channel also helps to regulate Ca^{2+} , but to a lesser degree.

Health Conditions Related to Genetic Changes

Hypomagnesemia with secondary hypocalcemia

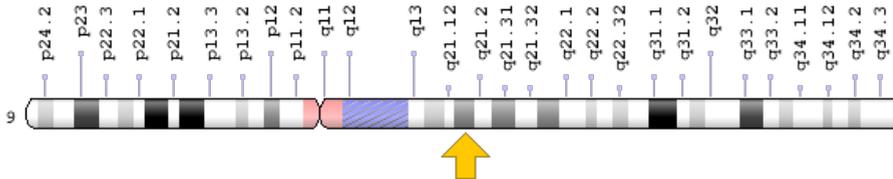
At least 38 mutations in the *TRPM6* gene have been found to cause hypomagnesemia with secondary hypocalcemia. This condition is characterized by low levels of magnesium (hypomagnesemia) and calcium (hypocalcemia) in the body, which leads to neurological problems that begin in infancy, including muscle spasms and seizures. *TRPM6* gene mutations result in a lack of functional protein.

Nonfunctional TRPM6 channels prevent Mg^{2+} absorption in the intestine and cause too much Mg^{2+} to be released in the urine. A lack of Mg^{2+} in the blood impairs the production of parathyroid hormone, which likely reduces blood Ca^{2+} levels. Additionally, hypomagnesemia and hypocalcemia can disrupt many cell processes and impair the function of motor neurons, leading to neurological problems and movement disorders characteristic of this condition. If the condition is not effectively treated and low Mg^{2+} levels persist, signs and symptoms can worsen over time and may lead to early death.

Chromosomal Location

Cytogenetic Location: 9q21.13, which is the long (q) arm of chromosome 9 at position 21.13

Molecular Location: base pairs 74,722,495 to 74,888,094 on chromosome 9 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CHAK2
- channel kinase 2
- FLJ22628
- HMGX
- melastatin-related TRP cation channel 6
- transient receptor potential cation channel, subfamily M, member 6

Additional Information & Resources

Educational Resources

- TRP Ion Channel Function in Sensory Transduction and Cellular Signaling Cascades (2007): Structural Insights into the Function of TRP Channels
<https://www.ncbi.nlm.nih.gov/books/NBK5266/>
- TRP Ion Channel Function in Sensory Transduction and Cellular Signaling Cascades (2007): TRP Channel Trafficking
<https://www.ncbi.nlm.nih.gov/books/NBK5269/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28TRPM6%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 6
<http://omim.org/entry/607009>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TRPM6.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TRPM6%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:17995
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:140803>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/140803>
- TRIP Database
<http://www.trpchannel.org/summaries/TRPM6>
- UniProt
<https://www.uniprot.org/uniprot/Q9BX84>

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