TRPM6 Gene

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Contributor: Rui Liu

Transient receptor potential cation channel subfamily M member 6: The TRPM6 gene provides instructions for making a protein that acts as a channel, which allows charged atoms (ions) of magnesium (Mg2+) to flow into cells; the channel may also allow small amounts of calcium ions (Ca2+) to pass through cells.

Keywords: genes

1. 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.

2. Health Conditions Related to Genetic Changes

2.1. 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.

3. 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

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