KCNK9 Gene

Subjects: Genetics & Heredity

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Potassium two pore domain channel subfamily K member 9

genes

1. Introduction

The *KCNK9* gene provides instructions for making a protein called TASK3, which functions as a potassium channel. Potassium channels transport positively charged atoms (ions) of potassium into and out of cells.

TASK3 channels are found throughout the body. They are especially abundant in nerve cells (neurons) in the brain, particularly the region of the brain that coordinates movement (cerebellum). The flow of ions through potassium channels in neurons is involved in activating (exciting) the neurons and sending electrical signals in the brain. Unlike some potassium channels that open and close in response to certain triggers, TASK3 channels are always open, although their activity can be controlled by the environment surrounding the cell. Because the channels are always open, they are often called background or leak channels. TASK3 channels maintain the cell's ability to generate electrical signals and regulate the activity (excitability) of cells. These channels also appear to play a role in the movement (migration) of certain neurons in the brain.

People inherit two copies of their genes, one from their mother and one from their father. Usually both copies of each gene are active, or "turned on," in cells. For some genes, however, only one of the two copies is normally turned on. Which copy is active depends on the parent of origin: some genes are normally active only when they are inherited from a person's father; others are active only when inherited from a person's mother. This phenomenon is known as genomic imprinting. The *KCNK9* gene is a maternally expressed imprinted gene, which means that only the copy of the gene that comes from the mother is active. The copy of the gene that comes from the father is turned off (silenced).

2. Health Conditions Related to Genetic Changes

2.1. KCNK9 Imprinting Syndrome

At least two changes in the *KCNK9* gene, both of which have the same effect on the TASK3 channel protein, have been found to cause *KCNK9* imprinting syndrome. This condition is characterized by weak muscle tone (hypotonia) from birth that can affect the ability to eat. Affected individuals typically have intellectual disability and delayed

development of speech and motor skills, such as walking. Because the copy of the *KCNK9* gene from the father is silenced, the condition occurs only when there is a mutation in the copy of the gene inherited from the mother.

The gene mutations that cause *KCNK9* imprinting syndrome change a single protein building block (amino acid) in the TASK3 channel; the amino acid arginine replaces the amino acid glycine at position 236 (written as Gly236Arg or G236R). This alteration reduces the flow of ions through TASK3 channels by 80 percent. Research suggests that certain neurons with altered TASK3 channels are unable to repeatedly generate electrical signals. The reduction of ion transport through TASK3 channels disrupts normal neuron development and excitability. Impairment of neuron function likely underlies the hypotonia, intellectual disability, and developmental problems characteristic of *KCNK9* imprinting syndrome.

3. Other Names for This Gene

- acid-sensitive potassium channel protein TASK-3
- K2p9.1
- KT3.2
- potassium channel subfamily K member 9
- potassium channel, two pore domain subfamily K, member 9
- TASK-3
- TASK3
- TWIK-related acid-sensitive K(+) channel 3
- two pore K(+) channel KT3.2
- two pore potassium channel KT3.2

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