KCNA1 Gene

Subjects: Genetics
Submitted by: Dean Liu
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Definition

Potassium voltage-gated channel subfamily A member 1

1. Introduction

The *KCNA1* gene belongs to a large family of genes that provide instructions for making potassium channels. These channels, which transport positively charged atoms (ions) of potassium into and out of cells, play a key role in a cell's ability to generate and transmit electrical signals.

The *KCNA1* gene provides instructions for making one part (the alpha subunit) of a potassium channel called Kv1.1. These channels are found in the brain, where they transport potassium ions into nerve cells (neurons). The flow of certain ions, including potassium, into and out of neurons regulates communication between these cells.

2. Health Conditions Related to Genetic Changes

2.1. Episodic Ataxia

At least 20 mutations in the *KCNA1* gene have been identified in people with episodic ataxia type 1 (EA1). People with this form of the condition have brief, recurrent episodes of poor coordination and balance (ataxia). Between episodes, many affected individuals experience myokymia, a muscle abnormality that can cause involuntary muscle cramping, stiffness, and continuous, fine muscle twitching that appears as rippling under the skin.

Most of the *KCNA1* mutations responsible for episodic ataxia change single protein building blocks (amino acids) in the alpha subunit of the Kv1.1 channel. Some of these changes prevent the assembly of functional channels, while other mutations alter the channel's structure. When Kv1.1 channels are missing or abnormal, the flow of potassium ions into neurons is reduced. This decrease in potassium ions overexcites certain neurons in the brain, which disrupts normal communication between these cells. Although changes in signaling between neurons underlie the episodes of uncoordinated movement seen in people with episodic ataxia, it is unclear how altered potassium ion transport causes the specific features of the condition.

2.2. Other disorders

Mutations in the *KCNA1* gene have been found to cause a range of signs and symptoms affecting the nervous system. In at least one family, isolated myokymia (continuous muscle twitching and spasms without episodes of ataxia) has been attributed to *KCNA1* mutations. Changes in this gene have also been identified in a small number of people with epilepsy. Like the *KCNA1* mutations that underlie episodic ataxia, the mutations that cause isolated myokymia and epilepsy reduce the flow of potassium ions through Kv1.1 channels, disrupting normal communication between neurons in the brain. Researchers are working to determine why mutations in this single gene can cause several different disorders of the nervous system.

3. Other Names for This Gene

- AEMK
- EA1
- HBK1
- HUK1

- KCNA1_HUMAN
- KV1.1
- MBK1
- MGC126782
- MGC138385
- MK1
- potassium channel, voltage gated shaker related subfamily A, member 1
- potassium voltage-gated channel, shaker-related subfamily, member 1 (episodic ataxia with myokymia)
- RBK1
- voltage-gated potassium channel subunit Kv1.1

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Keywords

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