KCNT1 Gene

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Potassium sodium-activated channel subfamily T member 1

genes

1. Introduction

The *KCNT1* 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 specific function of a potassium channel depends on its protein components and its location in the body. Channels made with the KCNT1 protein are active in nerve cells (neurons) in the brain, where they transport potassium ions out of cells. This flow of ions is involved in generating currents to activate (excite) neurons and send signals in the brain.

Potassium channels are made up of several protein components (subunits). Each channel contains four alpha subunits that form the hole (pore) through which potassium ions move. Four alpha subunits from the *KCNT1* gene can form a channel. The KCNT1 alpha subunits can also interact with alpha subunits produced from the *KCNT2* gene to form a functional potassium channel.

Researchers have determined that a molecule called PKC can turn on channels made with the KCNT1 protein. While the channels can generate electrical currents without PKC, when PKC turns the channel on, the currents are stronger.

2. Health Conditions Related to Genetic Changes

2.1. Malignant Migrating Partial Seizures of Infancy

At least six *KCNT1* gene mutations have been found in individuals with malignant migrating partial seizures of infancy (MMPSI). This condition is characterized by recurrent seizures beginning before the age of 6 months as well as profound developmental delay. In MMPSI, seizure activity in the brain can spread (migrate) from one region to another during an episode.

The *KCNT1* gene mutations involved in MMPSI change single protein building blocks (amino acids) in the KCNT1 protein. The electrical currents generated by potassium channels made with the altered KCNT1 protein are abnormally increased, as though the channels were turned on by PKC. The increased electrical currents allow unregulated excitation of neurons in the brain. When neurons are abnormally excited, seizures develop. Repeated seizures contribute to the developmental delay that is characteristic of this condition.

2.2. Other Disorders

Mutations in the *KCNT1* gene have been found in several people with autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE), which causes seizures that usually occur at night (nocturnally) while an affected person is sleeping. In addition to seizures, most affected individuals with *KCNT1* gene mutations have psychiatric problems, such as aggression, episodes of unresponsiveness (catatonia), or a distorted view of reality (psychosis), and about half have intellectual disability. The *KCNT1* gene mutations involved in this condition change single amino acids in the KCNT1 protein; however, it is unclear what effects these changes have on the function of potassium channels or how they lead to the features of ADNFLE.

3. Other Names for This Gene

- EIEE14
- ENFL5
- KCa4.1
- KCNT1_HUMAN
- KIAA1422
- potassium channel subfamily T member 1
- potassium channel, sodium activated subfamily T, member 1
- potassium channel, subfamily T, member 1
- SLACK
- Slo2.2

References

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