LGI1 Gene

Subjects: Genetics & Heredity
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Leucine rich glioma inactivated 1

Keywords: genes

1. Introduction

The *LGI1* gene provides instructions for making a protein called leucine-rich glioma inactivated 1 (Lgi1) or epitempin. This protein is found primarily in nerve cells (neurons) in the brain, including a part of the brain called the lateral temporal lobe. The temporal lobe of the brain is involved in hearing, speech, memory, and emotion.

Although researchers have proposed several functions for epitempin, its precise role in the brain remains uncertain. This protein is probably involved in normal brain development.

Some studies have suggested that epitempin plays a role in the normal function of potassium channels in neurons. These channels are embedded in the cell membrane, where they transport charged potassium atoms (potassium ions) out of neurons. Potassium channels are critical for normal electrical signaling in these cells. Other studies have found that epitempin is transported (secreted) out of neurons. The function of this protein outside cells is unclear.

Epitempin may also help regulate the communication between neurons. Researchers have determined that epitempin attaches (binds) to a receptor protein called ADAM22 on the surface of neurons. Together, these proteins help control the release of certain brain chemicals called neurotransmitters. These chemicals allow neighboring neurons to communicate with each other, which is how signals are relayed throughout the brain.

2. Health Conditions Related to Genetic Changes

2.1. Autosomal Dominant Partial Epilepsy with Auditory Features

At least 22 mutations in the *LGI1* gene have been identified in people with autosomal dominant partial epilepsy with auditory features (ADPEAF). Some *LGI1* mutations change a single protein building block (amino acid) in the epitempin protein, which alters the protein's structure. Other mutations lead to the production of an abnormally short, nonfunctional version of the protein. Researchers suspect that the altered protein is unable to be secreted, which would leave it trapped within cells and unable to perform its usual functions. Although *LGI1* mutations disrupt the function of epitempin, it is unclear how the altered protein leads to seizure activity in the brain.

2.2. Cancers

When the *LGI1* gene was first described, researchers believed that it might play a role in the growth and progression of brain tumors called gliomas. Epitempin was thought to act as a tumor suppressor, which is a protein that keeps cells from growing and dividing too fast or in an uncontrolled way. More recent studies have called into question the role of epitempin in cancerous tumors. Because no *LGI1* mutations have been identified in gliomas and people with ADPEAF do not appear to have a greatly increased risk of these tumors, it now appears unlikely that epitempin functions as a tumor suppressor.

3. Other Names for This Gene

- EPITEMPIN
- Epitempin 1
- EPT

- ETL1
- IB1099
- Leucine-Rich Glioma-Inactivated Protein 1
- leucine-rich, glioma inactivated 1
- LGI1_HUMAN

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