KCNE1 Gene

Subjects: Genetics & Heredity
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Potassium voltage-gated channel subfamily E regulatory subunit 1

genes

1. Introduction

The *KCNE1* gene provides instructions for making a protein that regulates the activity of potassium channels. These channels, which transport positively charged potassium atoms (ions) 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. The KCNE1 protein regulates a channel made up of four parts, called alpha subunits, which are produced from the *KCNQ1* gene. One beta subunit, produced from the *KCNE1* gene, binds to the channel and regulates its activity.

These channels are active in the inner ear and in heart (cardiac) muscle, where they transport potassium ions out of cells. In the inner ear, the channels play a role in maintaining the proper ion balance needed for normal hearing. In the heart, the channels are involved in recharging the cardiac muscle after each heartbeat to maintain a regular rhythm. The KCNE1 protein is also produced in the kidneys, testes, and uterus, where it probably regulates the activity of other channels.

2. Health Conditions Related to Genetic Changes

2.1. Jervell and Lange-Nielsen Syndrome

At least four mutations in the *KCNE1* gene have been identified in people with Jervell and Lange-Nielsen syndrome, a condition that causes an abnormal heart rhythm (arrhythmia) and profound hearing loss from birth. About 10 percent of cases are caused by mutations in this gene. Affected people typically have mutations in both copies of the *KCNE1* gene in each cell. These mutations change a single protein building block (amino acid) in the KCNE1 protein, which disrupts the protein's normal structure. An altered KCNE1 protein cannot regulate the flow of potassium ions through channels in the inner ear and cardiac muscle. This loss of channel function leads to the arrhythmia and hearing loss characteristic of Jervell and Lange-Nielsen syndrome.

2.2. Other Disorders

Certain drugs, including medications used to treat arrhythmias, infections, seizures, and psychotic disorders, can lead to an abnormal heart rhythm in some people. This drug-induced heart condition, which is known as acquired long QT syndrome, increases the risk of cardiac arrest and sudden death. A small percentage of cases of acquired long QT syndrome occur in people who have an underlying mutation in the *KCNE1* gene.

3. Other Names for This Gene

- delayed rectifier potassium channel subunit IsK
- IKs producing slow voltage-gated potassium channel beta subunit Mink
- ISK
- JLNS2
- KCNE1_HUMAN
- LQT5
- minimal potassium channel
- minK
- potassium channel, voltage gated subfamily E regulatory beta subunit 1
- potassium voltage-gated channel, Isk-related family, member 1

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