KCNJ1 Gene

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Potassium voltage-gated channel subfamily J member 1

genes

1. Introduction

The *KCNJ1* gene belongs to a large family of genes that produce 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 KCNJ1 protein, also known as ROMK, are predominantly found in the kidneys. ROMK is one of several proteins that work together to regulate the movement of ions into and out of kidney cells. In particular, the transport of potassium ions by ROMK is necessary for the normal function of another ion transporter called NKCC2 (which is produced from the *SLC12A1* gene). This transporter plays an essential role in the reabsorption of salt (sodium chloride or NaCl) from the urine back into the bloodstream. The retention of salt affects the body's fluid levels and helps maintain blood pressure.

2. Health Conditions Related to Genetic Changes

2.1. Bartter Syndrome

Several dozen mutations in the *KCNJ1* gene have been identified in people with Bartter syndrome type II. This form of the disorder causes severe or life-threatening health problems that become apparent before or soon after birth.

Some of the *KCNJ1* gene mutations responsible for Bartter syndrome change single protein building blocks (amino acids) in the ROMK protein. These mutations prevent the protein from reaching the cell membrane or alter the channel's ability to transport potassium ions. Other mutations in the *KCNJ1* gene delete amino acids from the protein or lead to the production of an abnormally short, nonfunctional version of ROMK.

A loss of functional ROMK affects the normal activity of the NKCC2 protein, preventing it from transporting ions into kidney cells. As a result, the kidneys cannot reabsorb salt normally and excess salt is lost through the urine (salt

wasting). The abnormal salt loss disrupts the normal balance of sodium, potassium, and other ions in the body. These imbalances underlie the major features of Bartter syndrome.

2.2. Other Disorders

Studies suggest that normal variants (polymorphisms) in the *KCNJ1* gene may help explain variations in blood pressure seen in different people. Certain rare polymorphisms appear to protect against high blood pressure (hypertension), and researchers speculate that other genetic variants might increase the risk of high blood pressure. Changes in the *KCNJ1* gene may affect blood pressure by altering the kidneys' ability to reabsorb salt into the bloodstream.

3. Other Names for This Gene

- ATP-regulated potassium channel ROM-K
- ATP-sensitive inward rectifier potassium channel 1
- inward rectifier K(+) channel Kir1.1
- inwardly rectifying K+ channel
- IRK1_HUMAN
- KIR1.1
- potassium channel, inwardly rectifying subfamily J member 1
- potassium channel, inwardly rectifying subfamily J, member 1
- potassium inwardly-rectifying channel, subfamily J, member 1
- ROMK
- ROMK1

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