CLCNKA Gene

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chloride voltage-gated channel Ka

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1. Normal Function

The *CLCNKA* gene belongs to the CLC family of genes, which provide instructions for making chloride channels. These channels, which transport negatively charged chlorine atoms (chloride ions), play a key role in a cell's ability to generate and transmit electrical signals. Some CLC channels regulate the flow of chloride ions across cell membranes, while others transport chloride ions within cells.

The *CLCNKA* gene provides instructions for making a chloride channel called ClC-Ka. These channels are found predominantly in the kidneys. ClC-Ka is one of several proteins that work together to regulate the movement of ions into and out of kidney cells. The transport of chloride ions by ClC-Ka channels is part of the mechanism by which the kidneys reabsorb 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.

ClC-Ka channels are also located in the inner ear, where they play a role in normal hearing.

2. Health Conditions Related to Genetic Changes

2.1. Bartter syndrome

Several people with Bartter syndrome have had mutations in both the *CLCNKA* gene and a closely related gene called *CLCNKB*. The *CLCNKB* gene provides instructions for making a very similar chloride channel, ClC-Kb, that is also found in the kidneys and inner ear. A combination of *CLCNKA* and *CLCNKB* gene mutations causes a life-threatening form of the disorder called Bartter syndrome type IV. This condition is also known as antenatal Bartter syndrome with sensorineural deafness because affected individuals have hearing loss caused by abnormalities in the inner ear.

Mutations in the *CLCNKA* and *CLCNKB* genes prevent the ClC-Ka and ClC-Kb channels from transporting chloride ions in the kidneys. 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 ions in the body. This imbalance underlies many of the major features of Bartter syndrome, including a failure to grow and gain weight at the expected rate (failure to thrive), dehydration, constipation, and increased urine production (polyuria). A loss of ClC-Ka and ClC-Kb function in the inner ear is responsible for the hearing loss characteristic of Bartter syndrome type IV.

2.2. Other Disorders

Studies suggest that several normal variants (polymorphisms) in the *CLCNKA* gene may be associated with salt-sensitive hypertension, a form of high blood pressure related to increased levels of salt in the blood. However, this association between *CLCNKA* polymorphisms and hypertension has not been confirmed. Changes in the *CLCNKA* gene may affect blood pressure by altering the kidneys' ability to reabsorb salt into the bloodstream.

3. Other Names for This Gene

- chloride channel Ka
- chloride channel protein ClC-Ka
- chloride channel, kidney, A
- chloride channel, voltage-sensitive Ka

- ClC-K1
- CLCK1
- CLCKA_HUMAN
- hClC-Ka

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