

# KCNQ3 Gene

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Potassium voltage-gated channel subfamily Q member 3

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## 1. Introduction

The *KCNQ3* 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 *KCNQ3* protein are active in nerve cells (neurons) in the brain, where they transport potassium ions out of cells. These channels transmit a particular type of electrical signal called the M-current, which prevents the neuron from continuing to send signals to other neurons. The M-current ensures that the neuron is not constantly active, or excitable.

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 *KCNQ3* gene can form a channel. However, the *KCNQ3* alpha subunits can also interact with alpha subunits from the *KCNQ2* gene to form a functional potassium channel, and these channels transmit a much stronger M-current.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Benign Familial Neonatal Seizures

A mutation in the *KCNQ3* gene has been identified in some people with benign familial neonatal seizures (BFNS), a condition characterized by recurrent seizures in newborn babies. The seizures begin around day 3 of life and usually go away within 1 to 4 months. At least three mutations have been identified in people with this condition, and these mutations change single protein building blocks (amino acids) in the *KCNQ3* protein. As a result of these mutations, the M-current is reduced. Researchers believe that a reduction of the current by 25 percent is enough to cause BFNS. A reduced M-current leads to excessive excitability of neurons, which is known to cause seizures. It is unclear why the seizures stop around the age of 4 months. It has been suggested that potassium channels formed from the *KCNQ2* and *KCNQ3* proteins play a major role in preventing excessive excitability of neurons in newborns, but other mechanisms develop during infancy.

## 3. Other Names for This Gene

- BFNC2
- EBN2
- KCNQ3\_HUMAN
- KV7.3
- potassium channel subunit alpha KvLQT3
- potassium channel, voltage gated KQT-like subfamily Q, member 3
- potassium channel, voltage-gated, subfamily Q, member 3
- potassium voltage-gated channel subfamily KQT member 3

- potassium voltage-gated channel, KQT-like subfamily, member 3
- voltage-gated potassium channel subunit Kv7.3

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