

# KCNQ4 Gene

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## 1. Introduction

The *KCNQ4* gene provides instructions for making a protein that is part of a family of potassium channels. These channels transport positively charged potassium atoms (potassium ions) between neighboring cells. The channels play a key role in the ability of cells to generate and transmit electrical signals. The specific function of a potassium channel depends on its protein components and its location in the body. Potassium channels made with the KCNQ4 protein are found in certain cells of the inner ear and along part of the nerve pathway from the ear to the brain (the auditory pathway). To a lesser extent, KCNQ4 potassium channels are also found in the heart and some other muscles.

Because KCNQ4 potassium channels are present in the inner ear and auditory pathway, researchers have focused on their role in hearing. Hearing requires the conversion of sound waves to electrical nerve signals, which are then transmitted to the brain. This conversion involves many processes, including maintenance of the proper levels of potassium ions in the inner ear. KCNQ4 channels help to maintain these levels, playing a critical role in the efficient transmission of electrical nerve signals from the inner ear to the brain.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Nonsyndromic Hearing loss

Several *KCNQ4* gene mutations have been reported in individuals with nonsyndromic hearing loss, which is loss of hearing that is not associated with other signs and symptoms. Mutations in this gene cause a form of nonsyndromic hearing loss called DFNA2. This form of hearing loss generally begins after a child learns to speak (postlingual) and particularly affects the ability to hear high-frequency sounds. DFNA2 is described as progressive, which means it becomes more severe over time.

Most *KCNQ4* gene mutations change one of the building blocks (amino acids) used to make the KCNQ4 protein. Some mutations prevent the channel from reaching the cell membrane, where it is needed to transport potassium ions. Other mutations lead to the formation of abnormal channels that cannot transport these ions effectively. The loss of functional KCNQ4 channels appears to cause a buildup of potassium ions in certain cells of the inner ear, which damages those cells and leads to progressive hearing loss in people with DFNA2.

## 3. Other Names for This Gene

- DFNA2
  - KCNQ4\_HUMAN
  - KQT-like 4
  - KV7.4
  - potassium channel, voltage gated KQT-like subfamily Q, member 4
  - potassium voltage-gated channel, KQT-like subfamily, member 4
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