We have over 20,000 different genes in the body. These genes are like instruction manuals for how to build a protein, and each protein has an important function that helps to keep our body working how it should. The *KCNQ1* gene makes a protein called the KCNQ1 protein that helps to build potassium channels in the body. These potassium channels allow cells to make and transmit signals which help to recharge the heart after it beats, which in turn helps to keep the heart beat at a normal rhythm. The potassium channels that are made by the *KCNQ1* gene are primarily in the heart muscles and the tissue in the inner ear (which helps with our hearing).

If someone has a harmful change (called a pathogenic variant) in one of their *KCNQ1* genes, then their body is not going to make enough of the KCNQ1 protein as it should. If there is not enough KCNQ1 protein, then there will not be enough potassium channels. This would mean that the heart's ability to send these signals is not going to work as well as it should. This can lead to several different types of health concerns, including Long QT syndrome and familial atrial fibrillation.

Pathogenic variants in the *KCNQ1* gene are passed through a family in an autosomal dominant pattern, meaning that anyone who carries the variant has a 50% chance to pass it down to any children they have. Women and men both have the *KCNQ1* gene and have the same chances to inherit and pass down pathogenic variants.

If someone inherits a pathogenic variant from BOTH of their parents, they have zero working *KCNQ1* genes. This can cause an autosomal recessive condition called Jervell and Lange-Nielsen syndrome, which causes Long QT syndrome and profound hearing loss from birth. Ninety percent of cases of Jervell and Lange-Nielsen syndrome are caused by non working *KCNQ1*, while the rest are caused by non working *KCNH2* genes.

Genetic Testing for *KCNQ1*

Genetic testing for pathogenic variants in *KCNQ1* is currently available, but there are a few different ways to approach testing:

- *Single site analysis*: Testing specific to a known pathogenic variant in the family
- Full gene sequencing and rearrangement analysis: Comprehensive testing to search for all currently detectable pathogenic variants in the gene
- Gene panels: Newer, more broadly based gene tests that would include not only the *KCNQ1* gene, but other genes known or suspected to be associated with hereditary cardiovascular disease.

Click here to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.