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 *RYR2* gene makes a protein called ryanodine receptor 2. The ryanodine receptor 2 protein helps to form channels to move calcium between cells. These calcium channels allow cells to make and transmit signals which help to make the heart beat at a normal rhythm. The calcium channels that are made by the *RYR2* gene are primarily in the heart muscles.

If someone has a harmful change (called a pathogenic variant) in one of their *RYR2* genes, then their body is not going to make enough of the ryanodine receptor 2 protein as it should. If there is not enough ryanodine receptor 2 protein, then there will not be enough calcium 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 a heart condition called catecholaminergic polymorphic ventricular tachycardia.

Pathogenic variants in the *RYR2* 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 *RYR2* gene and have the same chances to inherit and pass down pathogenic variants.

Genetic Testing for *RYR2*

Genetic testing for pathogenic variants in *RYR2* 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 *RYR2* 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.