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 CASQ2 gene makes a protein called calsequestrin 2. The calsequestrin 2 protein helps to store calcium. The amount of calcium in the cells of our heart controls when our heart muscles contract and relax, which make the heart beat at a normal rhythm. The calcium that is stored by the CASQ2 gene is primarily in the heart muscles.

If someone has a harmful change (called a pathogenic variant) in one of their CASQ2 genes, then their body is not going to make enough of the calsequestrin 2 protein as it should. If there is not enough calsequestrin 2 protein, then there will not be enough stored calcium. This would mean that the heart’s ability to contract and relax (and thus beat) at a normal rate 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 CASQ2 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 CASQ2 gene and have the same chances to inherit and pass down pathogenic variants.

**Genetic Testing for CASQ2**

Genetic testing for pathogenic variants in CASQ2 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 CASQ2 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.