



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.