



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 gene *CACNA1S* makes a protein called the CACNA1S protein. This protein helps to make calcium channels, which move calcium in and out of cells. Calcium is involved in the contraction of the muscles; as calcium levels increase, the muscle contracts. The calcium channels are an important part of that process.

If there is a harmful error (called a pathogenic variant) in the *CACNA1S* gene, then the body may not be able to make as many calcium channels as it needs to function how it should. If there are not enough calcium channels, this can lead to increased levels of calcium in the muscle cells. This can prevent the muscle from contracting normally, and can cause various different health problems.

Pathogenic variants in the *CACNA1S* gene are thought to cause approximately 1% of all cases of [malignant hyperthermia susceptibility](#) (MHS), which can include a potentially life-threatening sensitivity to certain medications. At least four specific pathogenic variants in the *CACNA1S* gene have been found to cause hypokalemic periodic paralysis, which causes periods of extreme muscle weakness starting in childhood or adolescence.

Pathogenic variants in the *CACNA1S* 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 *CACNA1S* gene and have the same chances to inherit and pass down pathogenic variants.

Genetic Testing for *CACNA1S*

Genetic testing for pathogenic variants in *CACNA1S* 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 *CACNA1S* gene, but other genes known or suspected to be associated with health concerns like those that can be related to *CACNA1S*.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.