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 *ACTC1* gene makes a protein called the cardiac muscle alpha actin (CMAA) protein. The CMAA protein is found primarily in the heart muscles in our body. The CMAA protein works with other proteins to create the force that is needed for our heart muscles to contract. This muscle contraction is how our heart pumps blood throughout our bodies.

If someone has a harmful change (called a pathogenic variant) in one of their *ACTC1* genes, then their body does not make as much CMAA protein as it should. If there is not enough CMAA protein, then the heart muscles cannot contract as well as they should. This causes damage to these muscles, which can lead to several different types of health issues, such as familial hypertrophic cardiomyopathy, left ventricular noncompaction, and familial dilated cardiomyopathy.

Pathogenic variants in the *ACTC1* 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 *ACTC1* gene and have the same chances to inherit and pass down pathogenic variants.

Genetic Testing for *ACTC1*

Genetic testing for pathogenic variants in *ACTC1* 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 *ACTC1* 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.