

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 MYBPC3 gene makes a protein called cardiac myosin binding protein C (MyBP-C). Our muscles, including our heart muscles, are made up of different fibers that help to control when our muscles contract. This process is necessary in our heart, because these contractions are how our heart pumps blood throughout our bodies. The cardiac MyBP-C protein works to keep these fibers in good working order so they can make sure our heart muscles are contracting how they should.

If someone has a harmful change (called a pathogenic variant) in one of their MYBPC3 genes, then their body does not make as much MyBP-C protein as it should. If there is not enough MyBP-C protein, then the fibers that control our heart contractions may get damaged. This damage is what can lead to several different types of health issues, including left ventricular noncompaction, familial hypertrophic cardiomyopathy, and familial dilated cardiomyopathy.

Pathogenic variants in the MYBPC3 gene are passed through the family in an <u>autosomal</u> dominant pattern, meaning that anyone who carries a pathogenic variant has a 50% chance to pass it down to any children they have. Women and men both have the MYBPC3 gene and have the same chances to inherit and pass down pathogenic variants.

Genetic Testing for MYBPC3

Genetic testing for pathogenic variants in *MYBPC3* is currently available, but there are a few different ways to approach testing:

- <u>Single site analysis</u>: Testing specific to a known pathogenic variant in the family
- Full gene <u>sequencing</u> and <u>rearrangement analysis</u>: Comprehensive testing to search for all currently detectable variants in the gene
- Gene panels: Newer, more broadly based gene tests that would include not only the MYBPC3 gene, but other genes known or suspected to be associated with hereditary heart disease.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.