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 *MYH11* gene makes a protein called smooth muscle myosin heavy chain 11 (MYH11 protein). The MYH11 protein is found primarily in our smooth muscles (muscles that line our blood vessels, stomach, intestines, and other internal organs). Smooth muscles relax and contract automatically as part of their normal function, and the MYH11 protein works with other proteins to help the smooth muscles contract, which helps them to keep their shape instead of stretching out too much while blood is pumping through.

If someone has a harmful change (called a pathogenic variant) in one of their *MYH11* genes, then their body does not make as much MYH11 protein as it should. If there is not enough MYH11 protein, then the smooth muscles cannot contract as well as they should. This causes damage to these muscles, which can lead to health issues like familial thoracic aortic aneurysm and dissection along with a congenital (from birth) heart defect called patent ductus arteriosus.

Pathogenic variants in the *MYH11* 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 *MYH11* gene and have the same chances to inherit and pass down pathogenic variants.

Genetic Testing for *MYH11*

Genetic testing for pathogenic variants in *MYH11* 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 *MYH11* 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.