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 *COL3A1* gene makes a protein called type 3 collagen. Type 3 collagen works to strengthen and support many of the tissues in our bodies (primarily in the skin, lungs, intestines, and walls of our blood vessels).

If someone has a harmful change (called a pathogenic variant) in one of their *COL3A1* genes, then their body does not make as much type 3 collagen as it should. Without enough type 3 collagen, these tissues in our bodies are not as strong and stable as they should be. This can lead different health conditions, such as the vascular form of Ehlers-Danlos syndrome, or familial thoracic aortic aneurysm and dissection.

Pathogenic variants in the *COL3A1* 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 *COL3A1* gene and have the same chances to inherit and pass down pathogenic variants.

## Genetic Testing for *COL3A1*

Genetic testing for pathogenic variants in *COL3A1* 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 *COL3A1* gene, but other genes known or suspected to be associated with cardiovascular disease or Ehlers-Danlos syndrome.

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