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 *TMEM43* gene makes a protein called a nuclear envelope protein. Our body is made up of billions of cells, and there are many parts of the cell. The 'mothership' of the cell is called the nucleus. The nucleus holds our DNA and helps to protect and regulate it. Nuclear envelope proteins surround the nucleus to help protect it, and to monitor different things that travel in and out of the nucleus.

If someone has a harmful change (called a pathogenic variant) in one of their *TMEM43* genes, then their body does not make as much nuclear envelope protein as it should. If there is not enough nuclear envelope protein, then the nucleus is not as well protected. This leads to a higher rate of cell death and damage because the nucleus of the cells are more vulnerable. This progressive cell death can cause damage to the muscles, which can lead to different types of health issues, such as arrhythmogenic right ventricular cardiomyopathy or Emery-Dreifuss muscular dystrophy.

Pathogenic variants in the *TMEM43* gene are passed through the 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 *TMEM43* gene and have the same chances to inherit and pass down pathogenic variants.

Genetic Testing for *TMEM43*

Genetic testing for pathogenic variants in *TMEM43* 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 *TMEM43* 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.