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 *PRKAG2* gene makes part of a protein called AMP-activated protein kinase (AMPK). The AMPK protein works in many parts of our bodies, particularly in the heart and skeletal muscles (which help with movement), and monitors if our cells need more energy. Our cells break down a molecule called adenosine triphosphate (ATP), and that is what they use for energy to keep functioning how they should. The AMPK protein helps to control the amount of ATP is broken down to make sure the cell has enough energy to do its job.

If someone has a harmful change (called a pathogenic variant) in one of their *PRKAG2* genes, then their body does not make as much AMPK protein as it should. If there is not enough AMPK protein, then the amount of ATP that is available to the cells for energy is not as high as it should be. This can lead to the cells not being able to perform their functions, which can lead to several different types of health issues, such as familial hypertrophic cardiomyopathy, Wolff-Parkinson-White syndrome, and familial atrial fibrillation.

Pathogenic variants in the *PRKAG2* 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 *PRKAG2* gene and have the same chances to inherit and pass down pathogenic variants.

## Genetic Testing for *PRKAG2*

Genetic testing for pathogenic variants in *PRKAG2* 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 *PRKAG2* 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.