PRKAG2

The *PRKAG2* gene is one of many genes that helps muscles tense up (contract). The *PRKAG2* gene makes a protein that works with other proteins to help sense and respond to energy demands within cells, especially in the heart muscle and in the muscles that allow the body to move (skeletal muscle). When this protein doesn't work properly, it decreases the heart's ability to pump blood to the rest of the body.

Impact of *PRKAG2* mutations
Individuals with a mutation in the *PRKAG2* gene are at an increased risk for developing hypertrophic cardiomyopathy, a hereditary cardiovascular (heart and blood vessel) disorder, which can affect the heart's ability to pump blood.

Disorders associated with the *PRKAG2* gene

Mutations in the *PRKAG2* gene have been associated with the following disorder:

Hypertrophic Cardiomyopathy
Hypertrophic cardiomyopathy (HCM) is associated with an abnormal thickening of the heart muscle, which can make it hard for the heart to pump blood.

HCM is associated with an abnormal thickening (hypertrophy) of the heart muscle in the major pumping chamber of the heart, called the left ventricle. This means blood is pumped out of the heart less efficiently and blood flow may even be blocked in some individuals. Symptoms of HCM may include fatigue, shortness of breath with exertion, pounding sensations in the heart (palpitations), light-headedness, dizziness or fainting.

The majority of people with HCM will have mild symptoms and a normal life expectancy. However, in some cases, symptoms of HCM can be life-threatening. People with HCM can have a problem with the electrical system of the heart that controls the heartbeat's regular rhythm (arrhythmias), which can increase the risk of sudden cardiac death. Individuals can also develop heart failure that is potentially fatal if untreated. Age of onset and severity of symptoms may vary, even within the same family.

Diagnosing HCM typically involves evaluating an individual's medical and family histories, as well as a regular physical exam, an imaging test used to see whether the heart muscle is abnormally thick (echocardiogram), and a test of the heart's electrical system called an electrocardiogram (EKG or ECG). Additional screening and diagnostic tests may be ordered, including an MRI.

Individuals with HCM are advised to make certain lifestyle changes, such as staying well-hydrated and avoiding strenuous exercise and certain medications. Depending on whether

HCM symptoms are present, medications that help control blood pressure may be prescribed. Antibiotics may be prescribed before certain medical and dental procedures to guard against infections in the heart. Some individuals may also need a device that detects a dangerously fast heart rhythm and delivers a shock to correct it called an implantable cardioverter defibrillator (ICD) or other surgical procedures. If medications and surgical procedures are not working to manage heart failure, a heart transplantation may be considered.

Regular visits to a cardiologist specializing in HCM are recommended in order to check that treatment is effective.

Useful resources

American Heart Association
Focused on building lives free of heart disease by providing accessible education and funding innovative research.
www.heart.org

Hypertrophic Cardiomyopathy Association
Provides support, education, advocacy and advancing research, understanding and care to those with hypertrophic cardiomyopathy.
http://www.4hcm.org

SHARE Registry
Advancing the understanding of cardiomyopathy by increasing community awareness and supporting research.
https://thesharereregistry.org

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