

KCNQ1

The *KCNQ1* gene is one of many genes that helps maintain a regular heartbeat. The *KCNQ1* gene makes a protein whose primary role is to form a potassium channel which generates and transmits electrical charges in the body when needed, such as when the heart beats. When this protein doesn't work properly the heartbeat can be abnormal.

Impact of *KCNQ1* mutations

Individuals with a mutation in the *KCNQ1* gene are at an increased risk for developing different hereditary cardiovascular (heart and blood vessel) disorders called arrhythmias, which can affect the heartbeat's regular rhythm. These include long QT syndrome and short QT syndrome.

Disorders associated with the *KCNQ1* gene

Mutations in the *KCNQ1* gene have been associated with the following disorders:

Long QT Syndrome

Long QT syndrome (LQTS) is a hereditary disorder associated with a problem with the electrical system of the heart that controls the heartbeat's regular rhythm (arrhythmia). LQTS may increase risk of sudden cardiac arrest or sudden cardiac death at young ages.

LQTS is associated with problems in the heart's electrical system in which there is a dangerously fast heart beat in the lower pumping chambers of the heart (torsade de pointes). Symptoms of LQTS may include fainting, seizures, or sudden cardiac arrest. Exercise and heightened or intense emotions may be a trigger for sudden cardiac arrest, which can happen from infancy through middle age. An individual's risk depends on their sex, age, and previous symptoms. The severity of the electrical problem in the heart and the specific gene that causes LQTS also play a role. Some individuals with LQTS experience no noticeable symptoms, but are still at risk for sudden cardiac arrest and death.

Women with LQTS have an increased risk of cardiac arrest or death in the first nine months after giving birth.

Diagnosing LQTS typically involves evaluating an individual's medical and family histories as well as results of a test of the heart's electrical system called an electrocardiogram (EKG or ECG). These evaluations may be combined with genetic testing to diagnose the type of LQTS an individual has.

Treatment is recommended for everyone since there is no way to predict who may have symptoms and who may not. Treatment typically involves taking medications that help moderate your heart's rhythm. 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. Individuals are recommended to avoid certain medications and drugs. Some individuals are advised to avoid strenuous exercise, competitive sports, or exposure to loud noises.

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

Short QT Syndrome

Short QT syndrome (SQTS) is a hereditary disorder associated with a problem with the electrical system of the heart that controls the heartbeat's regular rhythm (arrhythmia). SQTS may increase risk of sudden cardiac arrest or death at young ages.

SQTS is associated with problems in the heart's electrical system that can cause one of two rhythm problems. The first is called atrial fibrillation, in which there is an irregular heartbeat starting in the upper pumping chambers of the heart (atria). This can cause shortness of breath, dizziness, chest tightness, fatigue, and fainting. The second is called ventricular tachycardia or fibrillation, in which there is a very fast heartbeat starting in the lower pumping chambers of the heart (ventricles). This can cause fainting and sudden cardiac death. An individual's risk depends on their sex, age, and previous symptoms. Some individuals with SQTS experience no noticeable symptoms, but are still at risk for sudden cardiac arrest and death.

Diagnosing SQTS typically involves evaluating an individual's medical and family histories as well as results of a test of the heart's electrical system called an electrocardiogram (EKG or ECG). These evaluations may be combined with genetic testing to diagnose SQTS.

Treatment may involve use of a device that detects a dangerously fast heart rhythm and delivers a shock to correct it called an implantable cardioverter defibrillator (ICD). Medication can also be used for treatment.

Regular visits to a cardiologist specializing in SQTS 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

Heart Rhythm Society

Provides information sheets on types of arrhythmias and associated treatments, risk factors, signs and symptoms.

<http://resources.hrsonline.org>

Sudden Arrhythmia Death syndromes (SADS)

SADS advocates for nondiscriminatory treatment for people who are diagnosed with a SADS disorder. SADS is committed to supporting efforts that will improve the quality of life for patients with heart rhythm abnormalities.

www.sads.org

Last updated April 4, 2018