SCN5A

The *SCN5A* gene is one of many genes that helps maintain a regular heartbeat. The *SCN5A* gene makes a protein whose primary role is to form a sodium 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 *SCN5A* mutations
Individuals with a mutation in the *SCN5A* 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 Brugada syndrome and long QT syndrome.

Disorders associated with the *SCN5A* gene

Mutations in the *SCN5A* gene have been associated with the following disorders:

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

Brugada syndrome is associated with problems in the heart's electrical system in which there is a dangerously fast and chaotic heartbeat called ventricular fibrillation (VFib). Symptoms of Brugada syndrome may include fainting or sudden cardiac arrest. These can happen from infancy through middle age. An individual's risk depends on their sex, age, and previous symptoms. Some individuals with Brugada syndrome experience no noticeable symptoms, but are still at risk for sudden cardiac arrest and death.

Diagnosing Brugada syndrome 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), and a test that evaluates your heart's function during exercise (stress test). These evaluations may be combined with genetic testing to diagnose Brugada syndrome.

Treatment for people with symptoms 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) or other surgical procedures. Medication can also be used for treatment. Individuals are advised to avoid certain medications and drugs. Illnesses that include fever and electrolyte imbalance caused by dehydration are recommended to be treated promptly as they can increase risk. Individuals are advised to avoid excessive alcohol consumption and large meals.

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

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 control blood pressure. 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.

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

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