

## RYR2

The *RYR2* gene is one of many genes that helps maintain a regular heartbeat. The *RYR2* gene makes a protein whose primary role is to form a calcium 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 *RYR2* mutations

Individuals with a mutation in the *RYR2* gene are at an increased risk for developing catecholaminergic polymorphic ventricular tachycardia (CPVT), a hereditary cardiovascular (heart and blood vessel) disorder, called an arrhythmia, which can affect the heartbeat's regular rhythm.

### Disorders associated with the *RYR2* gene

Mutations in the *RYR2* gene have been associated with the following disorder:

#### Catecholaminergic polymorphic ventricular tachycardia

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is a hereditary disorder associated with a problem with the electrical system of the heart that controls the heartbeat's regular rhythm (arrhythmia). CPVT typically occurs in response to strenuous physical activity or heightened emotions and may increase risk of sudden cardiac arrest or death at young ages.

CPVT is associated with problems in the heart's electrical system in which there is a dangerously fast heartbeat in the lower pumping chambers of the heart (ventricles). Symptoms of CPVT may include fainting or sudden cardiac arrest. Sudden cardiac death can occur, even in individuals who have no other symptoms. Exercise, heightened or intense emotions, or other activities that cause an increase in adrenaline are typically a trigger for sudden cardiac arrest, which can happen from childhood through middle age. An individual's risk depends on their sex, age, and previous symptoms. Some individuals with CPVT experience no noticeable symptoms, but are still at risk for sudden cardiac arrest and death.

Diagnosing CPVT typically involves evaluating an individual's medical and family histories as well as results of a test that evaluates your heart's function during exercise (stress test). These evaluations may be combined with genetic testing to diagnose CPVT.

Treatment typically involves taking medication that helps 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 advised to avoid certain medications, strenuous exercise, competitive sports and stressful environments.

Regular visits to a cardiologist specializing in CPVT 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](http://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](http://www.sads.org)

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