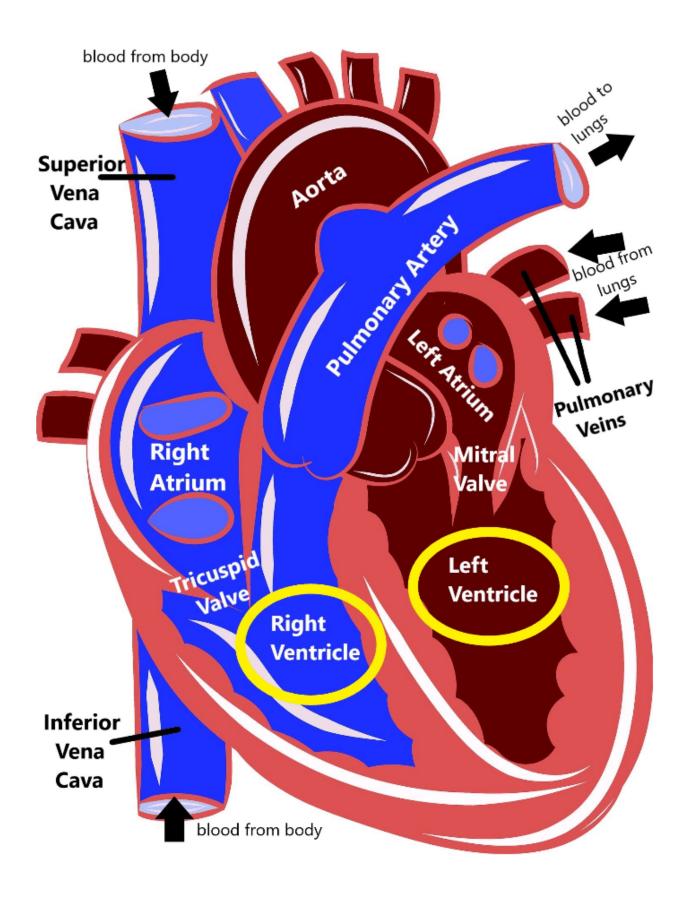




Catecholaminergic polymorphic ventricular tachycardia (CPVT) is a genetic condition that interferes with the heart's normal electrical rhythm (called an <u>arrhythmia</u>) in the lower part of the heart (the ventricles).







This interference makes the ventricles contract more quickly than normal, which speeds up the heart rate (tachycardia). Repeated occurrences of tachycardia increase the chance for heart failure (cardiac arrest) and sudden death.

People who have CPVT generally start to see symptoms between the ages of 7-12 years old. Symptoms of CPVT can include fainting, light-headedness, dizziness, and heart attack. Strenuous physical activity and emotional stress can increase the chance for someone with CPVT to show symptoms.

Causes

Approximately half of all cases of CPVT can be linked to the <u>RYR2</u> gene, and about 1-2% of all cases are linked to the CASQ2 gene. The remaining individuals may have a genetic link to their CPVT, but it is in a gene that we do not yet know about yet.

CPVT is thought to affect approximately 1 in 10,000 people. It is possible that it is more common than this, and that people who are affected that haven't shown any symptoms just don't know that they have it.

Diagnosing CPVT

While genetic testing for CPVT can be helpful to establish a diagnosis, medical providers may also use other medical tests, such as an ECG, or an echocardiogram. Some red flags that can increase the chance for CPVT in the family include:

- Fainting or passing out during physical activity or stressful emotional periods before the age of 40
- History of dizziness or a rapid, irregular heart beat (palpitations) during exercise or periods of emotional stress
- Emotional stress or strenuous activity that triggers sudden unexpected cardiac death
- Ventricular arrhythmias (abnormal heart beat) during exercise
- Ventricular fibrillation (when the lower chambers of the heart guiver instead of beating like normal) during times of acute stress
- A structurally normal heart

Many families may have these red flags in their family history and DO NOT have CPVT. However, someone with a strong pattern of these or other heart issues may be at a higher chance to have CPVT, and may benefit from talking about it more with a specialist, such as a cardiologist or a genetic counselor.



Medical Management for CPVT

Treatment for CVPT can sometimes vary depending on the individual person and their specific health concerns, and should be discussed with a medical provider who is familiar with CVPT. Some options for medical management can include medication, regular monitoring with an ECG, and a surgically-implantable defibrillator.

People who have CVPT should also be careful to avoid competitive sports or other strenuous exercises, as well as significant emotional stress (when possible).

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Additional Resources

Sudden Arrhythmia Death Syndromes (SADS) Foundation

American Heart Association (AHA)