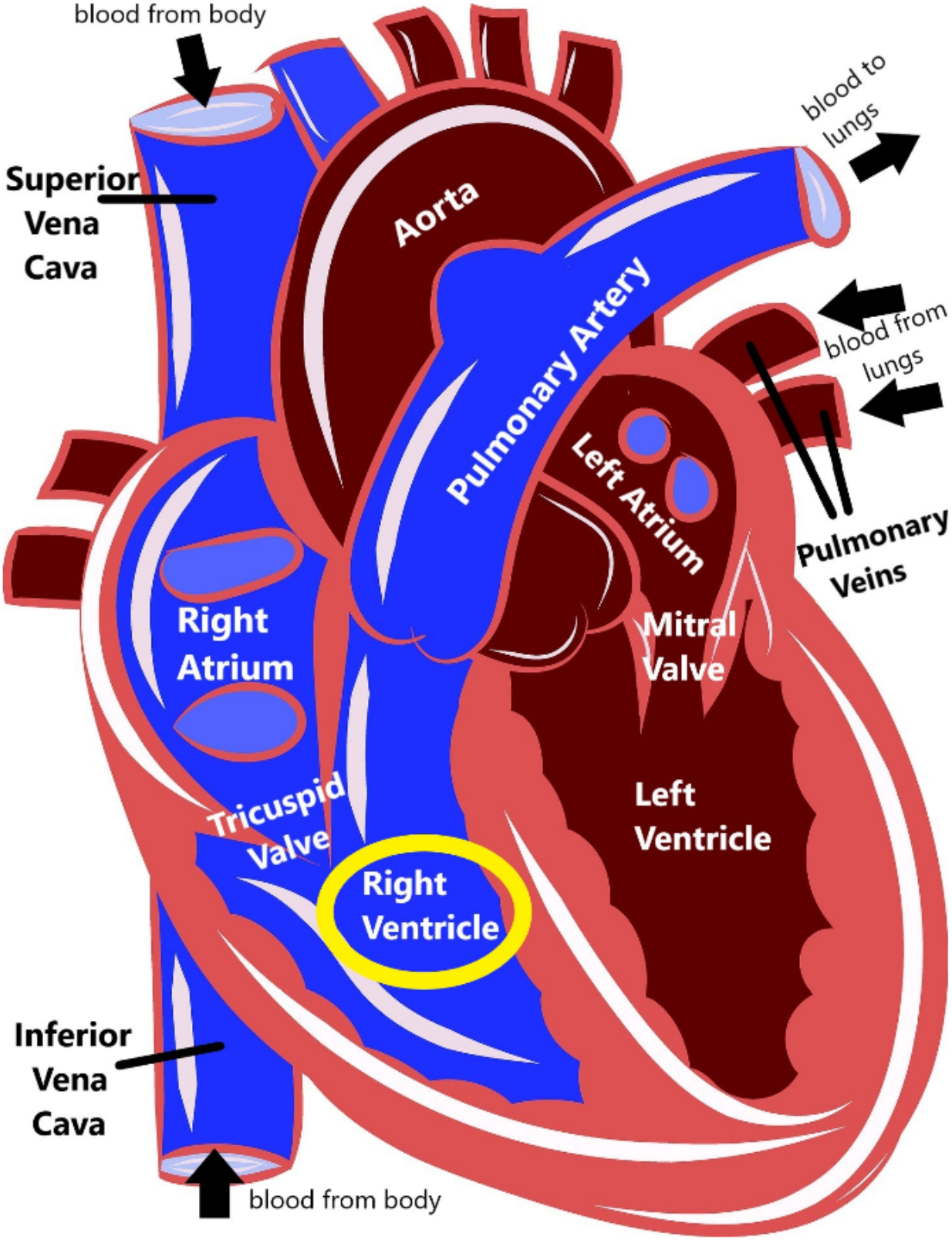
Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a disease that causes the heart muscle to break down, primarily in the right ventricle.


blood from body
blood to lungs
Superior Vena Cava
Aorta
Pulmonary Artery
blood from lungs
Left Atrium
Pulmonary Veins
Right Atrium
Mitral Valve
Tricuspid Valve
Left Ventricle
Right Ventricle
Inferior Vena Cava
blood from body

When this muscle breaks down, there is an increased risk that the heart's electrical system cannot work properly (causing an arrhythmia, or an abnormal heartbeat), which in turn increases the risk for sudden death. Because ARVC affects both the electrical system and the muscle of the heart, it technically fits into both the 'arrhythmia' and 'cardiomyopathy' categories of heart disease.

People who have ARVC may have no symptoms until adulthood, and the first things many individuals with ARVC will notice are heart palpitations, fainting, and feeling light-headed. People who have more advanced ARVC can also develop edema (swelling of the legs or abdomen) and shortness of breath. Over time if the heart muscle develops significant damage it can cause heart failure.

Some individuals who have ARVC may not have any obvious symptoms, but can still be at a higher risk of sudden death, particularly during strenuous exercise. The average age for someone with ARVC to be diagnosed is 31 years old.

Causes

There are many different genes that have been found to cause different types of ARVC in families, some of these include:

- PKP2 (34-74% of all cases of ARVC)
- DSG2 (5-26% of all cases of ARVC)
- DSP (2-39% of all cases of ARVC)
- DSC2 (1-2% of all cases of ARVC)
- TMEM43 (unknown percentage of all cases of ARVC)
- LMNA (unknown percentage of all cases of ARVC)

ARVC is thought to affect between 1 in 1000 to 1 in 1250 of 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 ARVC

While genetic testing for ARVC 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 ARVC in the family include:

- Fainting or passing out

- Heart palpitations
- Cardiac arrest (sudden heart failure)
- Abnormal ECG
- Abnormal right ventricle seen through heart imaging, such as an echocardiogram or cardiac MRI

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

Medical Management for ARVC

Treatment for ARVC 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 ARVC. Medications to help prevent arrhythmias and surgically implanting a defibrillator are some common therapies for ARVC. It is also usually recommended that people who have ARVC avoid regular strenuous activity because it can put more strain on the heart muscles.

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

Additional Resources

ARVC Patient Registry at Johns Hopkins

Sudden Arrhythmia Death Syndromes (SADS) Foundation