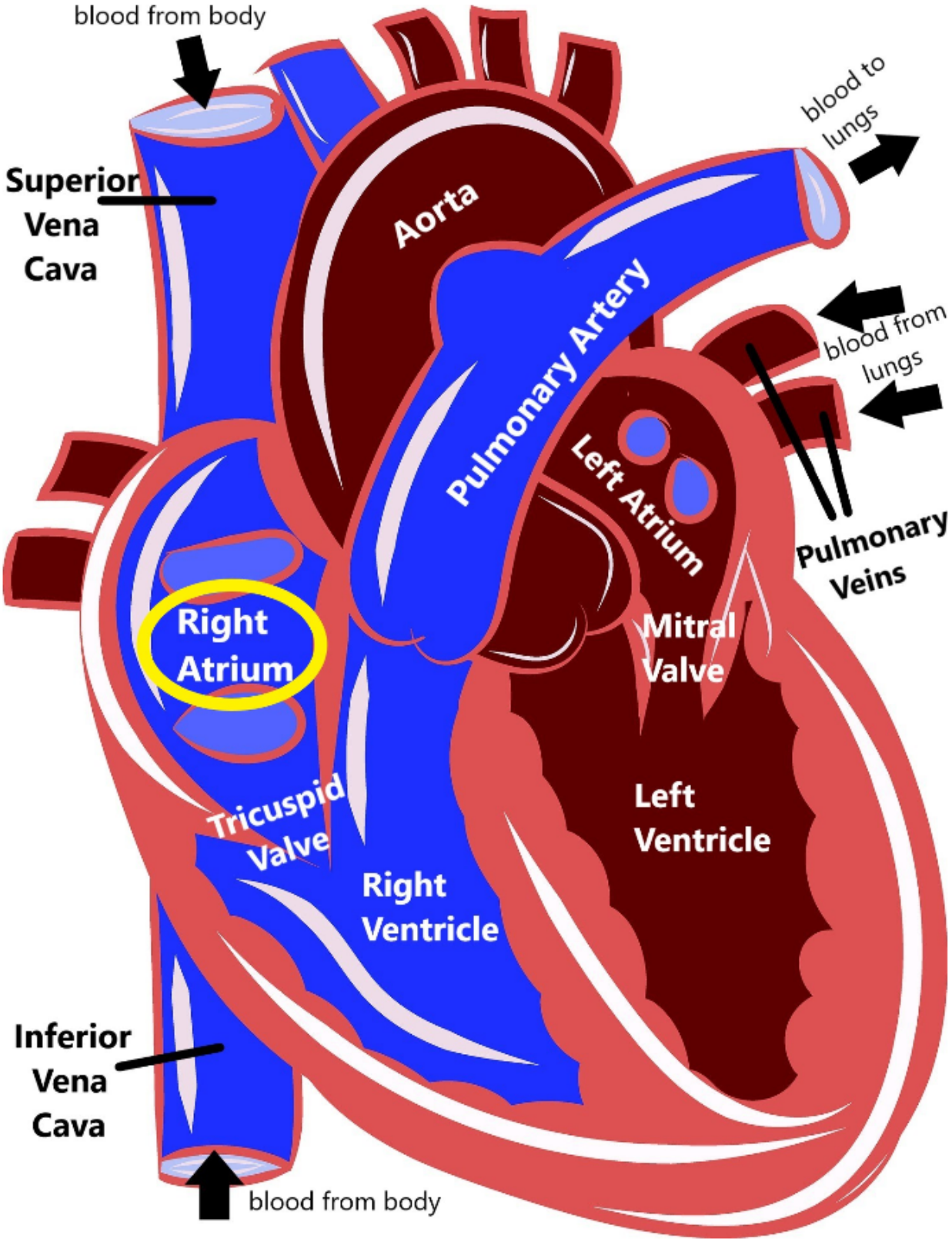
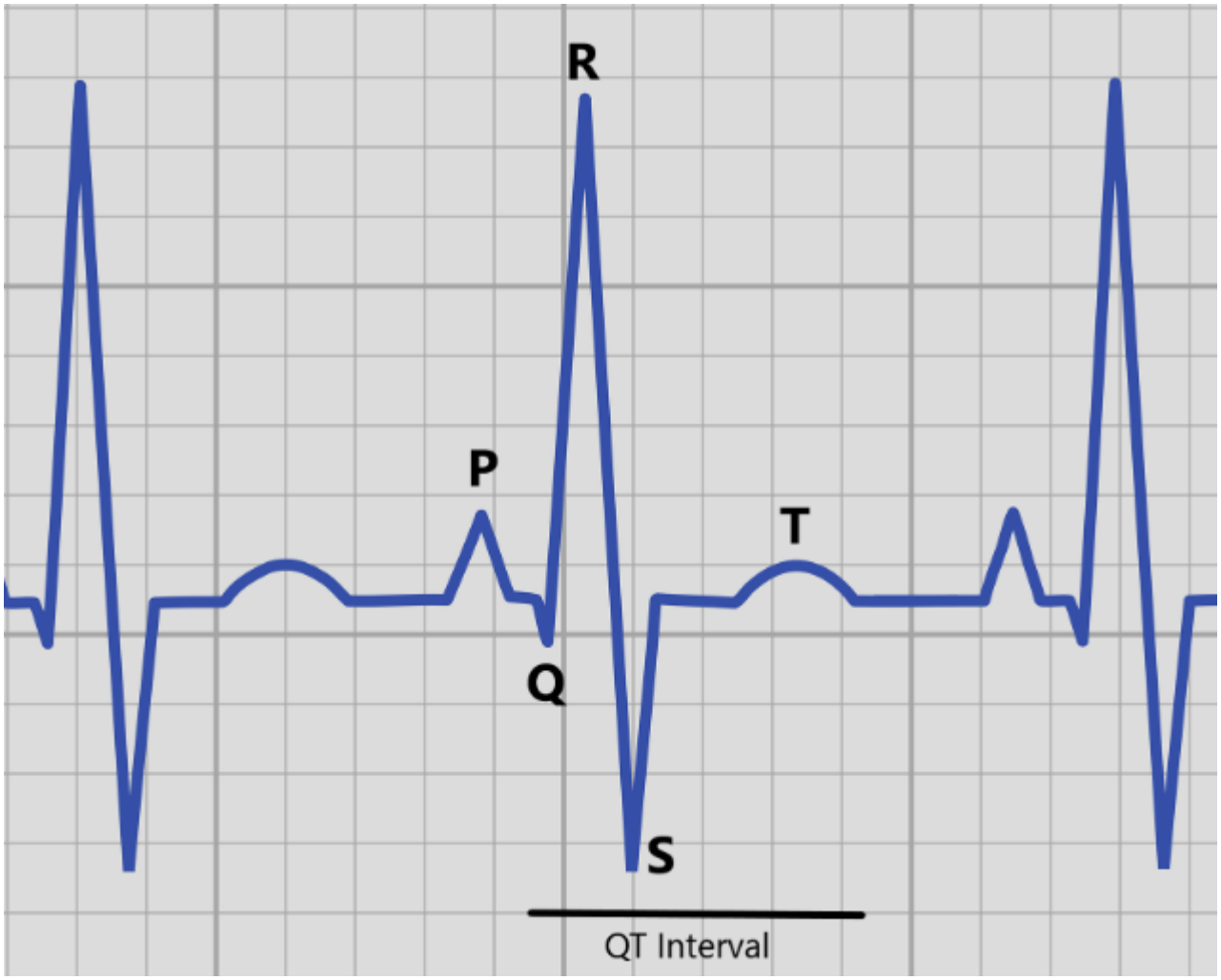
Long QT syndrome (LQTS) is a disorder that can cause erratic and irregular heartbeat. Our heart is controlled by an electrical system that helps determine how fast our heart should beat (your heart rate). It does this by using electrical signals to coordinate the heart muscles to beat together so the heart can pump blood efficiently. If this electrical system does not work properly, then it can cause the heart to beat too fast or too slow, or stop the heart from beating altogether. This electrical signal starts at a part of the heart called the sinoatrial node, which is located in the right atrium of heart.


blood from body
Superior Vena Cava
Aorta
blood to lungs
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

This electrical signal works its way down the heart to signal the muscles to contract together, which lets our heart more efficiently pump blood to the rest of our bodies. A medical device called an echocardiogram can measure this electrical pulse by looking five distinct waves, called P, Q, R, S, and T. The electrical activity in the bottom two chambers of the heart (the ventricles) is measured by looking at the length between the Q and T waves (called the QT interval).



If someone has a QT interval that is too long, that can lead to a longer time in between heartbeats (which explains the name Long QT syndrome). This abnormal heartbeat, called an arrhythmia, can lead to dizziness, lightheadedness, and fainting, or in more serious cases seizures or sudden cardiac death. Some people with LQTS may not have any obvious symptoms, but can still be at a higher risk of sudden death. Two people with LQTS, even within the same family, can have very different signs and symptoms.

Sudden cardiac death can happen anytime from infancy to advanced adulthood, but is more common in the teens and 20s. For about 10-15% of people who have LQTS, sudden death will be the first symptom they have.

Causes and subtypes

There are 15 different subtypes of LQTS that are all caused by different genes. The three most common subtypes make up about 75% of cases of LQTS:

- *KCNQ1*: type 1 (30-35% of all cases of LQTS)
- *KCNH2:* type 2 (25-30% of all cases of LQTS)
- *SCN5A*: type 3 (5-10% of all cases of LQTS)

The risk of sudden death is the same for all three of the common subtypes of LQTS (6-8%). However, the chance to have another symptoms (such as lightheadedness or fainting) is quite different (63% for type 1, 46% for type 2, and 18% for type 3). That means that 37% of people with LQTS type 1, 54% of people with LQTS type 2, and 82% of people with LQTS type 3 will never show any symptoms.

There are also different things that can trigger these health concerns depending on what type of LQTS someone has. Periods of intense emotion or strenuous exercise (particularly swimming) can trigger a heart issue in people with type 1, which people with type 2 can be triggered by emotion (such as excitement or fear), exercise, or during their sleep. Most people with LQTS type 3 will experience related heart issues during their sleep.

The other known 12 subtypes are more rare, and each account for less than 1% of all cases of LQTS. Approximately 20% of families who have LQTS do not have a genetic cause that can be found, which means there are likely other genes that we don't know about yet that cause LQTS in some families. A small number of cases of LQTS can be acquired, or caused by things like medications or other underlying health issues, rather than genetic.

Approximately 1 in 2000 to 1 in 2500 people are thought to have LQTS. 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 LQTS

While genetic testing for LQTS can be helpful to establish a diagnosis, medical providers may also use other medical tests, such as an ECG and your family history. Some red flags

that can increase the chance for LQTS in the family include:

- A history of fainting
- A history of a child or young adult that died suddenly
- A history of arrhythmia

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

Medical Management for LQTS

Treatment for LQTS 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 LQTS. This treatment can include medications (including beta blockers and sodium channel blockers) or surgically implanting a defibrillator.

Medical management is also different depending on what type of LQTS someone has. For example, because people with type 1 are more likely to have a heart issue during activity strenuous activity is not recommended, while people with type 2 should try to avoid loud noises because those startling noises can trigger a heart issue.

Click here 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

International Long QT Syndrome Registry: email heartajm@heart.rochester.edu