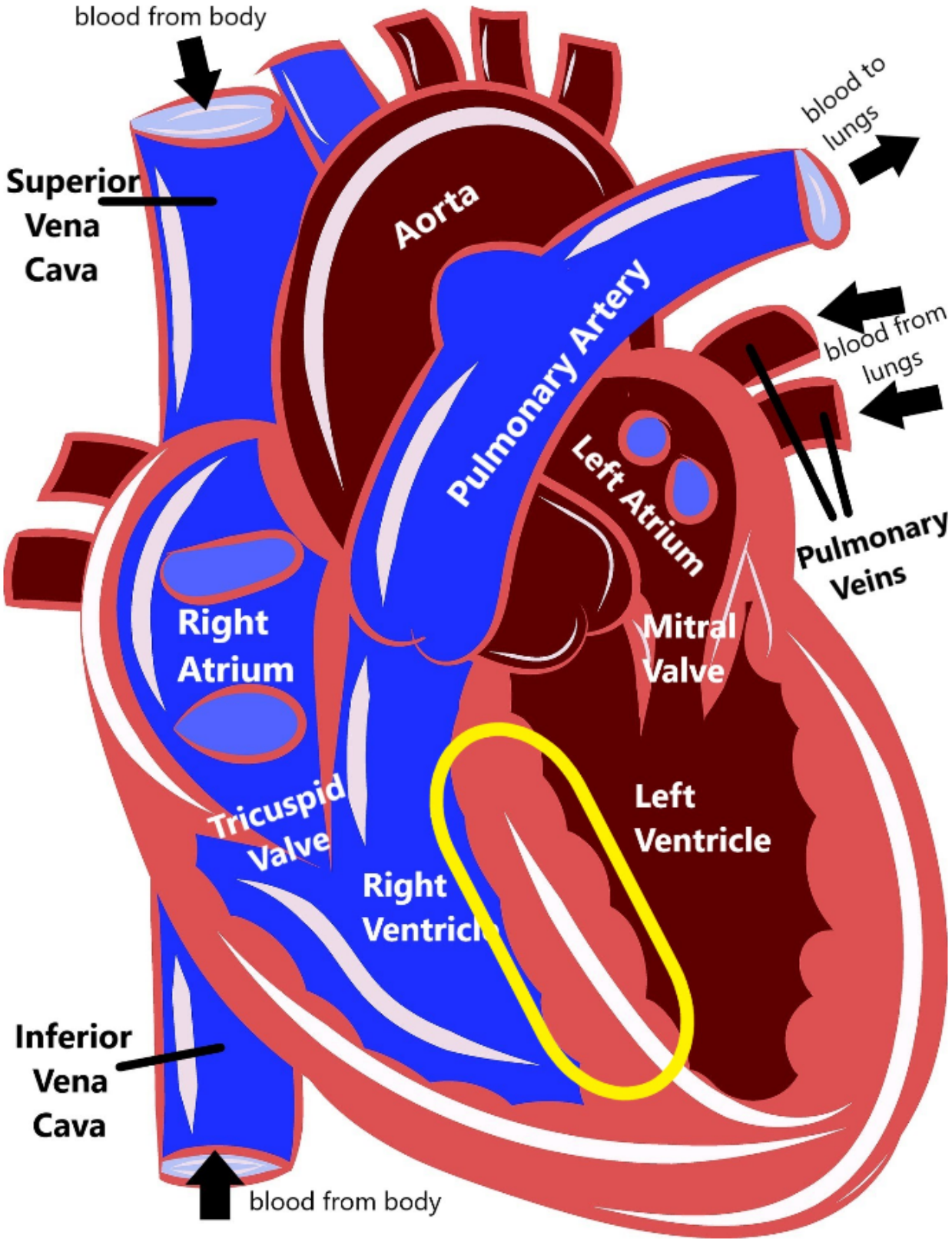
Familial hypertrophic cardiomyopathy (FHCM) is a disease that causes the muscle of the heart to become thicker than normal (called hypertrophy). There are several parts of the heart where the muscle can hypertrophy, but it most frequently happens in the muscle that separates the two bottom chambers of the heart (the ventricles).


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 becomes more thick, it can lead to the heart not being able to pump oxygenated blood around the body. Sometimes this thickened muscle can block the flow of blood out of the heart, which can lead to a heart murmur, or can disrupt the heart's electrical system, causing an arrhythmia. People who have FHCM are more likely to start showing symptoms in their teens to young adulthood, but some people do not have any health issues until later in life.

The specific health issues that affect someone with FHCM can be very different from person to person, even with people in the same family. Some people may begin to notice feeling dizzy or lightheaded, fainting, having chest pain or shortness of breath, or heart palpitations, while other people will not have any symptoms. However, a major concern for individuals with FHCM is the risk for sudden death, even if they have had no prior symptoms of heart trouble.

Causes

There are over a dozen different genes that have found to cause FHCM:

- *MYH7*: 40% of all FHCM
- *MYBPC3*: 40% of all FHCM
- *TNNT2*: 5% of all FHCM
- *TNNI3*: 5% of all FHCM
- *TPM1:* 2% of all FHCM
- *MYL3:* 1% of all FHCM
- *PRKAG2:* unknown percentage of all FHCM
- *MYL2:* unknown percentage of all FHCM
- *ACTC1:* unknown percentage of all FHCM

It is estimated that approximately 1 in 500 people have FHCM. Most (~70%) people with FHCM will have a parent who also has FHCM. For the other 30%, they will be the first ones in their family to have FHCM (called *de novo*).

Diagnosing FHCM

While genetic testing for FHCM can be helpful to establish a diagnosis, medical providers may also use other medical tests, such as an echocardiogram, a cardiac MRI, or your family history. Red flags in the family history that may increase the chance for FHCM include people with:

- Heart failure
- Hypertrophic cardiomyopathy
- Heart transplants
- Unexplained or sudden death, particularly before the age of 40
- Arrhythmias
- Unexplained stroke or blood clotting disease

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

Medical Management for FHCM

Treatment for FHCM 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 FHCM. Medications, certain therapies, and implantable devices, such as a pacemaker or implantable cardiac defibrillator may be part of the treatment plan. It is also recommended that people who are known to have FHCM avoid high intensity exercise (such as endurance training or running sprints), heavy weight lifting, dehydration, and certain medications (including some blood pressure medications).

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

Additional Resources

Children's Cardiomyopathy Foundation (CCF)

Hypertrophic Cardiomyopathy Association (HCMA)

American Heart Association: Hypertrophic Cardiomyopathy