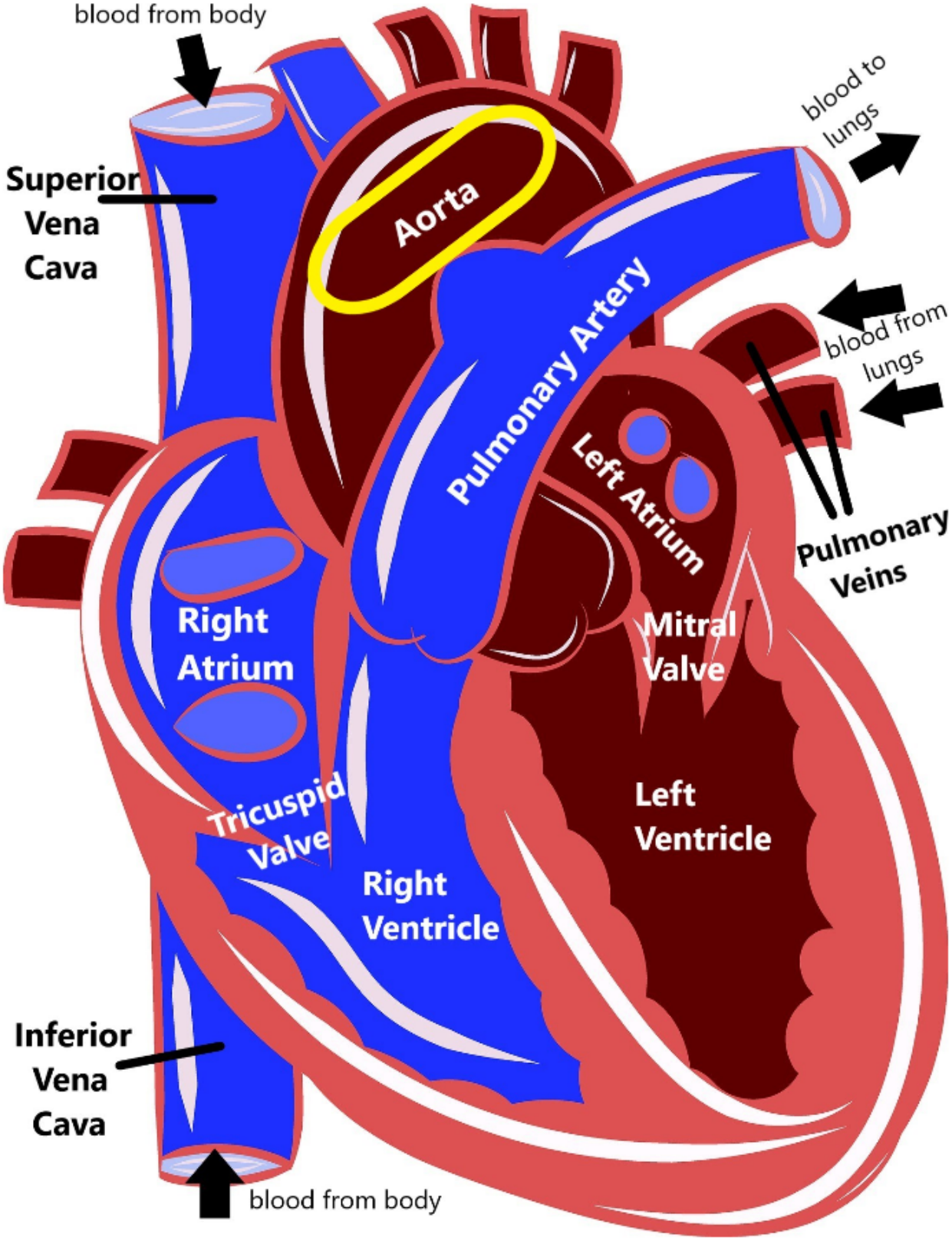
Familial thoracic aortic aneurysm and dissection (familial TAAD) is a hereditary condition that increases the chance for a specific type of heart issue. The aorta is the upper part of the heart that pumps oxygenated blood from our heart out to the rest of our body:


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

People who have familial TAAD can develop a weakened aorta. This weakening in the wall of the aorta can lead to pocket that bulges out of the side of the aorta, called an aneurysm. There are usually no physical signs or symptoms of an aneurysm, but some people can see changes in their breathing (shortness of breath, hoarseness, or wheezing), pain or swelling in the head, neck or upper body, or sudden sharp pain in the chest or back.

The weakening of the aortic wall can also lead to a widening of the aorta, called aortic dilation. This bulging and weakening of the wall of the aorta can also lead to an aortic dissection, or when the aorta ruptures. An aortic dissection can lead to death because it causes blood to flow outside of the aorta, and not enough blood is being pumped out to the rest of the body.

Individuals who have familial TAAD most frequently will have aortic dilation as the first sign, but some people may have a dissection without having any other previous symptoms. Familial TAAD may be an isolated hereditary health condition, or it could be associated to other underlying genetic conditions like Marfan syndrome, Loeys-Dietz syndrome, and vascular Ehlers-Danlos syndrome.

Causes

At least 20% of all aortic aneurysms and dissection are thought to be due to familial TAAD. That means the remaining 80% are likely not strongly genetic, and are multifactorial, or are due to genetic causes that we do not yet know about.

There are over a dozen genes that have been found to cause familial TAAD:

- *ACTA2* (12-21% of all cases of familial TAAD)
- *TGFBR2* (5% of all cases of familial TAAD)
- *FBN1* (3% of all cases of familial TAAD), which also causes Marfan syndrome
- *TGFBR1* (3% of all cases of familial TAAD)
- *SMAD3* (2% of all cases of familial TAAD)
- *LOX* (1.5% of all cases of familial TAAD)
- *FOXE3* (1.4% of all cases of familial TAAD)
- *MAT2A* (1% of all cases of familial TAAD)
- *MYH11* (1% of all cases of familial TAAD)
- *MYLK* (1% of all cases of familial TAAD)
- *PRKG1* (1% of all cases of familial TAAD)
- *TGFB2* (1% of all cases of familial TAAD)
- *MFAP5* (0.25% of all cases of familial TAAD)

- *BGN* (rare cause of familial TAAD)
- *COL3A1* (rare cause of familial TAAD), which also causes vascular Ehlers-Danlos syndrome
- *TGFB3* (rare cause of familial TAAD)

Diagnosing familial TAAD

While genetic testing for familial TAAD can be helpful to establish a diagnosis, medical providers may use other pieces of information from other lab tests or someone's medical and family history to make the diagnosis. Some these tests may include an echocardiogram, a cardiac CT scan, or a cardiac MRI. A family history of aortic or general cardiovascular disease could increase the chance for familial TAAD in a family. Many families will have a history of heart disease and DO NOT have familial TAAD, however someone with a strong pattern of these heart issues may be at a higher chance to have familial TAAD and may benefit from talking about it more with a specialist, such as a cardiologist or a genetic counselor.

Medical Management for familial TAAD

Treatment for familial TAAD can sometimes vary depending on the individual person and their specific health concerns, and should be discussed with a provider who is familiar with familial TAAD. Medical management for more specific forms of familial TAAD, such as Marfan syndrome, Loeys-Dietz syndrome, and vascular Ehlers-Danlos syndrome, may also include screening for other related health concerns. Some options for medical management include an echocardiogram, a cardiac CT scan, or a cardiac MRI in order to look at the aorta to assess for dilation or aneurysm, medications called beta blockers that help to lower the blood pressure (which reduces the stress on the heart), and avoiding contact sports. Other risk factors for heart disease, such as smoking and high cholesterol, should also be avoided when possible.

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

Additional Resources

The John Ritter Foundation for Aortic Health

The Marfan Foundation