A thoracic aortic aneurysm is an enlargement of the aorta in the thoracic cavity (chest area), which is the first part of the artery that takes blood away from the heart. A thoracic aortic aneurysm can progressively enlarge over time and doesn’t usually have symptoms. However, if an enlarged aneurysm is not surgically repaired, it can lead to an acute aortic dissection, a tear that causes the walls of the aorta to separate and allows blood to flow where it shouldn’t. This is potentially life-threatening.

Several factors put people at risk for thoracic aortic aneurysm and dissection. The condition does not always cause symptoms; therefore, it is important to know if you are at risk so that the aorta can be repaired before it tears.

In the case of thoracic aortic aneurysm passed on through generations of a family, it is referred to as familial thoracic aortic aneurysm and dissection (FTAAD).

**What other names do people use for familial thoracic aortic aneurysm and dissection?**

Familial thoracic aortic aneurysm and dissection (FTAAD) is also referred to as thoracic aortic aneurysm (TAA), thoracic aortic aneurysm and dissection (TAAD), familial aortic aneurysm (FAA), or annuloaortic ectasia.
How prevalent is familial thoracic aortic aneurysm and dissection?

Thoracic aortic aneurysm and dissection is fairly common in the general population, but only 20 percent of the cases are caused by a genetic condition. There does not appear to be any difference between ethnic or racial groups. Many people do not know that their family has a predisposition for familial thoracic aortic aneurysm and dissection. Therefore, it is recommended that the first-degree relatives (i.e., parent, sibling, and child) of people affected are also screened for thoracic aneurysms.

What are the characteristics of familial thoracic aortic aneurysm and dissection?

The changes in the aorta associated with familial thoracic aortic aneurysm and dissection may appear in early childhood or at any time later in life.

Aortic aneurysms can be asymptomatic (without symptoms) or symptomatic (with symptoms). If there are symptoms, they may be related to the location, size, and growth rate of the aneurysm. The symptoms include pain in the chest, neck, and/or back; swelling of the head, neck, and arms; wheezing, coughing, or shortness of breath; and coughing up blood.

An aortic dissection usually causes severe, sudden, constant pain in the chest and/or upper back that patients describe as “ripping” or “tearing,” and the pain may feel like it moves from one place to another. Aortic dissection may also result in unusually pale skin (pallor), a very faint pulse, numbness or tingling in one or both arms and legs (paresthesias), and paralysis.

Both aortic aneurysms and dissections increase the risk that the aorta will suddenly rupture, causing massive internal bleeding. Without surgery to prevent aortic rupture, these blood vessel abnormalities can be life-threatening.

In cases of familial thoracic aortic aneurysm, in which the disease is passed on through generations of a family, the occurrence and timing of the changes in the aorta associated with thoracic aortic aneurysm and dissection can vary even within the same family.

Occasionally, people with familial thoracic aortic aneurysm and dissection also may develop aneurysms in the brain or in the section of the aorta located in the abdomen (abdominal aorta). Some people with FTAAD have congenital (at birth) heart abnormalities, such as a bicuspid aorta or patent ductus arteriosus (blood vessels in the heart that are supposed to close one to two days after birth but do not). Other families also have a predisposition for blockage of smaller arteries, leading to early onset stroke and coronary artery disease.

People with familial thoracic aortic aneurysm and dissection may also have features such as a soft out-pouching in the lower abdomen (inguinal hernia), an abnormal curvature of the spine (scoliosis), or a purplish skin discoloration (livedo reticularis) caused by abnormalities in the tiny blood vessels of the skin (dermal capillaries), but these features are also common in the general population.

What are the causes of familial thoracic aortic aneurysm and dissection?

The ACTA2, MYH11, MYLK, and PRKG1 genes are known to cause familial aortic aneurysm and dissection. In addition, the TGFBRI, TGFBRII, SMAD3, TGFB2, and FBN-1 genes have been associated with FTAAD and other disorders that have specific outward physical characteristics. Genetic testing is available for all of these genes. If a person has a genetic test, they should discuss the results with a geneticist or genetic counselor.
Each child of an affected parent has an up to 50 percent chance of inheriting the disorder. Prenatal testing is possible for pregnancies if there is a known disease-causing mutation in the family.

**How is familial thoracic aortic aneurysm and dissection diagnosed?**

Thoracic aortic aneurysm and dissection is diagnosed based on the presence of dilatation and/or dissection of the thoracic aorta and the absence of clinical features of other connective tissue disorders, such as Marfan syndrome, Loeys-Dietz syndrome, or vascular Ehlers-Danlos syndrome. Speak to your doctor about differential diagnosis for these connective tissue disorders.

A diagnosis of familial thoracic aortic aneurysm and dissection is made when there is a positive family history of aneurysm and dissection.

Aortic aneurysms are diagnosed using imaging techniques such as echocardiography (sound wave picture), computed tomography (CT or CAT scan), magnetic resonance imaging (MRI), transesophageal echocardiogram (TEE), chest x-ray, or angiography. Aortic dissections can be diagnosed through computed tomography (CT or CAT scan) or transesophageal echocardiogram (TEE).

**How is familial thoracic aortic aneurysm and dissection managed?**

The management of familial thoracic aortic aneurysm and/or dissection requires the coordinated input from a multidisciplinary team of specialists familiar with this condition, including a medical geneticist, cardiologist, and cardiovascular surgeon.

Medications (drugs) that reduce the stress on the aorta can be helpful. Beta-blockers help lower blood pressure and reduce the force of the heartbeat. They also may help prevent or slow aortic enlargement and reduce the risk of aortic dissection.

People with familial thoracic aortic aneurysm and dissection must have routine tests to monitor their aortic health and identify problems before there is an emergency. These are the same imaging tests that doctors use to diagnose the condition: echocardiogram, MRI, CT scan, or TEE.

For individuals with an ACTA2 mutation, screening for coronary artery disease and cerebrovascular disease is reasonable. For individuals with a TGFBR1, TGFBR2, TGFB2, or SMAD3 mutation, annual imaging of the aorta and its branches and cerebrovascular circulation is recommended.

Surgery is most effective if it is done before an aortic dissection or other life-threatening situation. Surgery is considered when:

- The rate of enlargement of the ascending aorta approaches 0.5 cm per year
- The diameter of the ascending aorta is between 4.2 and 5.0 cm (depending on the underlying mutation or family history)

The two most common types of surgery are:

- The Bentall procedure, which involves an aortic graft repair plus insertion of an aortic mechanical valve. After this surgery, a patient is required to take blood-thinning medication for the rest of their life to avoid blood clots, which could be life-threatening.
• Valve-sparing surgery, which replaces the damaged part of the aorta, but keeps the patient’s own aortic valve. This type of surgery, which does not require blood-thinning medication afterward, can only be performed if the patient’s valve is working properly.

Regardless of which type of surgery is performed, patients must continue to take their blood pressure medicine and have their aorta monitored at least once a year to safeguard against additional enlargements or tears in other parts of the aorta.

What can you do to help safeguard you and your family’s health?
If you are at risk of familial thoracic aortic aneurysm and dissection, there are steps you can take to try to prevent a serious situation:

• Have an echocardiogram or other aortic imaging study at least once a year. Your doctor may want you to have more frequent echocardiograms (every three to six months) to make sure your aorta is growing very little or not at all.

• Do not put extra stress on your aorta. You should only participate in gentle exercise, such as walking, slow jogging, and leisurely biking. Avoid competitive and contact sports, as well as bodybuilding and weight training.

• Learn about aortic disease and any other cardiovascular problems that you have. People with these conditions often need to teach others about this condition. You can help by talking with family members and your healthcare team about cardiovascular problems that may affect you.

• Talk with your healthcare team if your child has been diagnosed with FTAAD. Sometimes doctors suggest medication for very young children.

• Encourage family members to be evaluated. Imaging is recommended for first-degree relatives (i.e., parent, sibling, and child) of an individual with FTAAD. Aortic imaging is warranted once a year, or every few years, for at-risk relatives because the onset of an aneurysm can happen at any age. If the disease-causing mutation is known, genetic counseling and genetic testing of at-risk first-degree relatives assures that only relatives with the mutation undergo aortic imaging.

What is the life expectancy for someone with familial thoracic aortic aneurysm and dissection?
With proper management, including medical therapy and elective (planned) surgery to repair an aneurysm before it tears or ruptures, the life expectancy of people with this condition should approach that of the general population. At-risk family members who are screened for aortic dilatation and undergo elective aortic surgery are expected to have a better prognosis than relatives who were treated after an aortic dissection has occurred.

Do you have questions? Would you like more information?

• Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.

• Visit our website at marfan.org. You can print information that interests you and ask questions online.