The aorta is one of the largest blood vessels in your body. It goes from your heart to your lower abdomen. This blood vessel carries oxygen-rich blood from your heart to all parts of your body. The thoracic aorta is the upper part of the aorta located in your chest. The lower part of the aorta is called the abdominal aorta.

A weak spot in the aortic wall can cause the aorta to bulge out. This is called an aneurysm. Over time, pressure from blood flowing over an aneurysm may cause the layers of the aortic wall to tear apart. This is an aortic dissection. Although aneurysms and dissections are related, aneurysms can occur without dissection and dissections can occur without aneurysms.

How is an aortic aneurysm diagnosed?
There are usually no symptoms with an aortic aneurysm. An ultrasound machine uses sound waves to let the doctor see and measure the aorta. The diameter of the aorta is measured in several places, including the point where it connects to the heart, called the aortic “root”. Normal aortic size varies by age, sex, and body size. Doctors can estimate the expected size of the aorta for each individual. An aneurysm is diagnosed when any area of the aorta measures larger than expected.

How is an aortic dissection diagnosed?
Aortic dissection usually causes sudden, intense pain, similar to symptoms of a heart attack. These types of symptoms should prompt immediate medical care. The diagnosis can be made using special medical imaging, such as an echocardiogram, CT (computerized tomography), or MRI (magnetic resonance imaging).

What causes aortic aneurysms and dissections?
There are many factors that increase the chance for an aortic aneurysm or dissection. High blood pressure, older age, physical trauma (like a car accident), smoking, and inflammation of the blood vessels (vasculitis) are all risk factors for aortic aneurysms and dissections. There are also genetic conditions that can make the aortic wall more prone to aneurysms and dissection. Aneurysms and dissections that occur in the thoracic aorta are more likely to be due to genetic causes than an aneurysm or dissection in the abdomen. Up to 20% of thoracic aortic aneurysms and dissections (TAAD) are thought to be due to inherited or genetic causes.

When is familial TAAD suspected?
Your doctor may suspect a genetic or familial form of TAAD based on your personal and family history. However, it is not always easy to recognize familial TAAD. There may be no one else in the family who has had their aorta measured. Some forms of familial TAAD only affect the aorta and have no other symptoms. Other forms of familial TAAD might include additional medical findings, like tall stature, eye problems, or fragile skin. In some families, there may be only one or two people with symptoms that come to medical attention.

GATHERING YOUR FAMILY HISTORY: WHAT TO ASK
Many families with TAAD are not aware of the condition. When you gather family history, it is helpful to ask about medical problems that might be related to TAAD.

Ask your relatives whether any of the following medical problems have happened in your family:
- Known aortic aneurysms or dissections
- Bicuspid aortic valve
- Dislocated lens in one or both eyes
- Heart surgery
- Sudden cardiac death

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How is familial TAAD inherited?
There are different ways that familial TAAD can be inherited. Most types of familial TAAD are inherited in an autosomal dominant pattern. This means that when one parent has a mutation (disease-causing change) in a TAAD gene, there is a 50% chance to pass the mutation on to any child. A mutation in a TAAD gene can be inherited from either a mother or a father and can cause aortic problems in both males and females who have the mutation. Less often, familial TAAD occurs due to a new mutation in a TAAD gene that is not found in either parent. This is called a de novo mutation. A de novo mutation happens as a random genetic change when an egg or sperm is formed. A person with a de novo mutation can still pass it to their offspring, but other family members are not at risk for the condition. Rarely, TAAD may be caused by a mutation in a gene with a different pattern of inheritance.

How is a gene mutation identified?
A blood test can be done to look for mutations in one or more of the TAAD genes. Hundreds of mutations in many different genes have been found to cause familial TAAD. A genetic testing panel lets us look at many of the TAAD genes with just a single blood test. However, current genetic testing only finds a mutation in about one out of every five families. Testing is more likely to find a disease-causing mutation when your medical history and family history has a clear pattern of familial TAAD. Your genetics consultation will help determine whether or not testing is appropriate.

What are the benefits of genetic testing?
There are two main benefits of genetic testing: better medical care and family screening. Medical guidelines have been developed for certain forms of familial TAAD. By knowing the gene involved, your medical care can focus on risks related to that gene. In addition, once genetic testing finds a disease-causing mutation in a person with TAAD, family members can be offered testing. Genetic testing can identify relatives at-risk for aortic disease before they have serious symptoms. Aneurysms and dissections often have no warning signs. Regular cardiac screening, medication, and risk-reducing heart surgery (when needed), can all lower the risk for individuals with a TAAD mutation. Relatives without the family mutation can be given reassurance.

What if genetic testing doesn't find a mutation?
Genetic testing is not always able to find a mutation in a person with TAAD, even in families with a strong history of TAAD. There are some rare TAAD genes that may not be included on the testing panel. There also may be other TAAD genes that have not been discovered yet. If the history is suspicious for familial TAAD, cardiac screening may still be recommended for all close family members, even when genetic testing is negative (no mutation is found).

Does everyone with a TAAD mutation have aortic problems?
A person who inherits a TAAD mutation has a much higher chance than usual to have an aortic aneurysm or dissection during his or her lifetime. However, some people who inherit a mutation never develop any aortic problems. Regular cardiac screening is recommended for all mutation carriers.

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The information is not intended to diagnose health problems or to take the place of professional medical care. If you have persistent health problems or if you have further questions, please consult your health care provider.

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