

Aortic imaging for individuals with a family history of thoracic aortic aneurysms and dissections

Approximately 20% of patients with thoracic aortic aneurysms and dissections have a similarly affected first-degree relative, indicating a genetic or heritable cause of the condition in the family.

The [2022 ACC/AHA Guideline for the Diagnosis and Management of Aortic Disease](#) give a **Class 1 (strong) recommendation** that first-degree relatives (parents, children, siblings) of someone with thoracic aortic disease should have aortic imaging. TTE is the preferred test if the aortic root and ascending aorta are adequately visualized. If not, then a CT or MRI is recommended.

Early diagnosis of a thoracic aortic aneurysm can be lifesaving because aneurysms can be surgically repaired electively to prevent an acute aortic dissection.

Many patients with genetic aortic disease have no obvious physical features (e.g. like those seen in Marfan syndrome). A lack of syndromic features is **not** a reason to skip aortic imaging.

- For children, baseline echocardiography is recommended, and depending on findings and family history, repeat imaging later in childhood may be appropriate.

- If you are unsure, refer to a geneticist or cardiologist familiar with genetic cardiovascular disease. A timely evaluation can make all the difference.

- When possible, refer to a cardiologist with expertise in aortic disease. Many providers are not familiar with aortic imaging and diagnosis.

- If you don't know of an appropriate provider, [The John Ritter Foundation for Aortic Health](#) can help connect your patient to appropriate resources.

These recommendations are especially relevant when a family member with thoracic aortic disease receives a negative genetic testing result or genetic testing has not been done. Imaging the aorta of family members at risk to diagnosis asymptomatic aneurysms can be lifesaving. If a pathogenic (disease-causing) variant is known, relatives can have targeted genetic testing for that specific variant. Only those who test positive need to undergo aortic imaging. A genetics professional can work with the family to get everyone in the family tested.

Questions or need resources?

Contact **The John Ritter Foundation for Aortic Health** at info@johnritterfoundation.org or **(213) 218-3329**.