




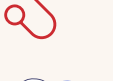





CONSIDER REFERRAL FOR GENETIC ASSESSMENT FOR THOSE WITH:

- Thoracic aortic **dilation** reported on imaging as mild or greater, at age <50y or <60y in the absence of hypertension
- Thoracic aortic **dissection** at age <60y or <70y in the absence of hypertension
- Thoracic aortic **dilation** at any age in the presence of any of the following family histories in a 1st or 2nd degree relative:
 - Thoracic aortic aneurysm (TAA) or thoracic aortic **dissection**
 - Sudden cardiac death age at <50y without a confirmed alternate etiology
- Personal or family history of thoracic aortic **dilation** or TAA and/or features that suggest an underlying syndromic condition, such as:
 - Tall for family
 - Ectopia lentis (lens dislocation)
 - Spontaneous pneumothorax (particularly if recurrent)
 - Hypertelorism (wide-spaced eyes)
 - Bifid uvula
 - Hollow organ rupture
 - Spontaneous tendon rupture
 - Large and unprovoked bruising (prior to anti-coagulation)
 - Very translucent skin
 - Pectus carinatum or significant pectus excavatum
 - Scoliosis requiring bracing or surgery
 - Significant varicose veins at a young age
- 1st or 2nd degree relative in whom a pathogenic variant in one of the HTAD genes has been identified
referral of 3rd degree relatives can be considered when intervening relatives are not available or decline testing

BOTTOM LINE

-  HTAD accounts for ~20-25% of all thoracic aortic aneurysms and dissections.
-  Most individuals with HTAD do not have additional associated features (non-syndromic).
-  HTAD presents at a younger age and is more aggressive than other TAA.
-  Appropriate recognition of HTAD allows initiation of imaging surveillance in at-risk relatives.
-  A positive genetic test result can help guide pharmacotherapy, determine vascular regions which require ongoing imaging surveillance, influence surgical threshold, and allow for cascade testing of at-risk relatives.
-  In most families with HTAD, genetic testing does NOT identify the responsible genetic variant. Thus negative testing does not exclude HTAD and at-risk relatives would still need ongoing imaging surveillance.
-  Pharmacological management for those with TAA may include:
 - beta-blockers or angiotensin receptor blockers to limit aneurysmal dilation.
 - avoidance of medications and recreational drugs with potential vasoactive effect (e.g. triptans, cocaine).
-  Fluoroquinolones should be avoided where possible in anyone with or at risk for aortic aneurysms of any type because of the associated increased risk of aortic dissection.
-  Participation in competitive sports and isometric exercises are advised against.

KEY DEFINITIONS

- **Dilation:** when the diameter of the aorta exceeds the norms for a given age and body size. Reported as borderline, mild, moderate or severe on imaging.
- **Aneurysm:** a dilation >50% larger than the blood vessel should be. All aneurysms are dilations, however not every dilation will reach the size of an aneurysm.
- **Dissection:** a rip or tear in the inner lining of a blood vessel.

Further details on surveillance, management, resources and more can be found in our [additional HTAD resources](#).