Heritable Thoracic Aortic Disease (HTAD)

GECKO Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

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 or likely 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 as much as possible in anyone with aortic aneurysms or at risk for aortic aneurysm (thoracic or not, heritable or not), 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.
- Degenerative: an aneurysm or dilation caused by the deterioration of a blood vessel over time, associated with risk factors such as high blood pressure and age.

Further details on surveillance, management, resources and more can be found in our additional HTAD resources.

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