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

Hereditary Thoracic Aortic Disease (HTAD)

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.

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