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

GENETIC CONSIDERATIONS WHEN ASSESSING BICUSPID AORTIC VALVE (FIG 1.)

Oct 2025

INDIVIDUAL WITH BICUSPID AORTIC VALVE (BAV)

FIRST-DEGREE RELATIVES

Recommend screening by transthoracic echocardiogram (TTE)

If there is no evidence of BAV or aortic dilatation on TTE, no suggestion of family history of aortic aneurysm/dissection, no suggestive signs of a syndromic aortopathy then one time TTE is reasonable for FDRs.

NON-SYNDROMIC

isolated with or without thoracic aortic disease

SYNDROMIC

Features of a connective tissue disorder such as Marfan syndrome, vascular Ehler-Danlos syndrome, Loeys-Dietz syndrome **or** features suspicious of Turner syndrome in those assigned female at birth.

FAMILY HISTORY

Take a 3-generation family history asking about other relatives with:

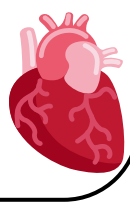
- ☐ BAV
- ☐ left ventricular outflow track obstructive (LVOTO) defects
- ☐ thoracic aortic disease
- ☐ other congenital heart defect
- ☐ sudden cardiac death

- ☐ Family history of a connective tissue disorder
- ☐ Tall for family
- ☐ Ectopia lentis (lens dislocation)
- ☐ Spontaneous pneumothorax (particularly if recurrent)
- ☐ Hypertelorism (wide-spaced eyes)
- ☐ Bifid uvula
- ☐ Hollow organ rupture
- ☐ Spontaneous tendon rupture
- ☐ Extensive 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
- ☐ Suspicion of Turner syndrome. Highly variable presentation but may include: short stature, low posterior hairline, low set ears, short webbed neck, premature ovarian failure, delayed or failure to attain puberty, congenital anomalies e.g. heart, kidney

IF ALL ARE NO

REFER

Consider referral to cardiology for surveillance and management.



IF ANY ARE YES

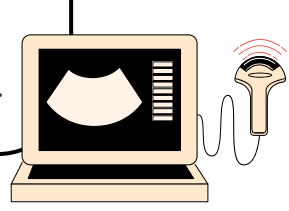
REFER

Consider referral to cardiology for surveillance and management **AND** to genetics

IMAGING AND SURVEILLANCE

Transthoracic echocardiography (TTE) is the first-line imaging for BAV diagnosis and phenotyping. Computed tomography (CT) angiography, or cardiac magnetic resonance imaging (MRI) can be considered when TTE visualization is poor.

Frequency of surveillance will be determined based on results, personal and family history.



IF ANY ARE YES

REFER

Consider referral to genetics for an assessment.