DISEASE:
Familial bicuspid aortic valve

NAME:	Familial bicuspid aortic valve
DESCRIPTION:	Familial bicuspid aortic valve is a rare, genetic, aortic malformation defined as a presence of abnormal two-leaflet aortic valve in at least 2 first-degree relatives. It is frequently asymptomatic or may be associated with progressive aortic valve disease (aortic regurgitation and/or aortic stenosis, typically due to valve calcification) and a concomitant aortopathy (i.e. aortic dilation, aortic aneurysm and/or dissection).
ORPHACODE:	402075
SYNONYMS:	Familial BAV
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	NKX2-5 GATA5 NOTCH1 SMAD6
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Aneurysm, Thoracic Aortic, familial (gene panel)
- Bicuspid aortic valve
- Cardiopathies, hereditary (gene panel)
- Familial Thoracic Aortic Aneurysm (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- GATA binding protein 5
- NK2 homeobox 5
- notch receptor 1
- SMAD family member 6

Related Gene Panels

- Bicuspid aortic valve - UGent
- Cardiopathies, hereditary (102 genes) - KUL

- Congenital heart disease (29 genes) - VUB
- Familial Thoracic Aortic Aneurysm (21 genes) - UGent
- Familial Thoracic Aortic Aneurysm (genepanel) - UZA
