DISEASE:
Familial thoracic aortic aneurysm and aortic dissection

NAME:	Familial thoracic aortic aneurysm and aortic dissection
DESCRIPTION:	Familial thoracic aortic aneurysm and aortic dissection is a rare genetic vascular disease characterized by the familial occurrence of thoracic aortic aneurysm, dissection or dilatation affecting one or more aortic segments (aortic root, ascending aorta, arch or descending aorta) in the absence of any other associated disease. Depending on the size, location and progression rate of dilatation/dissection, patients may be asymptomatic or may present dyspnea, cough, jaw, neck, chest or back pain, head, neck or upper limb edema, difficulty swallowing, voice hoarseness, pale skin, faint pulse and/or numbness/tingling in limbs. Patients have increased risk of presenting life threatening aortic rupture.
ORPHACODE:	91387
SYNONYMS:	Familial TAAD Familial non-syndromic thoracic aortic aneurysm and aortic dissection

XREF(S):	Orphanet OMIM OMIM OMIM ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	FOXE3 THSD4 SMAD2 LOX MAT2A ELN HEY2 TGFB3 TGFBR1 TGFBR2 FBN1 ACTA2 MYLK SMAD3 PRKG1 MFAP5 TGFB2 SMAD4 MYH11
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Aneurysm, Thoracic Aortic, familial (gene panel)
- Cardiomyopathy, hereditary (gene panel)
- Cardiopathies, hereditary (gene panel)
- Cataract (gene panel)
- Familial Thoracic Aortic Aneurysm (gene panel)

Related Laboratories

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

Related Analytes

- actin alpha 2, smooth muscle
- elastin
- fibrillin 1
- forkhead box E3
- hes related family bHLH transcription factor with YRPW motif 2
- lysyl oxidase
- methionine adenosyltransferase 2A
- microfibril associated protein 5

- myosin heavy chain 11
- myosin light chain kinase
- protein kinase cGMP-dependent 1
- SMAD family member 2
- SMAD family member 3
- SMAD family member 4
- transforming growth factor beta 2
- transforming growth factor beta 3
- transforming growth factor beta receptor 1
- transforming growth factor beta receptor 2
- thrombospondin type 1 domain containing 4

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- Cardiopathies, hereditary (102 genes) - KUL
- Cataract - UGent
- Familial Thoracic Aortic Aneurysm (21 genes) - UGent
- Familial Thoracic Aortic Aneurysm (genepanel) - UZA
- Familiale thoracale aorta aneurysmata (19 genes) - UGent
- test
