

Full name:	Familial Thoracic Aortic Aneurysm (genepanel) - UZA
Abbreviation:	FTAA / TAAD
Version number:	v12
Laboratory:	<u>Centrum Medische Genetica - UZ Antwerpen</u>
Created:	04 Jul 2019 - 14:34
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Related Diseases

- Arterial tortuosity syndrome
- Autosomal dominant cutis laxa
- Cardiac-valvular Ehlers-Danlos syndrome
- Classical Ehlers-Danlos syndrome
- Familial bicuspid aortic valve
- Familial thoracic aortic aneurysm and aortic dissection
- Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency
- Loeys-Dietz syndrome
- Marfan syndrome type 1
- Shprintzen-Goldberg syndrome
- Stiff skin syndrome
- Vascular Ehlers-Danlos syndrome

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ABL1</u>	100.00	0	
<u>ACTA2</u>	100.00	0	
<u>ARIH1</u>	100.00	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ASPH</u>	100.00	0	
<u>BGN</u>	100.00	0	
<u>COL3A1</u>	100.00	0	
<u>EFEMP2</u>	100.00	0	
<u>ELN</u>	100.00	0	
<u>EMILIN1</u>	100.00	0	
<u>FBN1</u>	100.00	1	
<u>FBN2</u>	100.00	0	
<u>FKBP14</u>	100.00	0	
<u>FLNA</u>	100.00	0	
<u>FOXE3</u>	100.00	0	
<u>HCN4</u>	100.00	0	
<u>IPO8</u>	100.00	0	
<u>JAG1</u>	100.00	0	
<u>LMOD1</u>	100.00	0	
<u>LOX</u>	100.00	0	
<u>LTBP3</u>	100.00	0	
<u>MAT2A</u>	100.00	0	
<u>MFAP5</u>	100.00	0	
<u>MYH11</u>	100.00	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MYLK</u>	100.00	0	
<u>NOTCH1</u>	100.00	0	
<u>NPR3</u>	100.00	0	
<u>PLOD1</u>	100.00	0	
<u>PMEPA1</u>	100.00	0	
<u>PRKG1</u>	100.00	0	
<u>ROBO4</u>	100.00	0	
<u>SKI</u>	100.00	0	
<u>SLC2A10</u>	100.00	0	
<u>SMAD2</u>	100.00	0	
<u>SMAD3</u>	100.00	0	
<u>SMAD4</u>	100.00	0	
<u>SMAD6</u>	100.00	0	
<u>TBX20</u>	100.00	0	
<u>TGFB2</u>	100.00	0	
<u>TGFB3</u>	100.00	0	
<u>TGFBR1</u>	100.00	0	
<u>TGFBR2</u>	100.00	1	
<u>THSD4</u>	100.00	0	