

**Appendix:** Composition of the TAAD\_v9 gene panel (thoracic aortic aneurysm/dissection)

Gene	Transcript reference (Ensembl)	Alternative exon	Transcript reference for alternative exon (Ensembl)
<i>ABL1</i>	ENST00000372348		
<i>ACTA2</i>	ENST00000458208		
<i>ARIH1</i>	ENST00000379887		
<i>BGN</i>	ENST00000331595		
<i>COL3A1</i>	ENST00000304636		
<i>EFEMP2/FBLN4</i>	ENST00000307998		
<i>ELN</i>	ENST00000358929		
<i>EMILIN1</i>	ENST00000380320		
<i>FBN1</i>	ENST00000316623		
<i>FBN2</i>	ENST00000262464		
<i>FLNA</i>	ENST00000369850		
<i>FOXE3<sup>a</sup></i>	ENST00000335071		
<i>HCN4</i>	ENST00000261917		
<i>LMOD1</i>	ENST00000367288		
<i>LOX</i>	ENST00000231004		
<i>LTBP3</i>	ENST00000301873		
<i>MAT2A</i>	ENST00000306434		
<i>MFAP5</i>	ENST00000359478		
<i>MYH11</i>	ENST00000452625	Exon 42B	ENST00000396324
<i>MYLK</i>	ENST00000360304		
<i>NOTCH1</i>	ENST00000277541		
<i>PLOD1</i>	ENST00000196061	Exon 2A	ENST00000449038
<i>PMEPA1/TMEPAI</i>	ENST00000341744		
<i>PRKG1<sup>b</sup></i>	ENST00000401604		
<i>SKI</i>	ENST00000378536		
<i>SLC2A10</i>	ENST00000359271		
<i>SMAD2</i>	ENST00000402690		
<i>SMAD3</i>	ENST00000327367	Exon 1A	ENST00000439724
<i>SMAD4</i>	ENST00000342988		
<i>SMAD6</i>	ENST00000288840		
<i>TGFB2</i>	ENST00000366929		
<i>TGFB3</i>	ENST00000238682		
<i>TGFBR1<sup>c</sup></i>	ENST00000374994		
<i>TGFBR2</i>	ENST00000359013		

<sup>a</sup>Forkhead domain only

<sup>b</sup>Exon 3 only

<sup>c</sup>*TGFBR1* exon 1 is partially insufficiently covered in the current version of the test; 63 bp, 15 of which are intronic bp, drop out at the 3' side of the exon.

For the listed genes, a coverage of 30x for >99% of the coding sequence is postulated. For the following core genes a coverage of 100% is guaranteed: *ACTA2*, *COL3A1*, *FBN1*, *FLNA*, *MAT2A*, *MFAP5*, *MYH11*, *MYLK*, *NOTCH1*, *PRKG1*, *SMAD2*, *SMAD3*, *TGFB2*, *TGFB3*, *TGFBR1* and *TGFBR2* (except for exon 1 of *TGFBR1*, see remark above). Large deletions/duplications are not detected with the current version of the assay.