

Appendix: Composition of the cardiomyopathy gene panel (CM_v3)

Gene	Transcript reference (Ensembl)	Alternative transcripts/exons (Ensembl)
<i>ABCC9</i>	ENST00000261200	ENST00000261201: exon 38
<i>ACTC1^a</i>	ENST00000290378	
<i>ACTN2</i>	ENST00000542672	ENST00000366578: exon 8
<i>ALPK3</i>	ENST00000258888	
<i>ANKRD1</i>	ENST00000371697	
<i>BAG3</i>	ENST00000369085	
<i>CALR3</i>	ENST00000269881	
<i>CAV3</i>	ENST00000343849	
<i>CRYAB</i>	ENST00000526180	ENST00000524660: exon 1; ENST00000533971: exon 2
<i>CSRP3</i>	ENST00000265968	
<i>CTNNA3</i>	ENST00000433211	
<i>DES</i>	ENST00000373960	
<i>DSC2</i>	ENST00000280904	ENST00000251081: exon 16
<i>DSG2</i>	ENST00000261590	
<i>DSP</i>	ENST00000379802	
<i>EMD (STA)</i>	ENST00000369842	
<i>FHL1</i>	ENST00000394155	ENST00000370683: exon 1, exon 6
<i>FHL2</i>	ENST00000409177	
<i>FLNC</i>	ENST00000325888	
<i>GLA</i>	ENST00000218516	
<i>JPH2</i>	ENST00000372980	
<i>JUP</i>	ENST00000393931	
<i>LAMA4</i>	ENST00000230538	
<i>LAMP2</i>	ENST00000371335	ENST00000434600: exon 9; ENST00000200639: exon 9
<i>LDB3</i>	ENST00000429277	ENST00000372066: exon 4; ENST00000372056: exon 8; ENST00000361373: exon 7
<i>LMNA</i>	ENST00000368300	ENST00000368297: exon 2, exon 11
<i>MIB1</i>	ENST00000261537	
<i>MYBPC3</i>	ENST00000545968	
<i>MYH6^b</i>	ENST00000405093	
<i>MYH7</i>	ENST00000355349	
<i>MYL2</i>	ENST00000228841	
<i>MYL3</i>	ENST00000292327	
<i>MYOZ2</i>	ENST00000307128	
<i>MYPN</i>	ENST00000358913	
<i>NEBL</i>	ENST00000377122	ENST00000417816: Exon 1, Exon 2, Exon 3, Exon 4
<i>NEXN</i>	ENST00000334785	
<i>PKP2</i>	ENST00000070846	
<i>PLN</i>	ENST00000357525	
<i>PPA2</i>	ENST00000341695	
<i>PRDM16</i>	ENST00000270722	
<i>PRKAG2</i>	ENST00000287878	
<i>RAF1</i>	ENST00000442415	
<i>RBM20</i>	ENST00000369519	
<i>RYR2</i>	ENST00000366574	
<i>SCN5A</i>	ENST00000333535	ENST00000413689: exon 6
<i>SDHA</i>	ENST00000264932	
<i>SGCD</i>	ENST00000337851	ENST00000517913: exon 10
<i>SYNE1</i>	ENST00000367255	
<i>TAZ</i>	ENST00000601016	
<i>TCAP</i>	ENST00000309889	
<i>TGFB3</i>	ENST00000238682	
<i>TMEM43</i>	ENST00000306077	
<i>TNNC1</i>	ENST00000232975	
<i>TNNI3</i>	ENST00000344887	
<i>TNNT2^c</i>	ENST00000236918	
<i>TPM1</i>	ENST00000403994	ENST00000334895: exon 1, exon 8; ENST00000559397: exon 2; ENST00000358278: exon 6; ENST00000288398: exon 9; ENST00000559556: exon 9
<i>TTN^d</i>	ENST00000589042	ENST00000360870: exon 46
<i>TTR</i>	ENST00000237014	
<i>VCL</i>	ENST00000211998	

^a*ACTC1* exon 6 is partially insufficiently covered in the current version of the test; 107 bp, 15 of which are intronic bp, drop out at the 5' side of the exon

^b*MYH6* exon 26 is partially insufficiently covered in the current version of the test; 88 bp drop out in the middle of the exon

^cAll annotations are based on the Hg19/GRCh37 genome build, with exception of *TNNT2*, for which the GRCh38 build is used

^dThe most complete *TTN* transcript is ENST00000589042 that contains a total of 363 exons. From this transcript, we have opted to exclude the exons that are not present in the cardiac isoforms: N2BA, N2B, Novex-1, Novex-2 and Novex-3. This means that the following exons are not included in the gene panel: exon 147, exon 149, exons 158 to 201 with the exception of exon 174, exons 212 to 216 (<https://cardiode.org/titin> website).

For the listed genes, a coverage of 30x for >99% of the coding sequence is postulated. Large deletions/duplications are not detected with the current version of the assay.