

Targeted Genes and Methodology Details for Cardiovascular/Connective Tissue/Dyslipidemia/ Cerebrovascular/Primary Ciliary Dyskinesia Custom Gene Panel

Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from June 2025 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>ABCA1</i>	NM_005502.4	chr9:g.107567035A>G (c.4465-34A>G); chr9:g.107599404G>A (c.1195-27G>A)	-
<i>ABCC9</i>	NM_005691.3	-	-
<i>ABCG5</i>	NM_022436.3	-	-
<i>ABCG8</i>	NM_022437.3	-	-
<i>ACAD9</i>	NM_014049.5	-	-
<i>ACADVL</i>	NM_000018.4	-	-
<i>ACTA2</i>	NM_001613.4	-	-
<i>ACTB</i>	NM_001101.5	-	-
<i>ACTC1</i>	NM_005159.5	-	-
<i>ACTG1</i>	NM_001614.5	-	-
<i>ACTN2</i>	NM_001103.3	-	-
<i>ACVRL1</i>	NM_000020.3	-	-
<i>ADA2</i>	NM_001282225.2	-	-
<i>ADAMTS10</i>	NM_030957.4	-	-
<i>ADAMTS17</i>	NM_139057.4	-	-
<i>ADAMTS2</i>	NM_014244.5	-	-
<i>AEBP1</i>	NM_001129.5	-	-
<i>AGL</i>	NM_000642.3	chr1:g.100381954A>G (c.4260-12A>G)	-
<i>AGPAT2</i>	NM_006412.4	-	-
<i>AKAP9</i>	NM_005751.4	-	-
<i>ALMS1</i>	ENST00000264448.6	-	-
<i>ALPK3</i>	NM_020778.4	-	-
<i>ALPL</i>	NM_000478.6	-	-
<i>ANGPTL3</i>	NM_014495.4	-	-
<i>ANK2</i>	NM_020977.4	-	-
<i>ANKRD1</i>	NM_014391.2	-	-
<i>ANO5</i>	NM_213599.2	-	CNV may not be detected in exon 2
<i>APOA1</i>	NM_000039.2	-	-
<i>APOA5</i>	NM_052968.5	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>APOB</i>	NM_000384.3	-	-
<i>APOC2</i>	NM_000483.5	-	-
<i>APOE</i>	NM_000041.4	-	-
<i>ARMC4 (ODAD2)</i>	NM_018076.5	-	Sequence variants and CNV may not be detected in exon 9
<i>ATP1A2</i>	NM_000702.4	-	-
<i>ATP1A3</i>	NM_152296.5	-	-
<i>ATP7A</i>	NM_000052.7	-	-
<i>B3GALT6</i>	NM_080605.4	-	-
<i>B3GAT3</i>	NM_012200.4	-	-
<i>B4GALT7</i>	NM_007255.3	-	-
<i>BAG3</i>	NM_004281.3	-	-
<i>BCOR</i>	NM_017745.6	-	-
<i>BGN</i>	NM_001711.6	-	-
<i>BMP1</i>	NM_006129.5	-	-
<i>BMP1</i>	NM_001199.4	-	CNV and sequence variants will be analyzed and reported in exon 16 only
<i>BMPR2</i>	NM_001204.7	chr2:g.203395505T>G (c.968-12T>G)	-
<i>BRAF</i>	NM_004333.6	-	-
<i>BSCL2</i>	NM_032667.6	chr11:g.62470032T>C (c.213-11A>G)	-
<i>CACNA1A</i>	NM_001127221.1	-	-
<i>CACNA1C</i>	NM_000719.7	-	-
<i>CACNA1D</i>	NM_000720.4	-	-
<i>CACNA2D1</i>	NM_000722.4	-	CNV may not be detected in exons 14 and 20
<i>CACNB2</i>	NM_201590.3	-	-
<i>CALM1</i>	NM_006888.6	-	-
<i>CALM2</i>	NM_001743.6	-	-
<i>CALM3</i>	NM_005184.4	-	-
<i>CASQ2</i>	NM_001232.3	-	-
<i>CAV1</i>	NM_001753.5	-	-
<i>CAV3</i>	NM_033337.3	-	-
<i>CAVIN1</i>	NM_012232.6	-	-
<i>CBL</i>	NM_005188.4	-	-
<i>CBS</i>	NM_000071.2	-	-
<i>CCDC103</i>	NM_213607.3	-	-
<i>CCDC114 (ODAD1)</i>	NM_144577.4	-	-
<i>CCDC151 (ODAD3)</i>	NM_145045.5	-	-
<i>CCDC39</i>	NM_181426.2	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CCDC40</i>	NM_017950.4	-	-
<i>CCDC65</i>	NM_033124.5	-	-
<i>CCM2</i>	NM_031443.3	-	-
<i>CCNO</i>	NM_021147.5	-	-
<i>CDH2</i>	NM_001792.5	-	-
<i>CETP</i>	NM_000078.3	-	-
<i>CFAP298</i>	NM_021254.4	-	CNV may not be detected in exon 3
<i>CFAP300</i>	NM_032930.3	-	-
<i>CHD7</i>	NM_017780.4	chr8:g.61763035G>A (c.5405-17G>A)	-
<i>CHRM2</i>	NM_000739.2	-	-
<i>CHST14</i>	NM_130468.3	-	-
<i>CITED2</i>	NM_006079.5	-	-
<i>COL1A1</i>	NM_000088.3	chr17:g.48273743A>T (c.904-15T>A); chr17:g.48273742C>T (c.904-14G>A); chr17:g.48272201C>T (c.1354-12G>A)	-
<i>COL1A2</i>	NM_000089.4	-	CNV may not be detected in exons 2–3
<i>COL3A1</i>	NM_000090.3	-	-
<i>COL4A1</i>	NM_001845.6	-	-
<i>COL4A2</i>	NM_001846.4	-	-
<i>COL5A1</i>	NM_000093.5	chr9:g.137686903T>G (c.2701-25T>G); chr9:g.137726806T>A (c.5137-11T>A)	-
<i>COL5A2</i>	NM_000393.5	-	CNV may not be detected in exon 3
<i>COL12A1</i>	NM_004370.6	-	-
<i>CPT2</i>	NM_000098.3	-	-
<i>CREB3L1</i>	NM_052854.4	-	-
<i>CREB3L3</i>	NM_032607.3	-	-
<i>CRTAP</i>	NM_006371.5	chr3:g.33160815C>G (c.472-1021C>G)	-
<i>CRYAB</i>	NM_001885.3	-	-
<i>CSRP3</i>	NM_003476.5	-	-
<i>CST3</i>	NM_000099.4	-	-
<i>CYP27A1</i>	NM_000784.4	-	-
<i>DES</i>	NM_001927.4	-	-
<i>DMD</i>	NM_004006.2	chrX:g.33192452C>T (c.31+36947G>A); chrX:g.32827744A>T (c.531-16T>A); chrX:g.32398808T>C (c.4675-11A>G); chrX:g.31279781T>C (c.9225-648A>G); chrX:g.31279780T>C (c.9225-647A>G); chrX:g.31279420G>T (c.9225-287C>A); chrX:g.31279418T>C (c.9225-285A>G)	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>DNAAF1</i>	NM_178452.6	-	-
<i>DNAAF2</i>	NM_018139.2	-	-
<i>DNAAF3</i>	NM_001256714.1	-	-
<i>DNAAF4</i>	NM_130810.4	-	-
<i>DNAAF5</i>	NM_017802.4	-	-
<i>DNAH1</i>	NM_015512.5	-	-
<i>DNAH5</i>	NM_001369.2	-	-
<i>DNAH8</i>	NM_001206927.2	-	-
<i>DNAH9</i>	NM_001372.4	-	-
<i>DNAH11</i>	NM_001277115.2	-	-
<i>DNAI1</i>	NM_012144.4	-	-
<i>DNAI2</i>	NM_023036.6	-	-
<i>DNAJB13</i>	NM_153614.3	-	Sequence variants and CNV may not be detected in exon 7
<i>DNAJC19</i>	NM_145261.4	-	-
<i>DNAL1</i>	NM_031427.4	-	CNV may not be detected in exon 5
<i>DOLK</i>	NM_014908.4	-	-
<i>DRC1</i>	NM_145038.5	-	-
<i>DSC2</i>	NM_024422.6	-	-
<i>DSE</i>	NM_013352.4	-	-
<i>DSG2</i>	NM_001943.5	-	-
<i>DSP</i>	NM_004415.4	-	-
<i>DTNA</i>	NM_001390.4	-	-
<i>EFEMP2</i>	NM_016938.5	-	-
<i>EIF2AK4</i>	NM_001013703.4	-	-
<i>ELAC2</i>	NM_018127.7	-	-
<i>ELN</i>	NM_001278939.1	-	-
<i>EMD</i>	NM_000117.3	-	-
<i>ENG</i>	NM_001114753.2	chr9:g.130616761G>A (c.-127C>T); chr9:g.130578359A>G (c.1742-22T>C)	-
<i>EPHB4</i>	NM_004444.5	-	-
<i>FBN1</i>	NM_000138.4	chr15:g.48818478G>A (c.863-26C>T); chr15:g.48802380T>C (c.1589-14A>G); chr15:g.48780451A>T (c.3209-13T>A); chr15:g.48721629T>C (c.6872-961A>G); chr15:g.48720692A>T (c.6872-24T>A); chr15:g.48713895A>T (c.7571-12T>A); chr15:g.48707358C>A (c.8051+375G>T)	-
<i>FBN2</i>	NM_001999.4	chr5:g.127670562A>C (c.3974-26T>G); chr5:g.127671284T>C (c.3725-15A>G)	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>FHL1</i>	NM_001449.5	-	-
<i>FKBP10</i>	NM_021939.4	-	-
<i>FKBP14</i>	NM_017946.4	-	-
<i>FKRP</i>	NM_024301.5	-	-
<i>FKTN</i>	NM_001079802.1	chr9:g.108368857G>T (c.648-1243G>T); ~3 kb retrotransposon insertion (c.*4392_*4393)	-
<i>FLNA</i>	NM_001456.3	-	-
<i>FLNC</i>	NM_001458.4	-	-
<i>FOXF1</i>	NM_001451.3	-	-
<i>FOXH1</i>	NM_003923.3	-	-
<i>FOXJ1</i>	NM_001454.4	-	-
<i>GAA</i>	NM_000152.5	chr17:g.78078341T>G (c.-32-13T>G); chr17:g.78082266T>G (c.1076-22T>G); chr17:g.78082485T>A (c.1195-11T>A); chr17:g.78092432T>G (c.2647-20T>G)	-
<i>GAS8</i>	NM_001481.3	-	-
<i>GATA4</i>	NM_002052.5	-	-
<i>GATA5</i>	NM_080473.5	-	-
<i>GATA6</i>	NM_005257.5	-	-
<i>GCKR</i>	NM_001486.4	-	-
<i>GDF1</i>	NM_001492.6	-	-
<i>GDF2</i>	NM_016204.4	-	-
<i>GLA</i>	NM_000169.2	chrX:g.100654735C>T (c.640-801G>A); chrX:g.100653945A>T (c.640-11T>A)	-
<i>GLMN</i>	NM_053274.3	-	CNV may not be detected in exons 2, 10, 16 and 17
<i>GNB5</i>	NM_016194.4	-	CNV may not be detected in exon 11
<i>GPD1</i>	NM_005276.4	-	CNV may not be detected in exon 8
<i>GPIHBP1</i>	NM_178172.6	-	-
<i>GUCY1A1</i>	NM_000856.6	-	CNV may not be detected in exon 6
<i>HCN4</i>	NM_005477.3	-	-
<i>HRAS</i>	NM_005343.4	-	-
<i>HTRA1</i>	NM_002775.5	-	-
<i>IFITM5</i>	NM_001025295.3	chr11:g.299504G>A (c.-14C>T)	-
<i>ILK</i>	NM_004517.4	-	-
<i>JAG1</i>	NM_000214.3	chr20:g.10629767A>C (c.1349-12T>G)	-
<i>JPH2</i>	NM_020433.4	-	-
<i>JUP</i>	NM_002230.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>KCND2</i>	NM_012281.3	-	-
<i>KCND3</i>	NM_004980.4	-	-
<i>KCNE1</i>	NM_000219.6	-	-
<i>KCNE2</i>	NM_172201.1	-	-
<i>KCNH2</i>	NM_000238.4	-	-
<i>KCNJ2</i>	NM_000891.3	-	-
<i>KCNJ5</i>	NM_000890.5	-	-
<i>KCNJ6</i>	NM_002240.5	-	-
<i>KCNJ8</i>	NM_004982.4	-	-
<i>KCNQ1</i>	NM_000218.3	-	-
<i>KRAS</i>	NM_004985.5	-	-
<i>KRIT1</i>	NM_194456.1	chr7:g.91856011A>T (c.990-15T>A); chr7:g.91855154G>C (c.1147-13C>G); chr7:g.91830749T>C (c.2026-12A>G)	-
<i>LAMA4</i>	NM_002290.5	-	-
<i>LAMP2</i>	NM_013995.2	-	-
<i>LCAT</i>	NM_000229.2	chr16:g.67976512A>G (c.524-22T>C)	CNV may not be detected in exon 6
<i>LDB3</i>	NM_007078.3	-	-
<i>LDB3</i>	NM_001080116.1	-	-
<i>LDLR</i>	NM_000527.5	chr19:g.11200029_11200044 (c.-196_c.-181) (Repeat 1 Sp1 binding site); chr19:g.11200064_11200079 (c.-161_c.-146) (Repeat 2 SREBP binding site); chr19:g.11200080_11200095 (c.-145_c.-130) (Repeat 3 Sp1 binding site); chr19:g.11231284C>G (c.2140+86C>G); chr19:g.11231301G>T (c.2140+103G>T)	-
<i>LDLRAP1</i>	NM_015627.3	-	-
<i>LIPA</i>	NM_000235.4	-	-
<i>LIPC</i>	NM_000236.3	chr15:g.58830518A>G (c.89-14A>G)	-
<i>LIPE</i>	NM_005357.4	-	-
<i>LMF1</i>	NM_022773.4	-	-
<i>LMNA</i>	NM_170707.4	chr1:g.156105681C>G (c.937-11C>G)	-
<i>LOX</i>	NM_002317.7	-	-
<i>LPL</i>	NM_000237.3	-	Sequence and CNV analyses for exon 10 will not be performed
<i>LRP5</i>	NM_002335.4	-	-
<i>LRP6</i>	NM_002336.3	-	-
<i>LRRC6 (DNAAF11)</i>	NM_012472.6	-	CNV may not be detected in exon 3
<i>LTBP3</i>	NM_001130144.2	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
LZTR1	NM_006767.4	chr22:g.21336623T>A (c.-38T>A); chr22:g.21340117G>A (c.264-13G>A); chr22:g.21349779C>T (c.1943-256C>T); chr22:g.21350968C>A (c.2220-17C>A)	-
MAP2K1	NM_002755.3	-	-
MAP2K2	NM_030662.3	-	-
MBTPS2	NM_015884.4	-	-
MCIDAS	NM_001190787.3	-	-
MED12	NM_005120.3	-	-
MED13L	NM_015335.4	-	-
MEGF8	NM_001271938.2	-	-
MEIS2	NM_170674.5	-	-
MFAP5	NM_003480.4	-	-
MRAS	NM_012219.4	-	-
MTO1	NM_012123.4	-	-
MTPP	NM_000253.3	chr4:g.100522736A>G (c.1237-28A>G)	-
MYBPC3	NM_000256.3	chr11:g.47368616C>T (c.906-36G>A); chr11:g.47364832C>T (c.1224-19G>A); chr11:g.47364834T>C (c.1224-21A>G); chr11:g.47364865C>T (c.1224-52G>A); chr11:g.47364709C>T (c.1227-13G>A); chr11:g.47362606T>C (c.1898-23A>G); chr11:g.47361954G>A (c.1927+600C>T); chr11:g.47354261_47354267del (c.3491-12_3491-6delTGTCACC); chr11:g.47353821G>C (c.3628-12C>G)	CNV may not be detected in exon 10
MYH11	NM_001040113.2	-	-
MYH6	NM_002471.3	-	-
MYH7	NM_000257.4	-	-
MYL2	NM_000432.3	-	-
MYL3	NM_000258.3	-	-
MYLK	NM_053025.4	-	-
MYLK3	NM_182493.3	-	-
MYPN	NM_032578.3	-	-
NEXN	NM_144573.3	-	-
NKX2-5	NM_004387.4	-	-
NKX2-6	NM_001136271.2	-	-
NODAL	NM_018055.5	-	-
NOTCH1	NM_017617.5	-	-
NOTCH2	NM_024408.4	-	Sequence and CNV analyses for exons 1-4 will not be performed
NOTCH3	NM_000435.3	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>NR2F2</i>	NM_021005.4	-	-
<i>NRAS</i>	NM_002524.5	-	-
<i>NSD1</i>	NM_022455.4	-	-
<i>NSUN2</i>	NM_017755.6	-	-
<i>OFD1</i>	NM_003611.3	chrX:g.13773248_13773251del (c.1130-22_1130-19del); chrX:g.13773250_13773254del (c.1130-20_1130-16del)	-
<i>P3H1</i>	NM_022356.4	chr1:g.43212925C>T (c.2055+18G>A)	-
<i>P4HB</i>	NM_000918.4	-	-
<i>PCCA</i>	NM_000282.4	-	CNV may not be detected in exon 10
<i>PCCB</i>	NM_000532.5	-	-
<i>PCSK9</i>	NM_174936.4	-	-
<i>PDCD10</i>	NM_145860.1	-	-
<i>PIH1D3 (DNAAF6)</i>	NM_001169154.1	-	-
<i>PIK3R1</i>	NM_181523.3	-	-
<i>PKP2</i>	NM_004572.3	-	-
<i>PLD1</i>	NM_002662.5	-	CNV may not be detected in exon 6
<i>PLEKHM2</i>	NM_015164.4	-	-
<i>PLIN1</i>	NM_002666.5	-	-
<i>PLN</i>	NM_002667.5	-	-
<i>PLOD1</i>	NM_000302.4	-	-
<i>PLOD2</i>	NM_182943.3	-	CNV may not be detected in exon 16
<i>PLS3</i>	NM_005032.7	chrX:g.114856534T>A (c.74-24T>A)	-
<i>PPA2</i>	NM_176869.3	-	CNV may not be detected in exon 11
<i>PPARG</i>	NM_015869.4	-	-
<i>PPCS</i>	NM_024664.4	-	-
<i>PPIB</i>	NM_000942.4	-	-
<i>PPP1CB</i>	NM_206876.1	-	-
<i>PQBP1</i>	NM_005710.2	-	-
<i>PRDM16</i>	NM_022114.4	-	CNV may not be detected in exon 1
<i>PRDM5</i>	NM_018699.3	-	-
<i>PRKAG2</i>	NM_016203.4	-	CNV may not be detected in exon 13
<i>PRKAR1A</i>	NM_002734.4	-	-
<i>PRKG1</i>	NM_006258.4	-	-
<i>PTPN11</i>	NM_002834.4	-	-
<i>RAF1</i>	NM_002880.3	-	-
<i>RASA1</i>	NM_002890.3	-	-
<i>RBM20</i>	NM_001134363.3	-	-
<i>RIT1</i>	NM_006912.6	-	-

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<i>RNF213</i>	NM_001256071.3	-	-
<i>RPGR</i>	NM_000328.3	-	-
<i>RRAS</i>	NM_006270.5	-	-
<i>RRAS2</i>	NM_012250.6	-	-
<i>RSPH1</i>	NM_080860.4	-	-
<i>RSPH3</i>	NM_031924.6	-	-
<i>RSPH4A</i>	NM_001010892.3	-	-
<i>RSPH9</i>	NM_152732.5	-	-
<i>RYR2</i>	NM_001035.3	-	CNV may not be detected in exons 71, 96, and 104
<i>SAR1B</i>	NM_001033503.3	-	-
<i>SCN1A</i>	NM_001165963.3	chr2:g.166917341G>A (c.265-2143G>A); chr2:g.166863778G>C (c.4002+2451G>C)	-
<i>SCN5A</i>	NM_198056.2	-	-
<i>SEC24D</i>	NM_014822.4	-	-
<i>SERPINF1</i>	NM_002615.7	chr17:g.1674512C>T (c.439+34C>T)	-
<i>SERPINH1</i>	NM_001235.4	-	-
<i>SGCD</i>	NM_000337.5	-	-
<i>SHOC2</i>	NM_007373.3	-	-
<i>SKI</i>	NM_003036.4	-	-
<i>SLC2A10</i>	NM_030777.4	-	-
<i>SLC4A3</i>	NM_201574.2	-	-
<i>SLC22A5</i>	NM_003060.4	chr5:g.131705516G>A (c.-149G>A); chr5:g.131714054T>A (c.394-16T>A); chr5:g.131722665G>A (c.825-52G>A)	-
<i>SLC39A13</i>	NM_152264.4	-	-
<i>SMAD2</i>	NM_005901.6	chr18:g.45396947T>C (c.237-12A>G)	-
<i>SMAD3</i>	NM_005902.4	-	-
<i>SMAD4</i>	NM_005359.6	-	-
<i>SMAD6</i>	NM_005585.5	-	-
<i>SOS1</i>	NM_005633.3	-	-
<i>SOS2</i>	NM_006939.4	-	-
<i>SP7</i>	NM_001173467.3	-	-
<i>SPAG1</i>	NM_172218.2	-	CNV may not be detected in exon 5
<i>SPARC</i>	NM_003118.4	-	-
<i>SPRED1</i>	NM_152594.3	-	-
<i>TAB2</i>	NM_015093.5	-	-
<i>TAPT1</i>	NM_153365.3	-	-
<i>TAZ (TFAFAZZIN)</i>	NM_000116.5	-	-

Targeted Genes and Methodology Details for Cardiovascular/Connective Tissue/Dyslipidemia/ Cerebrovascular/Primary Ciliary Dyskinesia Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>TBX1</i>	NM_080647.1	-	-
<i>TBX20</i>	NM_001077653.2	-	CNV may not be detected in exons 5–8
<i>TBX5</i>	NM_000192.3	-	-
<i>TCAP</i>	NM_003673.4	-	-
<i>TECRL</i>	NM_001010874.5	-	-
<i>TEK</i>	NM_000459.4	-	-
<i>TFAP2B</i>	NM_003221.4	-	-
<i>TGFB2</i>	NM_003238.6	-	-
<i>TGFB3</i>	NM_003239.4	-	-
<i>TGFBR1</i>	NM_004612.4	-	-
<i>TGFBR2</i>	NM_003242.6	-	-
<i>TMEM38B</i>	NM_018112.3	-	-
<i>TMEM43</i>	NM_024334.2	-	-
<i>TMEM70</i>	NM_017866.6	-	-
<i>TNNC1</i>	NM_003280.3	-	-
<i>TNNI3</i>	NM_000363.5	-	-
<i>TNNI3K</i>	NM_015978.3	-	CNV may not be detected in exon 1
<i>TNNT2</i>	NM_001001430.3	-	-
<i>TNXB</i>	NM_019105.8	-	CNV analysis for exons 32–44 will not be performed
<i>TPM1</i>	NM_001018005.2	-	-
<i>TRDN</i>	NM_006073.4	chr6:g.123957870T>C (c.22+29A>G); chr6:g.123850462C>T (c.484+1189G>A)	CNV may not be detected in exons 5, 7, 22, 26, and 28
<i>TREX1</i>	NM_033629.6	-	-
<i>TRIM63</i>	NM_032588.3	-	-
<i>TTC25 (ODAD4)</i>	NM_031421.5	-	Sequence and CNV analyses for exons 9–11 will not be performed
<i>TTN</i>	NM_001256850.1	-	Sequence and CNV analyses for exons 154–156 will not be performed
<i>TTR</i>	NM_000371.3	-	-
<i>VCL</i>	NM_014000.2	-	-
<i>WNT1</i>	NM_005430.4	-	-
<i>XYLT2</i>	NM_022167.4	-	-
<i>ZIC3</i>	NM_003413.4	-	-
<i>ZMPSTE24</i>	NM_005857.5	-	-
<i>ZMYND10</i>	NM_015896.4	-	-
<i>ZNF469</i>	NM_001367624.2	-	-

Targeted Genes and Methodology Details for Cardiovascular/Connective Tissue/Dyslipidemia/ Cerebrovascular/Primary Ciliary Dyskinesia Custom Gene Panel (continued)

Available Cardiovascular/Connective Tissue/Dyslipidemia/ Cerebrovascular/ Primary Ciliary Dyskinesia Panels

Test ID	Test Name	Genes
ARVGG	Arrhythmogenic Cardiomyopathy Gene Panel	<i>CDH2, DES, DSC2, DSG2, DSP, EMD, FLNC, JUP, LMNA, NKX2-5, PKP2, PLN, PPA2, PRKAG2, RBM20, RYR2, TMEM43, TTN</i>
CACMG	Comprehensive Arrhythmia and Cardiomyopathy Gene Panel	<i>ABCC9, ACAD9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, ANK2, BAG3, BRAF, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GLA, GNB5, HCN4, HRAS, JPH2, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ8, KCNQ1, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYLK3, MYPN, NEXN, NKX2-5, NRAS, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SGCD, SHOC2, SLC22A5, SLC4A3, SOS1, SOS2, TAZ (TAFAZZIN), TBX20, TCAP, TECRL, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TTN, TTR, VCL</i>
CAORG	Comprehensive Marfan, Loey's-Dietz, Ehlers-Danlos, and Aortopathy Gene Panel	<i>ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ATP7A, B3GALT6, B3GAT3, B4GALT7, BGN, CBS, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, LOX, LTBP3, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SPARC, TGFB2, TGFB3, TGFB3R1, TGFB3R2, TNXB, ZNF469</i>
CARGG	Comprehensive Arrhythmia Gene Panel	<i>ABCC9, ANK2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, DES, DSC2, DSG2, DSP, EMD, FLNC, GNB5, HCN4, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ8, KCNQ1, LMNA, NKX2-5, PKP2, PLN, PPA2, PRKAG2, RBM20, RYR2, SCN5A, SLC4A3, TECRL, TMEM43, TNNI3K, TRDN, TTN</i>
CCMGG	Comprehensive Cardiomyopathy Gene Panel	<i>ABCC9, ACAD9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, HRAS, JPH2, JUP, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYLK3, MYPN, NEXN, NKX2-5, NRAS, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SGCD, SHOC2, SLC22A5, SOS1, SOS2, TAZ (TAFAZZIN), TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, VCL</i>
CHDGG	Congenital Heart Disease Gene Panel	<i>ACTB, ACTG1, BRAF, CBL, CHD7, CITED2, ELN, FOXF1, FOXH1, GATA4, GATA5, GATA6, GDF1, HRAS, JAG1, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, MYH11, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NR2F2, NRAS, PLD1, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SMAD6, SOS1, SOS2, TAB2, TBX1, TBX20, TBX5, TFAP2B, ZIC3</i>
CPVTG	Catecholaminergic Polymorphic Ventricular Tachycardia Gene Panel	<i>CALM1, CALM2, CALM3, CASQ2, RYR2, TECRL, TRDN</i>
CVHGB	Comprehensive Cerebrovascular Gene Panel	<i>ACTA2, ACVRL1, ADA2, CBS, CCM2, COL3A1, COL4A1, COL4A2, CST3, ENG, EPHB4, GDF2, GLA, GUCY1A1, HTRA1, KRIT1, NOTCH3, PDCD10, RASA1, RNF213, SLC2A10, SMAD2, SMAD3, SMAD4, TEK, TGFB2, TGFB3, TGFB3R1, TGFB3R2, TREX1</i>
DCLNG	Dilated Cardiomyopathy and Left Ventricular Noncompaction Cardiomyopathy Gene Panel	<i>ABCC9, ACAD9, ACADVL, ACTC1, ACTN2, ALMS1, ALPK3, BAG3, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, FKRP, FKTN, FLNC, GAA, GLA, HCN4, JPH2, JUP, LAMP2, LMNA, MYBPC3, MYH7, MYLK3, MYPN, NEXN, NKX2-5, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PRDM16, RAF1, RBM20, RYR2, SCN5A, SGCD, SHOC2, SLC22A5, TAZ (TAFAZZIN), TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL</i>
EDSGG	Ehlers-Danlos Syndrome Gene Panel	<i>ADAMTS2, AEBP1, ATP7A, B3GALT6, B3GAT3, B4GALT7, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, PLOD1, PRDM5, SLC39A13, SPARC, TNXB, ZNF469</i>
HCHLG	Hypercholesterolemia Gene Panel	<i>ABCG5, ABCG8, APOB, APOE, CETP, CYP27A1, LDLR, LDLRAP1, LIPA, LPL, LRP6, PCSK9</i>

Targeted Genes and Methodology Details for Cardiovascular/Connective Tissue/Dyslipidemia/Cerebrovascular/Primary Ciliary Dyskinesia Custom Gene Panel (continued)

Available Cardiovascular/Connective Tissue/Dyslipidemia/ Cerebrovascular/ Primary Ciliary Dyskinesia Panels

Test ID	Test Name	Genes
HCMGG	Hypertrophic Cardiomyopathy Gene Panel	ACAD9, ACADVL, ACTC1, ACTN2, AGL, ALPK3, BRAF, CPT2, CSRP3, ELAC2, FHL1, FLNC, GAA, GLA, HRAS, JPH2, KRAS, LAMP2, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, NEXN, NRAS, PLN, PPA2, PRKAG2, PTPN11, RAF1, RIT1, SHOC2, SLC22A5, SOS1, SOS2, TCAP, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTR, VCL
HHTGG	Hereditary Hemorrhagic Telangiectasia and Vascular Malformations Gene Panel	ACVRL1, BMPR2, CCM2, ENG, EPHB4, GDF2, GLMN, KRIT1, PDCD10, RASA1, SMAD4, TEK
HYPBG	Hypobetalipoproteinemia Gene Panel	ANGPTL3, APOB, MTPP, PCSK9, SAR1B
HYPTG	Hypertriglyceridemia Gene Panel	APOA5, APOC2, APOE, CREB3L3, GPD1, GPIHBP1, LCAT, LIPA, LIPC, LMF1, LPL, LRP6
LIPOG	Lipodystrophy Gene Panel	AGPAT2, BSCL2, CAV1, CAVIN1, FBN1, KCNJ6, LIPE, LMNA, PIK3R1, PLIN1, PPARG, ZMPSTE24
LQTSG	Long QT Syndrome Gene Panel	CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN
MFBNG	FBN1 Full Gene Sequencing with Deletion/Duplication	FBN1
MFRGG	Marfan, Loeys-Dietz, and Aortopathy Gene Panel	ACTA2, ADAMTS10, ADAMTS17, BGN, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FLNA, LOX, LTBP3, MED12, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBRI1, TGFBRI2
NSRGG	Noonan Syndrome and Related Conditions Gene Panel	ACTB, ACTG1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1
OIBFG	Osteogenesis Imperfecta and Bone Fragility Gene Panel	ALPL, ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, WNT1, XYLT2
PCDGG	Primary Ciliary Dyskinesia Gene Panel	ARMC4 (ODAD2), CCDC103, CCDC114 (ODAD1), CCDC151 (ODAD3), CCDC39, CCDC40, CCDC65, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS8, LRRC6 (DNAAF11), MCIDAS, OFD1, PIH1D3 (DNAAF6), RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC25 (ODAD4), ZMYND10
PRKSG	PRKAR1A Full Gene Sequencing with Deletion/Duplication	PRKAR1A
SCN5A	Brugada Syndrome, SCN5A Full Gene Analysis	SCN5A
SQTSG	Short QT Syndrome Gene Panel	KCNH2, KCNJ2, KCNQ1, SLC4A3

Effective Date	Version	Synopsis of Test Change
6/4/2025	V2	<ul style="list-style-type: none"> Added Additional Evaluations for <i>BMPR2, BSCL2, COL1A1, COL5A1, DMD, ENG, FBN1, FBN2, GAA, GLA, JAG1, KRIT1, LCAT, LIPC, LMNA, LZTR1, MTPP, MYBPC3, OFD1, SERPINF1, and SMAD2</i> Added Technical Limitations for <i>ARMC4 (ODAD2), CACNA2D1, DNAJB13, DNAL1, GLMN, LCAT, LPL, LRRC6 (DNAAF11), PLOD2, PPA2, PRKAG2, TBX20, TNXB, and TRDN</i> Updated Technical Limitations for <i>TTC25 (ODAD4)</i> Removed Technical Limitations for <i>DNAH11, DNAI1, GDF1, SOS2, and TECRL</i> Added list of available panels