

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 November 2022 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	chr9:107567035A>G (c.4465-34A>G) chr9:107599404G>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	chr1:100381954A>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.

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>APOA1</i>	NM_000039.2	-	-
<i>APOA5</i>	NM_052968.5	-	-
<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	-	-
<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	-	-
<i>BRAF</i>	NM_004333.6	-	-
<i>BSCL2</i>	NM_032667.6	-	-
<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 exon 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	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CCDC114 (ODAD1)</i>	NM_144577.4	-	-
<i>CCDC151 (ODAD3)</i>	NM_145045.5	-	-
<i>CCDC39</i>	NM_181426.2	-	-
<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	chr8:61763035G>A (c.5405-17G>A)	-
<i>CHRM2</i>	NM_000739.2	-	-
<i>CHST14</i>	NM_130468.3	-	-
<i>CITED2</i>	NM_006079.5	-	-
<i>COL12A1</i>	NM_004370.6	-	-
<i>COL1A1</i>	NM_000088.3	chr17:48272201C>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	-	-
<i>COL5A2</i>	NM_000393.5	-	CNV may not be detected in exon 3.
<i>CPT2</i>	NM_000098.3	-	-
<i>CREB3L1</i>	NM_052854.4	-	-
<i>CREB3L3</i>	NM_032607.3	-	-
<i>CRTAP</i>	NM_006371.5	chr3:33160815C>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:33192452C>T (c.31+36947G>A) chrX:31279780T>C, (c.9225-647A>G) chrX: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>DNAH11</i>	NM_001277115.2	-	CNV may not be detected in exon 55.
<i>DNAH5</i>	NM_001369.2	-	-
<i>DNAH8</i>	NM_001206927.2	-	-
<i>DNAH9</i>	NM_001372.4	-	-
<i>DNAI1</i>	NM_012144.4	-	CNV may not be detected in exon 5.
<i>DNAI2</i>	NM_023036.6	-	-
<i>DNAJB13</i>	NM_153614.3	-	-
<i>DNAJC19</i>	NM_145261.4	-	-
<i>DNAL1</i>	NM_031427.4	-	-
<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	chr9:130616761G>A (c.-127C>T)	-
<i>EPHB4</i>	NM_004444.5	-	-
<i>FBN1</i>	NM_000138.4	chr15:48721629T>C (c.6872-961A>G)	-
<i>FBN2</i>	NM_001999.4	-	-
<i>FHL1</i>	NM_001449.5	-	-
<i>FKBP10</i>	NM_021939.4	-	-
<i>FKBP14</i>	NM_017946.4	-	-
<i>FKRP</i>	NM_024301.5	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>FKTN</i>	NM_001079802.1	chr9:108368857G>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	chr17:78078341T>G- (c. -32-13T>G) chr17:78082266T>G (c.1076-22T>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	-	CNV may not be detected for exon 8.
<i>GDF2</i>	NM_016204.4	-	-
<i>GLA</i>	NM_000169.2	chrX:100654735C>T (c.640-801G>A)	-
<i>GLMN</i>	NM_053274.3	-	CNV may not be detected in exons 2, 10 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	chr11:299504G>A- (c.-14C>T)	-
<i>ILK</i>	NM_004517.4	-	-
<i>JAG1</i>	NM_000214.3	-	-
<i>JPH2</i>	NM_020433.4	-	-
<i>JUP</i>	NM_002230.4	-	-
<i>KCND2</i>	NM_012281.3	-	-
<i>KCND3</i>	NM_004980.4	-	-
<i>KCNE1</i>	NM_000219.6	-	-
<i>KCNE2</i>	NM_172201.1	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<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	-	-
<i>LAMA4</i>	NM_002290.5	-	-
<i>LAMP2</i>	NM_013995.2	-	-
<i>LCAT</i>	NM_000229.2	-	-
<i>LDB3</i>	NM_007078.3	-	-
<i>LDB3</i>	NM_001080116.1	-	-
<i>LDLR</i>	NM_000527.5	chr19:11200029_11200044 (c.-196_c.-181) (Repeat 1 Sp1 binding site) chr19:11200064_11200079 (c.-161_c.-146) (Repeat 2 SREBP binding site) chr19:11200080_11200095 (c.-145_c.-130) (Repeat 3 Sp1 binding site) chr19:11231284C>G (c.2140+86C>G) chr19:11231301G>T (c.2140+103G>T)	-
<i>LDLRAP1</i>	NM_015627.3	-	-
<i>LIPA</i>	NM_000235.4	-	-
<i>LIPC</i>	NM_000236.3	-	-
<i>LIPE</i>	NM_005357.4	-	-
<i>LMF1</i>	NM_022773.4	-	-
<i>LMNA</i>	NM_170707.4	-	-
<i>LOX</i>	NM_002317.7	-	-
<i>LPL</i>	NM_000237.3	-	CNV may not be detected in exon 10.
<i>LRP5</i>	NM_002335.4	-	-
<i>LRP6</i>	NM_002336.3	-	-
<i>LRR6 (DNAAF1)</i>	NM_012472.6	-	-
<i>LTBP3</i>	NM_001130144.2	-	-
<i>LZTR1</i>	NM_006767.4	-	-
<i>MAP2K1</i>	NM_002755.3	-	-
<i>MAP2K2</i>	NM_030662.3	-	-
<i>MBTPS2</i>	NM_015884.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>MCIDAS</i>	NM_001190787.3	-	-
<i>MED12</i>	NM_005120.3	-	-
<i>MED13L</i>	NM_015335.4	-	-
<i>MEGF8</i>	NM_001271938.2	-	-
<i>MEIS2</i>	NM_170674.5	-	-
<i>MFAP5</i>	NM_003480.4	-	-
<i>MRAS</i>	NM_012219.4	-	-
<i>MTO1</i>	NM_012123.4	-	-
<i>MTTP</i>	NM_000253.3	-	-
<i>MYBPC3</i>	NM_000256.3	chr11:47368616C>T (c.906-36G>A) chr11:47364832C>T (c.1224-19G>A) chr11:47364834T>C (c.1224-21A>G) chr11:47364865C>T (c.1224-52G>A) chr11:47364709C>T (c.1227-13G>A) chr11:47361954G>A (c.1927+600C>T)	CNV may not be detected in exon 10.
<i>MYH11</i>	NM_001040113.2	-	-
<i>MYH6</i>	NM_002471.3	-	-
<i>MYH7</i>	NM_000257.4	-	-
<i>MYL2</i>	NM_000432.3	-	-
<i>MYL3</i>	NM_000258.3	-	-
<i>MYLK</i>	NM_053025.4	-	-
<i>MYLK3</i>	NM_182493.3	-	-
<i>MYPN</i>	NM_032578.3	-	-
<i>NEXN</i>	NM_144573.3	-	-
<i>NKX2-5</i>	NM_004387.4	-	-
<i>NKX2-6</i>	NM_001136271.2	-	-
<i>NODAL</i>	NM_018055.5	-	-
<i>NOTCH1</i>	NM_017617.5	-	-
<i>NOTCH2</i>	NM_024408.4	-	Sequence and CNV analyses for exons 1-4 will not be performed.
<i>NOTCH3</i>	NM_000435.3	-	-
<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	-	-
<i>P3H1</i>	NM_022356.4	chr1:43212925C>T (c.2055+18G>A)	-

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<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 for 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	-	-
<i>PLS3</i>	NM_005032.7	chrX:114856534T>A- (c.74-24T>A)	-
<i>PPA2</i>	NM_176869.3	-	-
<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	-	-
<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	-	-
<i>RNF213</i>	NM_001256071.3	-	-
<i>RPGR</i>	NM_000328.3	-	-
<i>RRAS</i>	NM_006270.5	-	-
<i>RRAS2</i>	NM_012250.6	-	-

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<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	chr2:166917341G>A (c.265-2143G>A) chr2:166863778G>C (c.4002+2451G>C)	-
<i>SCN5A</i>	NM_198056.2	-	-
<i>SEC24D</i>	NM_014822.4	-	-
<i>SERPINF1</i>	NM_002615.7	-	-
<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>SLC22A5</i>	NM_003060.4	chr5:131705516G>A (c.-149G>A) chr5:131714054T>A (c.394-16T>A) chr5:131722665G>A c.825-52G>A	-
<i>SLC2A10</i>	NM_030777.4	-	-
<i>SLC39A13</i>	NM_152264.4	-	-
<i>SLC4A3</i>	NM_201574.2	-	-
<i>SMAD2</i>	NM_005901.6	-	-
<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	-	CNV may not be detected in exon 18.
<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>TAZ (TAFAZZIN)</i>	NM_000116.5	-	-
<i>TAPT1</i>	NM_153365.3	-	-
<i>TBX1</i>	NM_080647.1	-	-

**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>TBX20</i>	NM_001077653.2	-	-
<i>TBX5</i>	NM_000192.3	-	-
<i>TCAP</i>	NM_003673.4	-	-
<i>TECRL</i>	NM_001010874.5	-	CNV may not be detected in exon 12.
<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	-	-
<i>TPM1</i>	NM_001018005.2	-	-
<i>TRDN</i>	NM_006073.4	chr6:123957870T>C- (c.22+29A>G) chr6:123850462C>T (c.484+1189G>A)	CNV may not be detected in exons 7 and 22.
<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 9–12 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	-	-