MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Comprehensive Cardiomyopathy Gene Panel

The following applies to CCMGG / Comprehensive Cardiomyopathy 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
ABCC9	NM_005691.3	-	-
ACAD9	NM_014049.5	-	-
ACADVL	NM_000018.4	-	-
ACTC1	NM_005159.5	-	-
ACTN2	NM_001103.3	-	-
AGL	NM_000642.3	chr1:g.100381954A>G (c.4260-12A>G)	-
ALMS1	ENST00000264448.6	-	-
ALPK3	NM_020778.4	-	-
BAG3	NM_004281.3	-	-
BRAF	NM_004333.6	-	-
CDH2	NM_001792.5	-	-
CPT2	NM_000098.3	-	-
CRYAB	NM_001885.3	-	-
CSRP3	NM_003476.5	-	-
DES	NM_001927.4	-	-
DMD	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)	-
DNAJC19	NM_145261.4	-	-
DOLK	NM_014908.4	-	-
DSC2	NM_024422.6	-	-
DSG2	NM_001943.5	-	-
DSP	NM_004415.4	-	-
ELAC2	NM_018127.7	-	-
EMD	NM_000117.3	-	-
FHL1	NM_001449.5	-	-
FKRP	NM_024301.5	-	-
FKTN	NM_001079802.1	chr9:g.108368857G>T (c.648-1243G>T); ~3 kb retrotransposon insertion (c.*4392_*4393)	-
FLNC	NM_001458.4	-	-

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GAA	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)	-
GLA	NM_000169.2	chrX:g.100654735C>T (c.640-801G>A); chrX:g.100653945A>T (c.640-11T>A)	-
HCN4	NM_005477.3	-	-
HRAS	NM_005343.4	-	-
JPH2	NM_020433.4	-	-
JUP	NM_002230.4	-	-
KRAS	NM_004985.5	-	-
LAMP2	NM_013995.2	-	-
LMNA	NM_170707.4	chr1:g.156105681C>G (c.937-11C>G)	-
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	-	-
MRAS	NM_012219.4	-	-
MTO1	NM_012123.4	-	-
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
MYH7	NM_000257.4	-	-
MYL2	NM_000432.3	-	-
MYL3	NM_000258.3	-	-
MYLK3	NM_182493.3	-	-
MYPN	NM_032578.3	-	-
NEXN	NM_144573.3	-	-
NKX2-5	NM_004387.4	-	-
NRAS	NM_002524.5	-	-
PCCA	NM_000282.4	-	CNV may not be detected in exon 10
PCCB	NM_000532.5	-	-
PKP2	NM_004572.3	-	-
PLN	NM_002667.5	-	-

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PPA2	NM_176869.3	-	CNV may not be detected in exon 11
PPCS	NM_024664.4	-	-
PRDM16	NM_022114.4	-	CNV may not be detected in exon 1
PRKAG2	NM_016203.4	-	CNV may not be detected in exon 13
PTPN11	NM_002834.4	-	-
RAF1	NM_002880.3	-	-
RBM20	NM_001134363.3	-	-
RIT1	NM_006912.6	-	-
RYR2	NM_001035.3	-	CNV may not be detected in exons 71, 96, and 104
SCN5A	NM_198056.2	-	-
SGCD	NM_000337.5	-	-
SHOC2	NM_007373.3	-	-
SLC22A5	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)	-
SOS1	NM_005633.3	-	-
SOS2	NM_006939.4	-	-
TAZ (TAFAZZIN)	NM_000116.5	-	-
TBX20	NM_001077653.2	-	CNV may not be detected in exons 5–8
TCAP	NM_003673.4	-	-
TMEM43	NM_024334.2	-	-
TMEM70	NM_017866.6	-	-
TNNC1	NM_003280.3	-	-
TNNI3	NM_000363.5	-	-
TNNI3K	NM_015978.3	-	CNV may not be detected in exon 1
TNNT2	NM_001001430.3	-	-
TPM1	NM_001018005.2	-	-
TRIM63	NM_032588.3	-	-
TTN	NM_001256850.1	-	Sequence and CNV analyses for exons 154–156 will not be performed
TTR	NM_000371.3	-	-
VCL	NM_014000.2	-	-

Effective Date	Version	Synopsis of Test Change
6/4/2025	V2	• Added Additional Evaluation for *DMD, GAA, GLA, LMNA, LZTR1, MYBPC3* • Added Technical Limitations for *MYBPC3, PPA2, PRKAG2,* and *TBX20* • Removed Technical Limitation from *SOS2*