MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Comprehensive Arrhythmia Gene Panel

The following applies to CARGG / Comprehensive Arrhythmia 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	-	-
ANK2	NM_020977.4	-	-
CACNA1C	NM_000719.7	-	-
CACNA1D	NM_000720.4	-	-
CACNA2D1	NM_000722.4	-	CNV may not be detected in exon 20
CACNB2	NM_201590.3	-	-
CALM1	NM_006888.6	-	-
CALM2	NM_001743.6	-	-
CALM3	NM_005184.4	-	-
CASQ2	NM_001232.3	-	-
CAV3	NM_033337.3	-	-
CDH2	NM_001792.5	-	-
DES	NM_001927.4	-	-
DSC2	NM_024422.6	-	-
DSG2	NM_001943.5	-	-
DSP	NM_004415.4	-	-
EMD	NM_000117.3	-	-
FLNC	NM_001458.4	-	-
GNB5	NM_016194.4	-	CNV may not be detected in exon 11
HCN4	NM_005477.3	-	-
JUP	NM_002230.4	-	-
KCND2	NM_012281.3	-	-
KCND3	NM_004980.4	-	-
KCNE1	NM_000219.6	-	-
KCNE2	NM_172201.1	-	-
KCNH2	NM_000238.4	-	-
KCNJ2	NM_000891.3	-	-

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Targeted Genes and Methodology Details for Comprehensive Arrhythmia Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
KCNJ8	NM_004982.4	-	-
KCNQ1	NM_000218.3	-	-
LMNA	NM_170707.4	-	-
NKX2-5	NM_004387.4	-	-
PKP2	NM_004572.3	-	-
PLN	NM_002667.5	-	-
PPA2	NM_176869.3	-	-
PRKAG2	NM_016203.4	-	-
RBM20	NM_001134363.3	-	-
RYR2	NM_001035.3	-	CNV may not be detected in exons 71, 96, and 104
SCN5A	NM_198056.2	-	-
SLC4A3	NM_201574.2	-	-
TECRL	NM_001010874.5	-	CNV may not be detected in exon 12
TMEM43	NM_024334.2	-	-
TNNI3K	NM_015978.3	-	CNV may not be detected in exon 1
TRDN	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
TTN	NM_001256850.1	-	Sequence and CNV analyses for exons 154–156 will not be performed