MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Arrhythmogenic Cardiomyopathy Gene Panel

The following applies to ARVGG / Arrhythmogenic 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
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	-	-
JUP	NM_002230.4	-	-
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 in exons 71, 96, and 104 may not be detected
TMEM43	NM_024334.2	-	-
TTN	NM_001256850.1	-	Analyses for sequence variants and CNV are not performed for exons 154–156