MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Congenital Heart Disease Gene Panel

The following applies to CHDGG / Congenital Heart Disease 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 numbers 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, or 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
ACTB	NM_001101.5	-	-
ACTG1	NM_001614.5	-	-
BRAF	NM_004333.6	-	-
CBL	NM_005188.4	-	-
CHD7	NM_017780.4	chr8:61763035G>A (c.5405-17G>A)	-
CITED2	NM_006079.5	-	-
ELN	NM_001278939.1	-	-
FOXF1	NM_001451.3	-	-
FOXH1	NM_003923.3	-	-
GATA4	NM_002052.5	-	-
GATA5	NM_080473.5	-	-
GATA6	NM_005257.5	-	-
GDF1	NM_001492.6	-	CNV may not be detected for exon 8.
HRAS	NM_005343.4	-	-
JAG1	NM_000214.3	-	-
KRAS	NM_004985.5	-	-
LZTR1	NM_006767.4	-	-
MAP2K1	NM_002755.3	-	-
MAP2K2	NM_030662.3	-	-
MRAS	NM_012219.4	-	-
MYH11	NM_001040113.2	-	-
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.
NR2F2	NM_021005.4	-	-
NRAS	NM_002524.5	-	-

Targeted Genes and Methodology Details for Congenital Heart Disease Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PLD1	NM_002662.5	-	CNV may not be detected for exon 6.
PPP1CB	NM_206876.1	-	-
PTPN11	NM_002834.4	-	-
RAF1	NM_002880.3	-	-
RIT1	NM_006912.6	-	-
RRAS2	NM_012250.6	-	-
SHOC2	NM_007373.3	-	-
SMAD6	NM_005585.5	-	-
SOS1	NM_005633.3	-	-
SOS2	NM_006939.4	-	CNV may not be detected for exon 18.
TAB2	NM_015093.5	-	-
TBX1	NM_080647.1	-	-
TBX20	NM_001077653.2	-	-
TBX5	NM_000192.3	-	-
TFAP2B	NM_003221.4	-	-
ZIC3	NM_003413.4	-	-