WES CONGENITAL HEARTDISEASE [1] DG 3.3

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ABL1	241.0	100.0	100.0	-
ACTC1	217.1	100.0	100.0	612794;613424;612098;613424
ACVR2B	173.3	100.0	100.0	613751
ALDH1A2	145.4	100.0	100.0	-
ANKRD1	143.5	100.0	100.0	-
BRAF	185.7	100.0	100.0	115150;613707;613706
CCDC114	172.5	100.0	100.0	-
CFAP53	150.0	100.0	100.0	614779
CFC1	274.6	100.0	100.0	605376
CHD7	180.1	100.0	100.0	214800;612370
CITED2	191.3	100.0	100.0	614433;614431
CRELD1	164.9	100.0	100.0	606217
EHMT1	178.9	99.9	99.7	610253
ELN	215.1	100.0	100.0	123700;185500
FBN1	157.9	100.0	100.0	616914;154700
FLT4	227.0	100.0	100.0	618780
FOXH1	219.3	100.0	100.0	-
FOXL1	241.9	100.0	100.0	-
GATA4	188.2	100.0	100.0	615542;607941;614430;187500;614429
GATA5	204.9	100.0	100.0	617912
GATA6	192.2	100.0	100.0	614475;614474;600001;217095;187500
GDF1	156.1	100.0	99.8	613854;208530
GJA1	204.2	100.0	100.0	600309;241550
GJA5	216.3	100.0	100.0	614049;108770
HAND1	180.6	100.0	100.0	-

HAND2	188.4	100.0	100.0	-
HEY2	212.5	100.0	100.0	-
JAG1	180.2	100.0	100.0	118450;187500
KDR	194.7	100.0	100.0	No OMIM phenotype
KMT2D	223.4	100.0	100.0	147920
KRAS	171.7	100.0	100.0	615278;609942
LEFTY2	250.6	100.0	100.0	-
MCTP2	145.3	100.0	100.0	-
MED13L	175.0	100.0	100.0	608808
MMP21	182.5	100.0	100.0	616749
MYH11	221.9	100.0	100.0	132900;619351;619350
MYH6	206.7	100.0	100.0	614089;613252;613251;614090
MYH7	200.1	100.0	100.0	613426;192600;160500;613426;608358;255160;181430
MYRF	180.7	100.0	100.0	618280
NAA15	125.6	96.8	96.8	No OMIM phenotype
NKX2-5	233.9	100.0	100.0	108900;217095;614435;225250;187500;614432
NKX2-6	218.3	100.0	100.0	217095;217095
NODAL	192.5	100.0	100.0	270100
NOTCH1	267.2	100.0	100.0	616028;109730
NOTCH2	247.6	100.0	100.0	610205
NR2F2	234.9	100.0	100.0	615779
PKD1L1	165.9	100.0	100.0	617205
PLD1	157.2	100.0	100.0	212093
PRKD1	165.9	100.0	100.0	617364
PTPN11	196.6	100.0	100.0	163950;151100
RAF1	193.0	100.0	100.0	615916;611554;611553
SHROOM3	208.6	100.0	100.0	604570
SMAD6	186.5	100.0	100.0	614823
SOS1	137.5	100.0	100.0	610733
SRF	177.8	100.0	100.0	No OMIM phenotype

TAB2	177.7	100.0	100.0	614980
TAF1	155.0	100.0	100.0	-
TBX1	144.8	97.3	94.9	217095;188400;187500;192430
TBX20	171.0	100.0	100.0	611363
TBX5	180.6	100.0	100.0	142900
TDGF1	178.3	100.0	100.0	-
TFAP2B	197.0	100.0	100.0	169100;617035
TLL1	148.8	100.0	100.0	613087
TMEM260	164.1	100.0	100.0	617478
TNNI3K	156.5	100.0	100.0	616117
ZFPM2	187.7	100.0	100.0	187500
ZIC3	212.2	100.0	100.0	306955

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.
Median Coverage describes the average number of reads seen across 50 exomes.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with no value for coverage are non protein coding genes.
Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors