WES CONGENITAL HEARTDISEASE [1] DG 3.5

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ABL1	113.0	100.0	100.0	-
ACTC1	185.7	100.0	100.0	612794;613424;612098;613424
ACVR2B	130.1	100.0	100.0	613751
ADNP	141.3	100.0	100.0	No OMIM phenotype
ALDH1A2	135.4	100.0	99.9	-
ANKRD1	134.7	100.0	99.9	-
ANKRD11	122.6	100.0	100.0	No OMIM phenotype
BRAF	139.7	100.0	100.0	115150;613707;613706
CACNA1C	132.6	100.0	100.0	No OMIM phenotype
CCDC114	136.4	100.0	100.0	-
CFAP53	138.2	100.0	100.0	614779
CFC1	217.5	100.0	100.0	605376
CHD4	133.0	100.0	100.0	617159
CHD7	133.9	100.0	100.0	214800;612370
CITED2	132.7	100.0	100.0	614433;614431
COL3A1	146.7	100.0	100.0	130050
CRELD1	133.4	100.0	100.0	606217
DYRK1A	147.1	100.0	100.0	No OMIM phenotype
EHMT1	130.8	100.0	99.9	610253
ELN	132.1	100.0	100.0	123700;185500
FBN1	149.7	100.0	100.0	616914;154700
FBN2	141.3	100.0	100.0	No OMIM phenotype
FLT4	125.0	100.0	100.0	618780
FOXH1	124.0	100.0	100.0	-
FOXL1	150.6	100.0	100.0	-

GATA4	164.3	100.0	100.0	615542;607941;614430;187500;614429
GATA5	147.5	100.0	100.0	617912
GATA6	147.3	100.0	100.0	614475;614474;600001;217095;187500
GDF1	128.7	100.0	100.0	613854;208530
GJA5	125.2	100.0	100.0	614049;108770
GLYR1	124.5	100.0	100.0	No OMIM phenotype
HAND1	120.5	100.0	100.0	-
HAND2	136.0	100.0	100.0	-
HEY2	136.1	100.0	100.0	-
JAG1	137.5	100.0	100.0	118450;187500
KAT6B	137.4	100.0	100.0	605880;603736
KDR	138.5	100.0	100.0	No OMIM phenotype
KMT2A	136.1	100.0	100.0	No OMIM phenotype
KMT2D	124.8	100.0	100.0	147920
KRAS	161.6	100.0	100.0	615278;609942
LEFTY2	200.9	100.0	100.0	-
MCTP2	139.6	100.0	99.9	-
MED13L	131.8	100.0	99.6	608808
MMP21	151.8	100.0	100.0	616749
MUC16	150.6	100.0	100.0	No OMIM phenotype
MYH11	129.0	100.0	100.0	132900;619351;619350
MYH6	149.9	100.0	100.0	614089;613252;613251;614090
MYH7	152.3	100.0	100.0	613426;192600;160500;613426;608358;255160;181430
MYRF	114.4	100.0	100.0	618280
NAA15	142.5	96.6	96.6	No OMIM phenotype
NF1	147.4	100.0	100.0	162210;162200;601321;193520
NKX2-5	144.1	100.0	100.0	108900;217095;614435;225250;187500;614432
NKX2-6	166.2	100.0	100.0	217095;217095
NODAL	117.9	100.0	100.0	270100
NOTCH1	130.3	100.0	100.0	616028;109730

NOTCH2	149.2	100.0	100.0	610205
NR2F2	164.1	100.0	100.0	615779
NSD1	126.6	100.0	100.0	No OMIM phenotype
PKD1L1	133.0	100.0	100.0	617205
PLD1	135.3	100.0	100.0	212093
PRKD1	140.1	100.0	100.0	617364
PTPN11	144.8	100.0	100.0	163950;151100
RAF1	133.3	100.0	100.0	615916;611554;611553
RBFOX2	140.4	100.0	100.0	No OMIM phenotype
ROBO4	122.2	100.0	100.0	618496
SHROOM3	121.0	100.0	100.0	604570
SMAD6	139.0	100.0	100.0	614823
SMARCA4	125.2	100.0	100.0	614609
SOS1	145.6	100.0	100.0	610733
SRF	136.6	100.0	100.0	No OMIM phenotype
TAB2	154.3	100.0	100.0	614980
TAF1	100.3	100.0	99.9	-
TBX1	117.8	97.7	95.5	217095;188400;187500;192430
TBX20	136.1	100.0	100.0	611363
TBX5	141.1	100.0	100.0	142900
TDGF1	142.1	100.0	100.0	-
TFAP2B	133.6	100.0	100.0	169100;617035
TLL1	142.4	99.4	98.9	613087
TMEM260	152.9	100.0	100.0	617478
TNNI3K	153.6	100.0	100.0	616117
TSC1	125.8	100.0	100.0	191100
ZFPM2	135.6	100.0	100.0	187500
ZIC3	118.4	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.