

WES CONGENITAL HEARTDISEASE ¹ DG 3.4

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABL1	233.2	100.0	100.0	-
ACTC1	218.1	100.0	100.0	612794;613424;612098;613424
ACVR2B	180.0	100.0	100.0	613751
ALDH1A2	153.7	100.0	100.0	-
ANKRD1	147.5	100.0	100.0	-
BRAF	174.2	100.0	100.0	115150;613707;613706
CCDC114	187.2	100.0	100.0	-
CFAP53	158.8	100.0	100.0	614779
CFC1	293.2	100.0	100.0	605376
CHD7	185.7	100.0	100.0	214800;612370
CITED2	203.1	100.0	100.0	614433;614431
CRELD1	176.3	100.0	100.0	606217
EHMT1	189.1	99.9	99.8	610253
ELN	221.0	100.0	100.0	123700;185500
FLT4	224.1	100.0	100.0	618780
FOXH1	233.0	100.0	100.0	-
FOXL1	259.6	100.0	100.0	-
GATA4	203.9	100.0	100.0	615542;607941;614430;187500;614429
GATA5	222.3	100.0	100.0	617912
GATA6	207.5	100.0	100.0	614475;614474;600001;217095;187500
GDF1	178.3	100.0	100.0	613854;208530
GJA5	231.3	100.0	100.0	614049;108770
GLYR1	161.8	100.0	100.0	No OMIM phenotype
HAND1	201.6	100.0	100.0	-
HAND2	207.1	100.0	100.0	-

HEY2	219.1	100.0	100.0	-
JAG1	188.5	100.0	100.0	118450;187500
KDR	181.9	100.0	100.0	No OMIM phenotype
KMT2D	227.9	100.0	100.0	147920
KRAS	158.7	100.0	100.0	615278;609942
LEFTY2	271.7	100.0	100.0	-
MCTP2	150.6	100.0	100.0	-
MED13L	185.7	100.0	100.0	608808
MMP21	196.9	100.0	100.0	616749
MUC16	213.8	100.0	100.0	No OMIM phenotype
MYH11	229.4	100.0	100.0	132900;619351;619350
MYH6	218.5	100.0	100.0	614089;613252;613251;614090
MYH7	212.1	100.0	100.0	613426;192600;160500;613426;608358;255160;181430
MYRF	195.2	100.0	100.0	618280
NAA15	130.8	96.8	96.8	No OMIM phenotype
NKX2-5	251.2	100.0	100.0	108900;217095;614435;225250;187500;614432
NKX2-6	237.9	100.0	100.0	217095;217095
NODAL	205.7	100.0	100.0	270100
NOTCH1	259.6	100.0	100.0	616028;109730
NOTCH2	240.8	100.0	100.0	610205
NR2F2	252.8	100.0	100.0	615779
PKD1L1	175.5	100.0	100.0	617205
PLD1	164.1	100.0	100.0	212093
PRKD1	171.0	100.0	100.0	617364
PTPN11	185.6	100.0	100.0	163950;151100
RAF1	181.9	100.0	100.0	615916;611554;611553
SHROOM3	218.5	100.0	100.0	604570
SMAD6	203.7	100.0	100.0	614823
SOS1	144.0	100.0	100.0	610733
SRF	195.0	100.0	100.0	No OMIM phenotype

TAB2	183.1	100.0	100.0	614980
TAF1	156.6	100.0	100.0	-
TBX1	158.0	98.1	95.9	217095;188400;187500;192430
TBX20	175.9	100.0	100.0	611363
TBX5	195.1	100.0	100.0	142900
TDGF1	186.6	100.0	100.0	-
TFAP2B	209.8	100.0	100.0	169100;617035
TLL1	154.2	100.0	100.0	613087
TMEM260	171.6	100.0	100.0	617478
TNNI3K	160.1	100.0	100.0	616117
ZFPM2	194.5	100.0	100.0	187500
ZIC3	217.2	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.