

# WES CONGENITAL HEARTDISEASE <sup>1</sup> DG 3.1

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ACTC1	119.1	100.0	99.7	102540
ACVR2B	133.0	98.3	95.0	602730
ALDH1A2	125.6	99.9	98.5	603687
ANKRD1	129.3	100.0	99.4	609599
BRAF	80.6	91.0	81.1	164757
CFAP53	150.5	99.6	97.4	614759
CFC1	122.7	84.2	74.1	605194
CHD7	158.7	100.0	99.5	608892
CITED2	111.9	99.2	99.0	602937
CRELD1	103.8	99.9	95.0	607170
EHMT1	138.6	94.5	93.7	607001
ELN	118.7	99.8	97.8	130160
FBN1	169.7	100.0	99.9	134797
FLT4	154.8	99.2	98.3	136352
FOXC2	99.8	100.0	96.7	602402
FOXH1	65.6	100.0	96.5	603621
FOXL1	106.3	96.6	89.0	603252
GATA4	80.9	84.1	74.5	600476
GATA5	57.8	99.7	93.7	611496
GATA6	94.1	89.8	83.0	601656
GDF1	28.8	73.9	54.0	602880
GJA1	187.7	100.0	100.0	600309;241550
GJA5	229.0	100.0	100.0	121013
HAND1	115.0	100.0	100.0	602406
HAND2	59.8	99.8	92.6	602407

HEY2	187.8	100.0	99.3	604674
JAG1	147.4	97.7	96.8	601920
KMT2D	142.1	100.0	99.4	602113
KRAS	84.3	99.5	96.9	190070
LEFTY2	51.9	88.9	81.4	601877
MCTP2	146.5	99.7	98.2	616297
MED13L	136.5	100.0	99.8	608771
MMP21	110.1	99.9	98.8	608416
MYH11	138.0	100.0	100.0	160745
MYH6	108.4	99.4	97.1	160710
MYH7	103.2	99.6	97.3	160760
MYRF	146.9	99.3	98.5	618280
NAA15	105.4	95.8	91.0	No OMIM phenotype
NKX2-5	86.7	100.0	99.7	600584
NKX2-6	128.9	100.0	99.5	611770
NODAL	167.9	100.0	100.0	601265
NOTCH1	121.7	99.2	97.2	190198
NOTCH2	146.7	100.0	99.5	610205
NR2F2	221.5	100.0	98.5	107773
PKD1L1	130.3	100.0	99.8	609721
PLD1	142.7	100.0	99.6	602382
PTPN11	100.3	99.1	93.7	176876
RAF1	125.8	100.0	100.0	164760
SHROOM3	156.0	98.6	97.8	604570
SMAD6	149.8	90.9	81.0	602931
SOS1	123.6	99.8	98.4	182530
TAB2	212.6	100.0	99.7	605101
TBX1	89.7	87.0	77.5	602054
TBX20	120.7	100.0	99.7	606061
TBX5	141.7	100.0	100.0	601620

TDGF1	157.7	99.9	96.7	187395
TFAP2B	166.7	99.9	98.6	601601
TLL1	160.1	100.0	100.0	606742
TNNI3K	128.4	100.0	99.4	613932
ZFPM2	181.3	100.0	100.0	603693
ZIC3	129.6	100.0	99.9	300265

*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.*

*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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.*

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