

WES HEART DISORDERS¹ DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AARS2	131.7	100.0	99.4	614096
ABCC6	110.8	93.6	92.5	614473
ABCC9	145.1	100.0	99.8	614050
ABL1	148.2	100.0	100.0	617602
ACAD8	132.5	100.0	100.0	611283
ACAD9	124.7	100.0	99.8	611126
ACADVL	124.7	99.7	96.6	201475
ACSF3	145.9	100.0	99.5	614265
ACTA2	98.9	100.0	98.9	611788
ACTC1	110.7	99.9	98.9	612794;613424;612098;613424
ACTN2	132.4	100.0	100.0	102573
ACVR2B	124.3	98.6	95.1	613751
ADAMTS19	117.5	95.7	92.6	-
ADCY5	133.1	95.9	92.5	606703
AGK	104.4	90.4	87.9	212350;614691
AGL	153.8	99.8	99.5	232400
AGPAT2	147.6	99.0	94.0	608594
AKAP9	85.1	98.4	94.6	611820
ALDH1A2	104.1	100.0	99.0	603687
ALMS1	174.7	99.7	99.5	203800
ALPK3	108.0	98.1	95.1	618052
ANK2	129.7	100.0	99.9	600919
ANKRD1	106.5	99.9	98.3	609599
ARIH1	104.3	99.7	99.3	-
ATPAF2	99.7	100.0	99.9	604273

BAG3	167.9	100.0	99.7	613881
BANF1	43.9	95.3	78.1	614008
BGN	146.9	100.0	99.9	300106;300989
BMPR2	162.0	99.9	99.9	178600
BRAF	63.9	89.4	77.6	211980
BSCL2	104.6	100.0	99.9	269700
BVES	117.4	99.4	98.6	616812
CACNA1C	151.1	99.9	99.2	611875
CACNA1D	133.5	97.9	97.7	614896
CACNA2D1	105.5	99.0	96.0	114204
CACNB2	148.8	98.5	98.4	611876
CALM1	103.4	99.9	97.3	614916
CALM2	42.5	66.5	59.7	114182
CALM3	108.7	100.0	99.2	114183
CASQ2	125.1	100.0	99.8	611938
CASZ1	107.1	96.5	93.2	No OMIM phenotype
CAV1	181.8	100.0	100.0	606721;612526;615343
CAV3	222.9	100.0	100.0	607801;192600
CCDC114	138.5	100.0	99.8	615067
CDH2	119.0	99.4	97.1	-
CFAP53	136.1	99.3	96.6	614779
CFC1	121.3	85.0	78.0	613853;217095;605376
CHD7	137.1	100.0	99.2	214800
CHKB	111.3	100.0	99.6	602541
CHRM2	120.7	100.0	100.0	-
CITED2	97.1	99.2	99.1	614433
COL3A1	101.5	99.6	96.2	130020
COQ2	83.7	97.6	96.7	607426;146500
COX15	91.4	99.9	97.8	256000;615119
CPT1A	141.6	99.8	97.6	255120

CPT2	148.5	98.2	97.4	255110;608836;614212;600649
CRELD1	95.3	99.5	94.0	606217
CRYAB	95.9	100.0	98.2	615184
CSRP3	96.9	98.9	93.9	607482
CTNNA3	142.7	99.9	99.8	615616
DCHS1	155.3	99.9	99.4	607829
DES	115.4	100.0	99.6	604765;615325
DMD	115.1	99.5	98.1	300376
DOLK	175.4	100.0	100.0	610768
DPM3	229.1	100.0	100.0	612937
DPP6	127.1	99.7	97.9	612956
DSC2	134.1	99.4	97.4	610476
DSG2	136.6	99.8	99.6	610193
DSP	130.7	99.9	99.4	607450
DTNA	129.4	99.9	99.9	604169
DZIP1	146.0	98.3	95.9	-
EEF1A2	184.6	100.0	100.0	616409;616393
EFEMP2	139.5	100.0	100.0	614437
EHMT1	128.4	94.5	93.6	610253
ELN	114.6	99.8	98.3	123700
EMD	137.4	99.8	97.9	310300
EMILIN1	108.5	98.3	90.3	-
ENPP1	129.7	96.5	90.6	125853;601665;208000;613312;615522
FAH	122.1	100.0	99.5	276700
FBN1	149.0	100.0	99.7	102370
FBN2	149.7	100.0	99.8	121050
FBXO32	136.6	100.0	100.0	-
FGF12	114.8	100.0	99.1	617166
FHL1	64.9	99.4	93.8	300696
FHL2	115.6	99.8	98.1	-

FHOD3	138.2	100.0	99.4	-
FKRP	140.6	100.0	100.0	607155
FKTN	108.4	99.8	95.2	611615
FLNA	143.7	100.0	99.9	314400
FLNC	160.9	100.0	99.4	614065
FLT4	161.0	99.2	98.9	602089;153100
FNIP1	163.3	99.9	99.9	-
FOXC2	94.3	100.0	98.1	153400
FOXD4	.8	22.4	5.0	-
FOXE3	52.2	87.6	79.0	617349
FOXH1	65.6	100.0	98.7	603621
FOXL1	108.1	97.0	88.8	-
GAA	177.6	100.0	99.9	232300
GATA4	84.9	87.4	78.5	607941
GATA5	58.6	99.9	95.5	611496
GATA6	101.5	91.5	84.5	614475
GATAD1	131.5	100.0	98.7	614672
GATB	104.7	100.0	99.0	-
GATC	125.6	100.0	100.0	-
GBE1	172.1	99.9	99.7	263570;232500
GDF1	32.3	80.8	59.0	217095
GDF2	147.0	100.0	100.0	615506
GJA1	151.1	100.0	100.0	600309;241550
GJA5	201.5	100.0	100.0	614049
GLA	72.8	91.0	85.9	301500
GLB1	77.0	99.2	92.8	230650;253010;230600;230500
GMPPB	257.0	100.0	100.0	615352
GNB2	202.7	100.0	100.0	-
GNPTAB	148.1	99.9	99.7	252500;252600
GPD1L	124.4	100.0	98.8	611777

HADHA	70.8	95.5	88.3	609015;609016
HADHB	73.3	97.7	87.0	609015
HAND1	114.3	100.0	100.0	602406
HAND2	66.7	99.7	94.9	-
HCN2	36.4	59.8	47.7	-
HCN3	148.5	99.9	98.5	-
HCN4	81.0	100.0	99.2	613123
HEY2	192.9	99.8	98.7	604674
HFE	101.9	99.9	97.8	235200
HJV	147.9	100.0	100.0	602390
HSPB6	73.5	92.7	83.0	-
IDUA	139.0	94.6	87.4	607014
ILK	136.9	100.0	99.9	-
ITPA	126.4	100.0	100.0	613850;616647
JAG1	127.4	97.8	96.7	118450
JPH2	84.8	96.0	85.6	613873
JUP	126.3	100.0	99.8	611528
KCNA5	154.3	100.0	99.3	612240
KCND2	167.1	100.0	100.0	-
KCND3	168.3	100.0	98.6	607346
KCNE1	384.7	100.0	100.0	612347
KCNE2	152.4	100.0	96.1	611493
KCNE3	144.3	100.0	100.0	613119
KCNE4	113.8	80.5	80.5	-
KCNE5	115.8	98.5	93.5	300328
KCNH2	103.4	95.9	92.1	613688
KCNJ11	174.3	100.0	100.0	606176
KCNJ2	149.7	100.0	100.0	170390
KCNJ5	163.3	100.0	100.0	613677
KCNJ8	130.8	100.0	100.0	600935

KCNK3	139.8	97.9	95.1	615344
KCNN3	128.6	100.0	99.7	-
KCNQ1	138.7	93.5	90.6	607554
KDR	122.4	100.0	99.7	No OMIM phenotype
KLF10	135.5	100.0	99.9	-
KLHL24	162.9	100.0	100.0	617294
KMT2D	133.9	99.9	99.0	147920
KRAS	68.4	99.0	97.8	109800
LAMA2	131.5	99.9	99.1	607855
LAMA4	123.2	100.0	99.7	615235
LAMP2	88.2	99.3	96.0	300257
LDB3	154.8	95.4	94.8	601493
LEFTY2	59.2	94.3	84.3	601877
LIMS2	116.7	94.1	92.7	616827
LMNA	100.3	96.1	90.6	159001;115200
LMOD1	145.0	100.0	99.8	602715
LOX	119.0	99.8	99.6	617168
LRRC10	130.2	100.0	100.0	610846
LTBP3	134.5	99.8	98.6	601216;617809
LZTR1	141.0	100.0	99.9	616564;615670
MAT2A	95.7	99.2	93.8	-
MCTP2	126.4	99.4	97.7	-
MED13L	111.6	100.0	99.5	608808
MFAP5	104.8	100.0	98.7	616166
MIB1	128.4	100.0	99.6	615092
MLYCD	85.3	96.8	92.5	248360
MMP21	101.4	99.8	99.2	616749
MYBPC3	146.0	99.8	97.6	115197
MYBPHL	81.3	99.5	96.6	-
MYH11	126.4	100.0	99.7	132900

MYH6	102.0	99.2	96.1	614089
MYH7	99.2	99.1	96.7	613426
MYH7B	119.4	98.3	94.8	-
MYL2	119.5	94.8	81.1	608758
MYL3	100.9	100.0	100.0	608751
MYL4	133.6	100.0	100.0	617280
MYL7	139.7	100.0	99.6	-
MYLK	132.7	100.0	99.6	613780
MYLK2	137.4	100.0	100.0	192600
MYLK3	123.7	99.7	98.1	-
MYO6	94.9	99.1	96.3	606346;607821
MYOM1	127.6	99.7	98.0	-
MYOT	141.1	100.0	99.2	159000
MYOZ2	156.5	100.0	100.0	613838
MYPN	134.3	100.0	99.5	615248
MYRF	144.5	99.0	97.8	618280
NAA15	85.7	94.8	91.2	No OMIM phenotype
NEBL	109.0	99.2	97.1	605491
NEXN	64.1	87.9	71.5	613122
NKX2-5	94.0	100.0	100.0	271400
NKX2-6	128.9	100.0	100.0	217095
NNT	118.6	96.4	96.0	614736
NODAL	154.7	100.0	100.0	270100
NOS1AP	182.6	100.0	100.0	-
NOTCH1	128.4	99.3	97.9	109730
NOTCH2	130.5	100.0	99.2	610205
NPPA	160.6	100.0	100.0	612201
NPPB	261.9	100.0	100.0	-
NR2F2	201.1	100.0	99.1	615779
NRAS	163.7	100.0	100.0	614470

NUP155	119.0	98.7	96.2	615770
OBSCN	147.2	99.4	98.5	608616
PCCA	103.7	98.9	93.4	606054
PCCB	107.3	96.7	95.4	606054
PDLIM3	145.0	100.0	99.7	605889
PDLIM5	120.9	92.9	90.1	-
PEX5	110.1	99.9	98.8	616716;202370;214110
PEX7	113.8	88.0	81.0	215100;614879
PGM1	124.5	94.2	94.1	614921
PHKA1	100.8	97.8	93.4	300559
PHYH	71.1	100.0	98.9	266500
PITX2	147.2	99.8	97.2	601542
PKD1L1	112.6	100.0	99.3	617205
PKP2	89.8	94.3	86.9	609040
PKP4	122.0	99.2	96.1	-
PLD1	125.1	99.8	98.7	212093
PLEKHM2	138.9	100.0	99.9	-
PLN	174.5	100.0	100.0	609909
PLOD1	140.5	100.0	98.2	225400
PMEPA1	112.5	100.0	98.8	-
PMM2	127.8	99.8	99.8	212065
PNPLA2	137.8	99.8	96.1	610717
POMT1	131.1	99.5	97.3	609308
POMT2	98.9	99.8	97.3	613158
PPA2	87.0	97.3	88.6	617223;617222
PPCS	130.5	100.0	99.1	618189
PRDM16	230.9	99.9	99.2	615373
PRKAG2	128.8	99.4	96.1	600858
PRKG1	118.0	92.4	91.0	615436
PTPN11	81.2	97.7	87.6	151100

QRSL1	90.0	98.6	92.8	-
RAF1	101.0	99.9	99.2	611554
RANGRF	156.2	100.0	99.2	607954
RBM20	190.2	100.0	99.4	613172
RIT1	158.7	100.0	100.0	615355
RRAD	142.5	88.0	82.4	179503
RRAGC	99.6	99.9	99.4	608267
RYR2	124.3	99.8	98.8	600996
SCN10A	127.0	99.9	98.5	615551
SCN1B	172.2	98.2	96.3	612838
SCN2B	190.2	100.0	100.0	615378
SCN3B	143.0	100.0	100.0	613120
SCN4B	71.5	99.9	97.1	611819
SCN5A	143.1	99.0	98.7	614022
SDHA	94.3	84.5	77.9	613642
SGCA	158.3	100.0	99.6	608099
SGCB	158.3	97.8	96.5	604286
SGCD	71.2	99.6	96.5	606685
SGCG	120.1	100.0	99.4	253700
SHOC2	141.2	99.8	99.6	607721
SHROOM3	150.4	98.5	97.5	604570
SKI	110.2	99.7	97.1	182212
SLC22A5	125.6	100.0	99.6	212140
SLC25A20	85.6	100.0	98.9	212138
SLC25A4	126.5	100.0	99.8	609283;615418;617184
SLC2A10	162.5	97.7	97.7	208050
SLC30A5	94.5	99.4	96.8	No OMIM phenotype
SLMAP	115.2	98.4	92.5	-
SMAD1	144.2	99.8	98.2	-
SMAD2	133.0	100.0	99.8	601366

SMAD3	132.6	99.9	98.4	613795
SMAD4	113.8	99.9	99.9	174900;260350;175050;139210
SMAD6	177.9	90.7	79.3	614823
SMAD9	106.9	100.0	99.3	615342
SNTA1	98.3	92.6	80.2	612955
SOD2	195.5	100.0	100.0	No OMIM phenotype
SOS1	104.8	99.6	97.9	135300
SRI	121.3	99.8	96.4	-
SYNE1	127.1	98.1	97.5	612998
SYNE2	105.8	99.4	97.2	612999
TAB2	170.6	99.8	99.2	614980
TAF1	89.4	99.2	95.7	No OMIM phenotype
TAZ	104.7	99.3	93.7	302060
TBX1	91.3	87.4	77.6	217095
TBX20	110.5	100.0	99.8	611363
TBX5	128.4	100.0	100.0	142900
TCAP	105.8	100.0	100.0	607487;601954
TDGF1	142.1	98.8	91.8	187395
TECRL	79.5	97.5	91.7	614021
TFAP2B	164.3	98.8	96.4	169100
TGFB2	169.0	100.0	100.0	614816
TGFB3	139.0	100.0	100.0	107970
TGFBR1	156.6	93.6	93.6	609192
TGFBR2	159.9	100.0	99.9	614331
THBS4	124.2	100.0	99.4	-
TJP1	145.3	100.0	99.7	601009
TLL1	134.1	99.9	99.7	613087
TMEM43	122.9	99.9	98.4	604400
TMPO	128.0	98.0	93.8	188380
TNNC1	182.2	100.0	100.0	611879

TNNI3	113.8	99.6	95.5	613286
TNNI3K	111.6	99.8	99.6	616117
TNNT2	99.2	94.6	90.7	601494
TOR1AIP1	125.4	99.2	96.1	617072
TPM1	110.9	100.0	99.3	611878
TRDN	79.2	97.7	89.1	615441
TRIM63	106.1	100.0	99.9	606131
TRPM4	133.4	100.0	99.8	604559
TSFM	121.7	100.0	99.3	610505
TTN	170.8	98.5	98.0	604145;608807
TTR	128.3	94.6	94.6	105210
TXNRD2	123.8	96.8	95.9	-
VCL	94.4	99.9	98.5	611407
XIRP2	117.0	99.9	99.7	-
XK	76.5	99.7	97.6	300842
ZBTB17	142.8	100.0	100.0	-
ZFPM2	151.2	100.0	99.9	610187
ZIC3	125.7	100.0	99.9	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