

# WES HEART DISORDERS\* DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ABCC6	116.4	93.6	92.6	614473
ABCC9	157.9	99.9	99.2	614050
ACAD8	141.5	100.0	100.0	611283
ACAD9	135.5	98.4	95.7	611126
ACADVL	119.0	98.7	95.1	201475
ACSF3	128.8	99.9	99.3	614265
ACTA1	99.7	99.2	95.3	616852;161800;255310
ACTA2	137.6	100.0	99.8	611788
ACTC1	164.1	100.0	99.6	612794
ACTN1	143.8	100.0	99.9	615193
ACTN2	156.5	100.0	100.0	102573
ACVR1	165.1	100.0	100.0	135100
ACVR2B	140.9	97.0	94.7	613751
ADCY5	129.2	92.4	89.2	606703
ADRB1	155.1	97.2	89.8	-
ADRB2	131.5	100.0	100.0	-
AGK	112.2	99.3	96.3	614691;212350
AGL	146.5	99.7	98.0	232400
AGPAT2	109.7	99.0	95.0	608594
AGRN	115.0	95.1	89.3	615120
AGT	214.7	100.0	100.0	267430
AGTR1	134.6	92.0	91.9	145500
AKAP9	98.0	98.3	94.2	611820
ALDH1A2	114.8	100.0	99.6	603687
ALMS1	179.9	99.9	99.7	203800

ALPK3	98.7	94.6	92.5	618052
ANK2	160.5	100.0	99.9	600919
ANKRD1	101.6	99.5	96.8	609599
ATP1A4	161.6	100.0	99.5	-
ATPAF2	101.5	100.0	100.0	-
BAG3	136.5	100.0	100.0	613881
BANF1	58.3	98.0	88.1	614008
BGN	128.9	100.0	99.5	300989300106
BRCC3	53.0	83.8	63.2	-
BSCL2	113.8	100.0	100.0	269700
BVES	114.8	99.8	98.2	616812
CACNA1C	154.6	99.9	99.2	611875
CACNA1D	149.9	98.0	97.8	614896
CACNA2D1	82.6	93.1	84.4	114204
CACNA2D4	112.0	99.2	97.7	610478
CACNB2	150.9	99.5	96.9	611876
CALM1	114.3	100.0	99.7	614916
CALM2	54.0	67.8	65.8	114182
CALM3	117.4	99.9	99.5	114183
CAMK2D	106.9	97.3	93.8	-
CARD6	149.6	97.5	97.1	-
CASQ2	143.3	99.9	99.2	611938
CAV1	265.5	100.0	100.0	612526606721615343
CAV3	305.3	100.0	100.0	192600607801
CAVIN4	159.7	100.0	100.0	-
CDH2	137.5	98.5	97.5	-
CFAP53	146.5	97.6	94.3	614779
CFC1	74.5	82.7	71.3	217095605376613853
CHD7	150.9	99.9	98.8	214800
CHKB	98.5	100.0	99.0	602541

CHRM2	142.9	100.0	99.8	-
CIB1	131.1	95.2	92.5	-
CITED2	111.9	99.2	99.0	614433
COL3A1	104.2	97.8	92.4	130020
COQ2	89.3	96.1	93.2	607426146500
COX15	98.7	100.0	99.7	256000;615119
CPT1A	169.3	100.0	98.7	255120
CPT2	163.0	97.2	95.4	600649608836255110614212
CRELD1	114.5	99.9	97.8	606217
CRKL	166.7	100.0	99.8	602007
CRYAB	125.7	99.9	98.7	615184
CSRP3	103.0	100.0	99.9	607482
CTF1	24.5	27.8	20.0	-
CTNNA3	138.2	100.0	99.9	615616
CXADR	95.0	95.9	88.6	-
DES	121.2	99.9	98.1	615325;604765
DMD	111.5	99.4	97.4	300376
DMPK	117.7	99.9	97.9	60900
DNM2	127.6	97.5	94.4	606482615368160150
DOLK	203.0	100.0	99.9	-
DPM3	183.7	100.0	100.0	612937
DPP6	145.5	96.5	94.5	612956
DSC2	128.5	99.4	96.2	610476
DSG2	140.6	99.9	98.7	610193
DSP	154.1	100.0	99.8	607450
DTNA	156.5	100.0	100.0	604169
EDN1	145.6	100.0	100.0	615706
EEF1A2	177.6	98.8	93.6	616409616393
EFEMP2	121.1	100.0	99.9	614437
ELN	91.2	99.5	97.5	123700

EMD	99.8	99.8	97.2	310300
EMILIN1	75.6	96.8	87.5	-
ENG	128.8	97.4	93.6	187300
ENPP1	134.8	92.4	83.2	125853;208000;615522;613312;601665
EPG5	126.0	99.3	97.7	242840
EYA4	160.3	100.0	99.5	605362
FAH	151.3	100.0	99.9	276700
FBN1	159.8	99.9	99.5	102370
FBXO32	150.5	100.0	100.0	-
FGF12	95.6	99.6	96.3	617166
FGF13	102.6	99.7	97.6	-
FHL1	86.6	98.8	92.9	300696
FHL2	149.6	99.6	98.4	-
FHOD3	135.3	99.9	98.5	-
FKRP	94.7	100.0	99.7	607155
FKTN	120.2	99.2	94.2	611615
FLNA	137.4	100.0	99.5	314400
FLNB	150.0	99.8	99.2	108720;150250272460;108721112310
FLNC	165.1	100.0	99.7	614065
FLT4	156.1	98.6	97.9	153100602089
FOXD4	3.1	25.1	13.3	-
FOXE3	20.6	68.9	48.0	617349
FOXH1	47.4	98.5	85.1	603621
FXN	75.4	85.9	75.9	229300
GAA	128.5	100.0	99.9	232300
GATA4	87.4	68.5	60.6	607941
GATA5	44.3	98.3	84.6	611496
GATA6	61.8	83.7	72.1	614475
GATAD1	126.8	97.0	92.0	614672
GBE1	145.4	99.6	97.2	232500;263570

GDF1	19.5	65.0	48.4	217095
GDF2	163.2	100.0	100.0	615506
GJA1	246.6	100.0	100.0	600309
GJA5	268.6	100.0	100.0	614049
GJC1	192.8	100.0	100.0	-
GLA	80.9	99.7	97.5	301500
GLB1	94.3	99.6	97.0	230500;230600230650253010
GMPPB	228.8	100.0	100.0	615352
GNB5	125.8	99.9	98.3	617173617182
GNE	153.8	100.0	99.8	605820269921
GNPTAB	167.7	98.3	97.4	252500;252600
GPD1L	138.4	100.0	98.5	611777
H19				130650
HADHA	84.4	96.5	90.3	609016609015
HADHB	80.5	92.5	79.5	609015
HAND1	84.8	100.0	98.9	602406
HAND2	32.1	87.8	67.7	-
HCN1	122.4	99.9	97.8	615871
HCN2	53.2	58.3	50.7	-
HCN3	146.4	99.9	99.2	-
HCN4	79.4	98.3	91.8	613123
HEY2	146.1	99.2	92.8	604674
HFE	142.3	100.0	99.7	235200
HFE2	117.0	100.0	100.0	602390
HRAS	165.0	99.8	98.1	218040
HSPB6	63.1	89.4	77.4	-
IDUA	123.1	88.3	80.1	607014
ILK	171.7	100.0	100.0	-
IRX3	73.8	82.9	66.2	-
IRX4	90.0	95.8	92.3	606199

ITGB1BP2	74.6	99.3	95.7	-
ITPA	120.2	100.0	100.0	613850;616647
JAG1	148.4	98.1	97.5	118450
JPH2	87.8	90.4	75.3	613873
JUP	145.3	100.0	99.7	611528
KAT6B	192.5	99.6	98.5	603736;606170
KCNA5	143.3	99.4	96.0	612240
KCND2	183.4	100.0	100.0	-
KCND3	182.8	99.9	99.1	607346
KCNE1	462.5	100.0	100.0	612347
KCNE2	181.9	100.0	100.0	611493
KCNE3	176.8	100.0	100.0	613119
KCNE4	84.9	79.9	77.6	-
KCNE5	87.4	97.6	90.1	300328
KCNH2	102.8	92.3	84.8	613688
KCNJ11	299.4	100.0	100.0	606176
KCNJ12	575.4	100.0	100.0	-
KCNJ2	229.1	100.0	100.0	170390
KCNJ3	168.1	100.0	100.0	-
KCNJ5	194.1	100.0	99.8	613677
KCNJ8	177.3	100.0	100.0	600935
KCNK3	165.4	98.9	96.1	615344
KCNMB1	116.0	100.0	100.0	-
KCNN2	169.8	99.5	99.5	-
KCNN3	151.3	100.0	99.9	-
KCNQ1	114.9	93.0	90.3	607554
KCNQ1OT1				130650
KLF10	141.4	100.0	99.7	-
KLHL24	193.1	100.0	100.0	617294
KRAS	64.7	99.9	98.6	109800

LAMA2	143.5	99.9	99.5	607855
LAMA4	132.7	100.0	99.9	615235
LAMP2	106.1	92.7	91.2	300257
LDB3	127.3	95.5	93.7	601493
LEFTY2	42.4	91.6	77.2	601877
LIMS2	110.8	93.0	92.3	616827
LMNA	89.4	97.9	91.2	115200;159001
LMOD1	177.4	100.0	100.0	-
LOX	104.6	99.8	97.6	-
LPL	147.3	100.0	100.0	144250238600
LRIT3	142.4	94.4	94.1	615058
LRP1	196.0	99.7	99.1	-
LRP2	176.3	100.0	99.8	222448
LRP6	169.3	100.0	99.7	610947
LRRC10	194.0	100.0	100.0	610846
LZTR1	134.2	100.0	99.4	615670;616564
MAP2K1	92.4	99.8	95.6	615279
MAP2K2	108.0	97.6	89.1	615280
MAT2A	115.4	99.7	96.9	-
MED13L	134.7	100.0	99.6	608808
MEF2C	137.7	97.9	93.5	613443
MFAP5	126.8	100.0	99.5	616166
MIB1	141.7	100.0	99.6	615092
MLYCD	75.8	91.2	86.8	248360
MMP2	164.7	100.0	100.0	259600
MMP21	93.5	90.3	84.6	-
MYBPC3	142.7	98.5	95.7	115197
MYBPHL	99.9	99.2	94.3	-
MYH11	132.8	100.0	99.3	132900
MYH6	113.5	99.0	96.1	614089

MYH7	111.5	99.4	96.8	613426
MYH7B	113.2	97.6	94.5	-
MYL2	134.8	98.7	90.2	608758
MYL3	103.4	100.0	100.0	608751
MYL4	159.0	100.0	100.0	617280
MYL7	130.9	100.0	100.0	-
MYLK	148.6	99.9	99.3	613780
MYLK2	120.1	100.0	100.0	192600
MYO1C	111.5	99.3	98.2	-
MYO6	89.7	98.1	92.3	606346607821
MYOCD	182.0	100.0	100.0	-
MYOM1	149.5	99.8	98.4	-
MYOM2	156.5	100.0	99.4	-
MYOT	139.6	99.3	95.4	159000
MYOZ1	97.0	100.0	100.0	604103
MYOZ2	145.9	100.0	100.0	613838
MYPN	142.5	99.3	98.4	615248
MYZAP	131.5	95.2	91.1	-
NCOA1	152.6	100.0	99.6	-
NEBL	102.2	96.7	92.9	605491
NEXN	79.8	94.2	79.9	613122
NGF	257.6	100.0	100.0	608654
NKX2-5	83.3	100.0	99.5	271400
NKX2-6	104.5	100.0	99.7	217095
NNT	136.9	98.6	97.1	614736
NODAL	160.7	100.0	99.9	270100
NOS1AP	192.2	100.0	100.0	-
NOS3	107.6	95.3	91.0	-
NOTCH1	137.5	99.1	98.0	109730
NOTCH2	172.4	100.0	99.9	610205



NPHP3	115.5	99.4	96.1	267010604387208540
NPPA	115.7	100.0	100.0	612201
NPPB	160.7	100.0	100.0	-
NR2F2	246.1	98.7	94.3	615779
NRAS	188.4	100.0	100.0	614470
NUP155	115.1	97.6	92.3	615770
NUP37	168.4	98.5	93.4	-
OBSCN	159.3	99.3	98.2	608616
PCCA	103.1	96.4	89.3	606054
PCCB	129.7	98.7	96.5	606054
PDLIM3	148.7	100.0	100.0	605889
PGM1	133.9	100.0	99.9	614921
PHKA1	106.7	98.9	95.3	300559
PHYH	74.7	97.5	90.8	266500
PITX2	147.8	99.7	97.5	601542
PKD1L1	123.8	100.0	99.6	-
PKP2	99.6	94.6	87.7	609040
PKP4	139.0	99.2	96.2	-
PLEC	114.4	99.7	98.7	613723
PLEKHM2	112.9	100.0	99.7	-
PLN	210.0	100.0	100.0	609909
PLOD1	138.0	99.8	97.6	225400
PMM2	141.1	99.9	99.4	212065
PNPLA2	113.5	99.7	97.4	610717
POMT1	155.7	99.7	98.1	609308
POMT2	111.1	98.9	97.5	613158
PPA2	80.5	94.6	82.4	617223;617222
PPARGC1A	142.7	99.9	99.3	-
PPP3CB	83.9	95.9	89.4	-
PPP3R2	221.4	100.0	100.0	-

PRDM16	161.7	100.0	99.1	615373
PRKAG2	125.7	98.1	91.6	600858
PRKG1	123.4	98.7	95.4	615436
PTPN11	103.1	97.9	92.5	151100
RAF1	127.4	100.0	99.7	611554
RANGRF	114.0	99.9	98.4	607954
RBM20	180.9	99.2	96.6	613172
RIT1	165.6	100.0	100.0	615355
RPSA	88.9	100.0	99.7	271400
RYR1	120.7	96.8	93.7	145600
RYR2	142.2	99.7	98.4	600996
SCN10A	165.4	100.0	99.5	615551
SCN1B	168.3	97.1	96.1	612838
SCN2B	185.8	100.0	100.0	615378
SCN3B	147.3	100.0	100.0	613120
SCN4B	77.5	100.0	97.9	611819
SCN5A	169.7	99.0	99.0	614022
SELENON	111.6	85.3	83.3	255310602771
SGCA	147.7	100.0	99.7	608099
SGCB	154.2	96.6	94.2	604286
SGCD	94.7	100.0	99.4	606685
SGCG	138.6	100.0	100.0	253700
SHOC2	140.6	100.0	99.4	607721
SHROOM3	137.5	99.9	98.9	604570
SKI	85.5	96.5	90.9	182212
SLC22A5	153.3	100.0	100.0	212140
SLC25A20	110.3	100.0	99.7	212138
SLC25A4	134.2	100.0	100.0	-
SLC2A10	166.9	97.7	97.6	208050
SLC8A1	198.2	99.7	99.0	-

SLMAP	121.2	93.5	85.2	-
SMAD1	184.0	99.9	99.7	-
SMAD2	151.7	99.9	99.1	601366
SMAD3	131.7	99.9	99.2	613795
SMAD4	125.5	100.0	100.0	-
SMAD6	100.5	80.0	72.0	614823
SMAD9	132.8	100.0	100.0	615342
SMYD1	140.0	100.0	100.0	-
SNTA1	97.2	82.4	77.3	612955
SOS1	94.4	96.7	90.5	135300
SRI	114.5	97.9	88.8	-
SYNE1	136.6	98.2	97.6	612998
SYNE2	123.1	98.6	96.0	612999
SYNPO2	195.8	99.7	99.4	-
TAB2	210.5	99.7	97.6	614980
TAZ	94.0	99.9	98.8	302060
TBX1	75.3	77.1	67.4	217095
TBX20	142.8	99.9	99.2	611363
TBX5	141.3	100.0	100.0	142900
TCAP	89.0	100.0	99.2	607487601954
TDGF1	151.4	99.8	96.4	187395
TECRL	59.2	89.8	77.1	614021
TFAP2B	153.7	98.8	96.3	169100
TGFB2	176.9	100.0	99.9	614816
TGFB3	171.5	100.0	100.0	107970
TGFBR1	173.3	93.7	93.6	609192
TGFBR2	193.7	100.0	99.9	614331
TLL1	140.1	100.0	99.9	613087
TMEM43	125.1	100.0	99.5	604400
TMOD1	109.8	100.0	100.0	190930

TMPO	117.8	98.6	94.4	188380
TNNC1	174.5	100.0	100.0	611879
TNNI3	86.8	98.2	86.7	613286
TNNI3K	118.8	98.7	96.0	-
TNNT2	106.2	100.0	99.9	601494
TOR1AIP1	143.8	97.6	95.9	617072
TPM1	133.0	99.7	97.9	611878
TRDMT1	100.0	91.8	85.1	-
TRDN	71.8	83.5	70.7	615441
TRIM63	118.5	100.0	99.6	606131
TRPM4	109.4	99.7	98.5	604559
TTN	187.8	98.2	97.2	604145608807
TTR	152.3	94.6	94.6	105210
TXNRD2	119.3	93.3	91.2	-
VCL	115.8	100.0	99.8	611407
XIRP2	138.5	100.0	99.7	-
XK	96.3	99.9	99.1	300842
ZBTB17	142.2	100.0	100.0	-
ZFH3	129.2	100.0	99.6	-
ZFPM2	196.3	100.0	99.6	610187
ZIC3	113.7	100.0	99.8	306955

*Gene symbols used follow HGNC 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*