WES HEREDITARY CANCER DG 3.2

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
A2ML1	105.6	99.9	99.3	-
ACD	159.3	100.0	99.9	616553
AIP	140.9	100.0	99.6	102200
AKT1	169.4	100.0	99.9	615109
ALK	129.0	100.0	99.4	613014
AMH	67.1	99.4	92.9	261550
AMHR2	159.4	100.0	99.6	261550
ANKRD26	82.9	94.6	88.5	188000
APC	132.4	99.9	99.7	175100
ARMC5	156.5	100.0	99.1	615954
ASXL1	128.2	99.8	98.9	614286
ATM	115.1	99.4	97.1	114480
ATR	164.5	99.7	99.0	614564
AXIN2	125.3	100.0	99.8	608615
BAP1	107.2	83.9	82.4	614327
BARD1	143.9	100.0	99.8	114480
BLM	113.6	99.3	97.7	210900
BMPR1A	82.0	99.5	94.0	174900
BRAF	63.9	89.4	77.6	613706
BRCA1	167.7	99.4	98.4	604370
BRCA2	97.7	99.1	98.2	612555
BRIP1	129.1	99.4	98.5	114480
BUB1	131.3	99.7	98.4	-
BUB1B	116.3	99.3	98.3	257300;176430
BUB3	119.2	99.1	97.9	-

CARD11	142.3	100.0	99.9	616452
CBL	133.8	97.3	96.9	613563
CD27	99.3	99.9	98.3	No OMIM phenotype
CD70	102.2	100.0	96.8	No OMIM phenotype
CDC73	115.4	99.8	98.3	145001
CDH1	108.4	99.2	98.6	114480;176807;176430
CDH23	189.6	100.0	100.0	617540
CDK4	101.0	99.9	99.0	609048
CDKN1A	163.5	100.0	100.0	-
CDKN1B	149.1	99.9	99.3	610755
CDKN1C	71.4	89.9	81.6	130650
CDKN2A	102.2	92.3	92.3	155601;606719;155755
CDKN2B	89.9	100.0	99.7	-
CDKN2C	132.1	100.0	100.0	-
CEBPA	84.4	95.9	80.1	601626
CHEK2	96.0	84.9	80.7	609265;114480;176807
CREBBP	104.9	99.6	97.8	180849
CTC1	116.1	100.0	99.1	612199
CTLA4	128.1	100.0	100.0	No OMIM phenotype
CTNNA1	113.3	98.8	97.2	-
CTR9	141.9	99.9	99.8	-
CYLD	110.6	99.6	98.8	601606;132700
DDB2	159.5	99.7	97.7	278740
DDX11	106.4	84.9	80.0	613398
DDX41	158.2	100.0	100.0	616871
DICER1	148.8	99.5	98.5	138800;601200;180295
DIS3L2	152.8	100.0	100.0	267000
DKC1	90.7	99.7	97.2	305000
DLST	92.4	95.7	87.7	618475
DNAJC21	117.0	99.5	97.4	617052

EGFR	151.0	100.0	100.0	211980
EGLN1	81.6	89.1	80.1	-
EGLN2	134.5	100.0	100.0	-
ELANE	128.3	99.9	98.8	202700;162800
ELP1	124.3	99.8	98.9	155255
EPCAM	74.1	97.5	89.6	613244
ERCC1	81.0	100.0	96.4	610758
ERCC2	130.5	100.0	99.4	278730
ERCC3	92.3	96.8	95.6	610651
ERCC4	122.5	100.0	99.9	278760;610965;615272
ERCC5	118.6	99.9	99.0	278780
ERCC6	169.3	100.0	100.0	600630;211980;133540;214150
ESR2	112.9	99.9	98.9	-
ETV6	177.8	100.0	99.3	616216
EXT1	89.6	99.6	97.1	133700;215300
EXT2	124.9	99.9	99.0	133701
EZH2	144.1	99.7	98.0	277590
FANCA	110.8	99.9	98.7	227650
FANCB	75.4	98.0	91.7	300514
FANCC	100.9	96.9	95.7	227645;613899
FANCD2	115.5	98.7	95.9	227646
FANCE	126.4	90.7	85.5	600901
FANCF	232.8	100.0	100.0	603467
FANCG	147.5	100.0	99.9	614082
FANCI	142.7	99.8	98.6	609053
FANCL	103.0	99.4	97.6	614083
FANCM	99.4	98.9	96.3	614087
FAS	337.2	100.0	99.6	601859
FASLG	95.5	100.0	99.1	601859
FBXW7	158.8	99.9	98.2	No OMIM phenotype

FH	128.8	93.2	87.2	150800;606812
FLCN	168.2	100.0	100.0	135150
G6PC3	116.4	100.0	99.9	612541
GALNT12	85.3	86.4	82.6	608812
GATA2	109.3	99.8	97.0	614286
GDNF	179.4	100.0	100.0	171300
GFI1	88.9	100.0	99.9	607847;613107
GPC3	77.3	98.8	92.9	312870
GPR161	175.8	100.0	100.0	155255
GREM1	107.3	100.0	100.0	601228
GRHL2	125.6	100.0	99.9	616029
HAVCR2	122.7	100.0	99.8	618398
HAX1	148.7	100.0	100.0	610738
HOXB13	182.2	100.0	99.6	610997
IDH1	72.6	90.6	75.5	614569
IDH2	108.9	99.8	97.4	614569
IKZF1	183.4	99.3	99.3	No OMIM phenotype
IPMK	102.4	98.9	89.9	-
ITK	102.0	99.8	98.6	No OMIM phenotype
KIF1B	137.3	99.9	99.2	171300
KIT	143.1	100.0	99.4	606764;154800;601626;273300
KRAS	68.4	99.0	97.8	601626
LHCGR	114.6	96.6	92.4	176410
LIG4	183.8	99.8	99.3	606593
LZTR1	141.0	100.0	99.9	615670
MAD2L2	147.2	100.0	99.9	617243
MAP2K1	91.3	99.6	96.1	615279
MAP2K2	128.3	98.5	95.3	615280
MAX	83.1	99.8	97.7	171300
MCM8	113.6	99.9	98.8	No OMIM phenotype

MCM9	132.6	99.9	99.0	No OMIM phenotype
MDH2	102.0	98.0	98.0	-
MEN1	122.0	96.2	94.1	131100
MET	159.2	100.0	99.4	605074
MITF	135.0	100.0	99.9	614456
MLH1	139.2	100.0	99.9	609310;276300;158320
MPL	132.8	100.0	99.8	601977;604498
MRE11	49.6	98.2	88.6	604391
MSH2	116.8	98.5	94.5	276300;158320;120435
MSH3	133.8	97.8	97.1	617100
MSH6	147.4	100.0	99.3	614350;608089;276300
MTAP	89.0	98.3	91.8	112250
MUTYH	157.2	100.0	100.0	132600;608456
NBN	96.9	99.2	97.8	251260
NF1	117.2	91.8	89.3	162200;193520;601321;607785;162210
NF2	103.5	100.0	99.6	101000;162091;607174
NHP2	143.7	100.0	100.0	613987
NOP10	106.3	100.0	99.2	224230
NPM1	78.3	95.3	84.9	601626
NRAS	163.7	100.0	100.0	613224
NSD1	137.5	100.0	99.8	601626;117550
NTHL1	116.2	100.0	99.9	602656
PALB2	150.5	100.0	99.9	114480;610832;613348
PARN	110.0	81.1	80.4	616353
PAX5	100.5	98.8	95.4	167414
PDGFB	125.7	100.0	100.0	607174
PDGFRA	125.7	100.0	100.0	173490
PHOX2B	103.1	100.0	100.0	613013
PIK3CA	128.2	97.7	97.3	612918
PMS2	100.9	83.9	81.6	276300;614337

PMS2CL				-
POLD1	127.6	98.4	95.1	612591
POLE	132.6	100.0	99.5	615083
POLH	115.5	100.0	99.1	278750
POT1	105.9	99.5	98.5	615848
POU6F2	137.0	95.2	95.1	601583
PPM1D	148.9	100.0	99.9	114480
PRF1	121.1	91.2	90.1	605027;603553
PRKAR1A	78.7	97.0	89.1	610489;160980;255960
PRKN	72.0	66.9	65.8	608935
PRSS1	164.5	100.0	100.0	167800
PTCH1	110.2	99.3	96.6	109400
PTEN	144.3	99.5	97.2	607174;158350;153480;613028;605309
PTPN11	81.2	97.7	87.6	163950
RAD50	93.7	96.6	89.7	613078
RAD51C	143.5	99.8	99.4	613390
RAD51D	145.6	100.0	99.6	614291
RAF1	101.0	99.9	99.2	611553
RB1	88.1	96.3	93.2	180200
RECQL4	158.3	99.9	98.6	266280;268400;218600
REST	104.1	98.4	97.4	616806
RET	140.9	100.0	98.7	155240;171300;171400;162300
RHBDF2	106.3	99.8	98.4	148500
RIT1	158.7	100.0	100.0	615355
RMRP				No OMIM phenotype
RNASEL	130.8	100.0	99.6	601518
RNF43	139.8	99.8	98.3	617108
RPL11	89.1	99.9	97.9	612562
RPL15	31.8	84.9	70.4	615550
RPL18	112.1	100.0	99.9	618310

RPL27	33.8	68.0	56.6	617408
RPL35A	80.0	94.7	84.9	612528
RPL5	32.6	81.9	59.7	612561
RPS10	90.6	96.6	87.6	613308
RPS15A	60.8	95.3	84.1	618313
RPS17	38.5	85.0	67.8	612527
RPS19	95.1	100.0	99.9	105650
RPS20	63.7	96.6	87.5	-
RPS24	79.4	96.2	90.3	610629
RPS26	89.3	93.2	81.2	613309
RPS27	38.2	95.5	70.0	603702
RPS28	47.1	99.7	86.3	603685
RPS29	61.7	78.0	70.5	615909
RPS7	89.6	81.7	66.9	612563
RTEL1	130.0	99.7	97.2	615190
RUNX1	83.1	98.6	93.0	601626;601399
SAMD9	158.4	99.9	99.8	617053
SAMD9L	169.1	100.0	99.9	159550
SBDS	176.5	100.0	99.9	260400
SDHA	94.3	84.5	77.9	252011;614165
SDHAF2	121.8	94.6	93.5	601650
SDHB	123.9	100.0	100.0	606764;606864;115310;171300
SDHC	91.3	100.0	98.9	605373;606764;606864
SDHD	38.8	53.8	49.0	171300;168000;114900;615106;606864
SEMA4A	127.6	100.0	99.4	-
SFTPA1	158.2	100.0	100.0	178500
SFTPA2	154.3	100.0	100.0	178500
SH2B3	113.3	99.0	94.7	-
SH2D1A	105.1	97.8	92.9	No OMIM phenotype
SHOC2	141.2	99.8	99.6	607721

SLX4	124.7	100.0	99.9	613951
SMAD4	113.8	99.9	99.9	174900;175050;139210
SMAD9	106.9	100.0	99.3	-
SMARCA4	154.0	99.9	99.2	613325
SMARCB1	201.9	100.0	99.9	614608;162091;609322
SMARCE1	74.4	93.7	85.9	607174
SOS1	104.8	99.6	97.9	610733
SPINK1	86.1	99.9	99.0	167800
SPRED1	146.8	99.8	98.2	611431
SQSTM1	120.9	99.8	97.8	602080
STK11	122.2	92.4	91.9	175200;260350
SUCLG2	55.7	91.7	79.1	No OMIM phenotype
SUFU	138.3	100.0	100.0	155255;109400;607174
TERC				127550;614743
TERF2IP	115.7	100.0	99.9	-
TERT	135.5	97.0	94.8	615134;614742;613989
TG	123.8	99.9	98.5	274700
THPO	74.2	81.4	78.7	187950
TINF2	160.3	100.0	100.0	268130;613990
TMEM127	106.0	99.9	97.7	171300
TNFRSF11A	119.0	94.9	93.8	602080
TP53	94.2	99.0	95.2	114480;151623;260350;137800;202300;114500;614740;607107;259500
TRIM28	147.1	97.7	96.3	No OMIM phenotype
TRIM37	123.0	98.3	97.1	No OMIM phenotype
TRIP13	142.0	100.0	99.9	617598
TSC1	118.1	99.5	98.2	191100
TSC2	140.3	100.0	99.8	613254
USB1	131.2	100.0	98.8	604173
VHL	131.2	95.5	90.6	263400;171300;193300
WAS	75.5	94.1	83.7	313900;300299

WRAP53	164.2	100.0	100.0	613988
WRN	133.2	99.3	98.2	277700
WT1	72.4	97.6	96.1	194070
XPA	73.4	99.2	97.3	278700
XPC	144.8	100.0	99.9	278720

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.