

WES HEREDITARY CANCER DG 2.18

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
A2ML1	123.4	100.0	99.6	-
ACD	148.6	100.0	99.9	616553
AIP	132.1	100.0	99.0	102200
AKT1	156.2	100.0	99.5	615109
ALK	137.8	100.0	99.4	613014
AMH	54.3	96.4	83.8	261550
AMHR2	157.4	100.0	99.5	261550
ANKRD26	94.5	95.0	89.3	188000
APC	168.3	100.0	99.7	175100
ARMC5	153.8	100.0	99.4	615954
ASXL1	146.1	100.0	99.5	614286
ATM	133.4	99.8	98.1	114480
ATR	178.2	99.9	99.4	614564
AXIN2	140.7	100.0	99.9	608615
BAP1	111.5	84.4	83.0	614327
BARD1	161.9	100.0	99.8	114480
BLM	133.4	99.8	98.3	210900
BMPR1A	98.0	99.8	96.6	174900
BRAF	83.8	95.6	85.1	613706
BRCA1	195.0	99.4	98.8	604370
BRCA2	119.2	99.8	98.5	612555
BRIP1	151.0	99.9	99.0	114480
BUB1	156.0	99.8	98.8	-
BUB1B	141.0	99.6	98.9	257300;176430
BUB3	139.0	99.8	99.1	-

CARD11	150.3	100.0	99.9	616452
CBL	152.5	97.3	97.1	613563
CDC73	136.4	100.0	99.4	145001
CDH1	126.7	99.2	99.1	114480;176807;176430
CDH23	187.3	100.0	100.0	617540
CDK4	114.2	100.0	99.7	609048
CDKN1A	138.3	100.0	100.0	-
CDKN1B	161.7	100.0	99.8	610755
CDKN1C	66.7	86.3	74.8	130650
CDKN2A	96.9	92.3	92.1	606719;155601;155755
CDKN2B	88.3	100.0	99.9	-
CDKN2C	159.2	100.0	100.0	-
CEBPA	84.1	98.6	83.9	601626
CHEK2	112.6	85.0	81.5	114480;176807;609265
CREBBP	124.1	99.7	98.5	180849
CTC1	128.0	100.0	99.6	612199
CTNNA1	125.7	99.3	98.1	-
CTR9	168.6	100.0	99.9	-
CYLD	127.1	99.8	98.0	132700;601606
DDB2	165.3	99.6	97.5	278740
DDX11	108.7	85.2	80.7	613398
DDX41	161.7	100.0	100.0	616871
DICER1	179.1	99.8	99.0	138800;601200;180295
DIS3L2	164.4	100.0	99.8	267000
DKC1	108.6	99.8	98.7	305000
DLST	97.1	96.7	90.3	618475
DNAJC21	146.2	99.9	99.0	617052
EGFR	166.1	100.0	100.0	211980
EGLN1	78.2	89.3	82.2	-
EGLN2	138.4	100.0	99.8	-

ELANE	143.2	99.7	97.4	162800;202700
ELP1	149.3	99.8	99.0	155255
EPCAM	93.7	98.6	90.3	613244
ERCC1	88.2	100.0	99.3	610758
ERCC2	135.5	100.0	99.7	278730
ERCC3	111.3	100.0	99.4	610651
ERCC4	141.7	100.0	99.9	615272;610965;278760
ERCC5	139.0	100.0	99.6	278780
ERCC6	197.0	100.0	100.0	211980;133540;214150;600630
ESR2	122.2	100.0	99.7	-
ETV6	185.3	100.0	99.9	616216
EXT1	114.3	99.9	98.4	215300;133700
EXT2	145.0	100.0	99.3	133701
EZH2	171.4	100.0	99.5	277590
FAN1	155.8	100.0	99.8	-
FANCA	124.7	100.0	99.4	227650
FANCB	88.6	98.6	94.1	300514
FANCC	121.7	99.9	99.3	613899;227645
FANCD2	139.2	99.5	97.5	227646
FANCE	134.6	89.8	85.1	600901
FANCF	268.5	100.0	100.0	603467
FANCG	161.0	100.0	99.9	614082
FANCI	166.6	99.9	99.2	609053
FANCL	126.2	100.0	98.6	614083
FANCM	112.9	99.6	97.3	614087
FAS	262.8	100.0	99.6	601859
FASLG	98.0	100.0	99.6	601859
FH	147.0	92.1	88.3	150800;606812
FLCN	173.1	100.0	100.0	135150
G6PC3	113.2	100.0	99.9	612541

GALNT12	99.4	85.8	82.7	608812
GATA2	119.1	100.0	98.3	614286
GDNF	204.6	100.0	100.0	171300
GFI1	88.3	100.0	99.2	613107;607847
GPC3	90.4	99.1	94.7	312870
GPR161	186.4	100.0	100.0	155255
GREM1	121.2	100.0	100.0	601228
GRHL2	139.4	100.0	100.0	616029
HAVCR2	141.9	100.0	100.0	618398
HAX1	166.6	100.0	100.0	610738
HNF1A	171.3	100.0	99.8	144700
HOXB13	194.7	100.0	99.1	610997
IDH1	91.4	93.3	80.1	614569
IDH2	111.5	99.7	97.4	614569
IPMK	112.5	99.2	92.0	-
KIF1B	167.4	100.0	99.6	171300
KIT	163.2	100.0	99.6	601626;273300;154800;606764
KRAS	84.3	99.5	96.9	601626
LHCGR	142.3	94.1	92.3	176410
LIG4	222.9	100.0	99.9	606593
LZTR1	136.8	100.0	99.9	615670
MAD2L2	151.7	100.0	99.9	617243
MAP2K1	111.1	99.8	97.1	615279
MAP2K2	123.0	98.5	95.1	615280
MAX	97.7	100.0	98.9	171300
MDH2	116.9	98.0	97.9	-
MEN1	141.0	99.9	99.1	131100
MET	178.5	100.0	99.5	605074
MITF	160.6	100.0	99.9	614456
MLH1	166.0	100.0	99.9	276300;609310;158320

MPL	140.2	100.0	99.5	601977;604498
MRE11	64.1	98.9	93.3	604391
MSH2	142.5	99.8	97.7	276300;120435;158320
MSH3	163.2	100.0	99.2	617100
MSH6	179.9	100.0	99.8	276300;614350;608089
MTAP	108.3	99.1	93.5	112250
MUC5B	86.0	82.5	72.9	178500
MUTYH	154.3	100.0	100.0	608456;132600
NBN	109.6	99.9	98.6	251260
NF1	131.8	92.6	90.2	193520;607785;162200;162210;601321
NF2	119.7	100.0	99.9	607174;101000;162091
NHP2	146.1	100.0	100.0	613987
NOP10	147.4	100.0	99.8	224230
NPM1	90.1	98.2	85.3	601626;-
NRAS	185.6	100.0	100.0	613224
NSD1	175.3	100.0	99.9	117550;601626
NTHL1	111.0	100.0	99.8	602656
PALB2	182.1	100.0	100.0	114480;610832;613348
PARN	165.2	100.0	99.9	616353
PAX5	115.0	98.7	96.1	167414
PDGFB	121.4	100.0	99.3	607174
PDGFRA	153.6	100.0	100.0	173490
PHOX2B	117.2	100.0	99.7	613013
PIK3CA	154.5	100.0	99.8	612918
PMS2	125.2	84.3	82.8	276300;614337
PMS2CL				-
POLD1	121.7	98.5	95.2	612591
POLE	140.6	100.0	99.8	615083
POLH	136.9	100.0	99.6	278750
POT1	120.7	99.9	99.0	615848

POU6F2	155.2	100.0	100.0	601583
PPM1D	177.8	100.0	99.9	114480
PRF1	114.3	91.2	90.8	603553;605027
PRKAR1A	94.5	99.3	93.5	255960;160980;610489
PRKN	97.4	79.8	78.8	608935
PRSS1	181.1	100.0	100.0	167800
PTCH1	127.2	99.2	97.6	109400
PTCH2	118.2	99.9	99.0	109400
PTEN	166.6	99.5	97.0	607174;605309;158350;153480;613028
PTPN11	100.3	99.1	93.7	163950
RAD50	118.2	97.5	91.6	613078
RAD51C	162.8	100.0	99.8	613390
RAD51D	166.9	100.0	99.9	614291
RAF1	125.8	100.0	100.0	611553
RB1	102.8	98.5	93.7	180200
RECQL4	149.8	99.8	98.1	218600;268400;266280
REST	119.0	98.5	98.2	616806
RET	139.5	99.9	99.1	171400;171300;162300;155240
RHBDF2	101.4	99.9	98.6	148500
RIT1	178.7	100.0	100.0	615355
RNASEL	157.0	100.0	99.8	601518
RNF43	155.2	99.9	99.1	617108
RPL11	111.0	100.0	100.0	612562
RPL15	35.2	86.8	78.0	615550
RPL18	106.9	100.0	100.0	618310
RPL27	33.7	73.6	56.5	617408
RPL35A	94.5	97.1	88.7	612528
RPL5	42.2	86.2	70.0	612561
RPS10	109.9	97.7	91.7	613308
RPS15A	68.5	96.9	86.7	618313

RPS17	43.8	84.2	69.8	612527
RPS19	92.9	100.0	99.6	105650
RPS20	81.0	98.6	93.6	-
RPS24	98.4	98.5	93.4	610629
RPS26	92.6	95.7	84.9	613309
RPS27	39.4	89.3	60.9	603702
RPS28	57.7	100.0	94.8	603685
RPS29	113.4	100.0	98.2	615909
RPS7	112.0	80.0	68.7	612563
RTEL1	127.7	99.5	96.8	615190
RUNX1	85.5	99.3	94.9	601626;601399
SAMD9	185.7	100.0	99.8	617053
SAMD9L	192.5	100.0	100.0	159550
SBDS	197.8	100.0	100.0	260400
SDHA	100.0	85.8	80.4	252011;614165
SDHAF2	139.9	94.6	94.2	601650
SDHB	146.3	100.0	100.0	115310171300;606864;606764;612359
SDHC	107.5	100.0	99.3	605373;606864;606764
SDHD	48.5	54.0	51.6	171300;606864;615106;114900;168000
SEMA4A	134.8	100.0	99.8	-
SFTPA1	169.4	100.0	100.0	178500
SFTPA2	154.1	100.0	100.0	178500
SH2B3	106.6	99.4	95.1	-
SHOC2	164.2	99.9	99.4	607721
SLX4	130.9	100.0	99.8	613951
SMAD4	127.2	100.0	99.9	175050;174900;139210
SMAD9	118.3	100.0	99.9	-
SMARCA4	157.4	99.9	99.0	613325
SMARCB1	210.4	100.0	100.0	609322;614608;162091
SMARCE1	81.3	96.1	88.1	607174

SOS1	123.6	99.8	98.4	610733
SPINK1	101.3	100.0	99.3	167800
SPRED1	171.2	100.0	98.9	611431
SQSTM1	125.7	98.8	95.5	602080
STK11	128.8	100.0	99.3	260350;175200
SUFU	153.1	100.0	100.0	155255;607174;109400
TERC				614743;127550
TERF2IP	100.5	99.9	97.3	-
TERT	132.4	96.2	94.5	613989;615134;614742
TG	137.4	100.0	99.4	274700
THPO	104.3	100.0	99.5	187950
TINF2	187.5	100.0	100.0	613990;268130
TMEM127	102.5	99.5	96.5	171300
TNFRSF11A	141.5	94.6	93.3	602080
TP53	113.9	99.9	97.7	114480;202300;260350;151623;137800;259500;607107;114500;614740
TRIP13	165.0	100.0	100.0	617598
TSC1	136.7	99.8	98.7	191100
TSC2	140.9	100.0	99.6	613254
USB1	139.6	100.0	99.4	604173
VHL	141.3	96.3	91.4	193300;171300;263400
WAS	81.1	95.9	85.3	313900;300299
WRAP53	187.5	100.0	100.0	613988
WRN	147.8	99.9	98.8	277700
WT1	83.5	99.9	98.3	194070
XPA	83.7	99.6	95.6	278700
XPC	162.6	100.0	100.0	278720

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