

WES HEREDITARY CANCER DG 2.16

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
A2ML1	104.6	99.9	99.5	-
ACD	159.6	100.0	100.0	616553
AIP	137.2	100.0	99.6	102200
ALK	128.1	100.0	99.4	613014
ANKRD26	83.3	95.3	90.1	188000
APC	141.4	99.9	99.6	175100
ARMC5	170.0	100.0	99.6	615954
ASXL1	132.4	100.0	99.5	614286
ATM	110.9	99.6	97.2	114480
ATR	144.6	99.8	98.6	614564
AXIN2	124.2	100.0	99.9	608615
BAP1	104.8	85.0	82.9	614327
BARD1	140.6	100.0	99.9	114480
BLM	111.0	99.6	98.0	210900
BMPR1A	78.2	99.5	92.9	174900
BRAF	72.5	92.4	80.2	613706
BRCA1	161.4	99.1	98.1	604370
BRCA2	106.2	99.6	98.7	612555
BRIP1	125.8	99.7	98.8	114480
BUB1	126.2	99.9	98.6	1
BUB1B	122.0	99.8	98.7	257300;176430
BUB3	125.3	99.8	98.1	2
CBL	126.0	97.3	97.0	613563
CDC73	113.6	99.9	98.8	145001
CDH1	104.5	99.2	99.0	114480;176807;176430

CDK4	100.0	100.0	99.1	609048
CDKN1A	171.9	100.0	100.0	-
CDKN1B	151.5	99.9	99.4	610755
CDKN1C	100.1	89.8	81.7	130650
CDKN2A	121.7	92.3	92.3	606719;155601;155755
CDKN2B	121.9	100.0	100.0	-
CDKN2C	139.7	100.0	100.0	-
CEBPA	139.8	99.9	99.1	601626
CHEK2	88.6	83.8	80.1	114480;176807;609265
CREBBP	110.7	99.4	97.0	180849
CTC1	105.5	100.0	99.3	612199
CTNNA1	109.6	99.1	97.2	4
CTR9	142.5	100.0	99.9	16
CYLD	109.2	99.7	97.8	132700;601606
DDB2	147.3	99.8	98.4	278740
DDX11	100.7	86.7	81.2	613398
DICER1	137.9	99.8	98.4	138800;601200;180295
DIS3L2	143.3	100.0	99.8	267000
DKC1	91.2	99.8	97.7	305000
DNAJC21	128.1	99.9	99.5	617052
EGFR	135.2	100.0	100.0	211980
EGLN1	102.0	97.6	85.9	-
EGLN2	151.2	100.0	100.0	-
ELANE	141.5	100.0	99.3	162800;202700
EPCAM	76.5	99.7	95.7	613244
ERCC1	85.7	100.0	98.1	610758
ERCC2	128.0	100.0	99.8	278730
ERCC3	92.0	99.9	98.4	610651
ERCC4	132.0	100.0	99.8	615272;610965;278760
ERCC5	126.3	99.9	99.5	278780

ERCC6	158.2	100.0	99.9	211980;133540;214150;600630
ESR2	111.6	100.0	99.6	17
ETV6	148.3	100.0	99.4	616216
EXT1	88.6	99.6	98.0	215300;133700
EXT2	118.0	99.9	99.1	133701
EZH2	130.0	99.4	97.6	277590
FAN1	132.2	100.0	99.9	6
FANCA	112.4	99.9	98.9	227650
FANCB	76.4	98.6	93.2	300514
FANCC	100.8	99.7	99.2	613899;227645
FANCD2	115.6	99.1	96.6	227646
FANCE	118.2	96.6	89.9	600901
FANCF	244.4	100.0	100.0	603467
FANCG	140.7	100.0	99.8	614082
FANCI	136.2	99.9	98.9	609053
FANCL	105.8	99.7	98.0	614083
FANCM	100.6	99.3	97.1	614087
FAS	226.0	99.9	99.6	601859
FH	128.0	95.0	88.5	150800;606812
FLCN	152.3	100.0	100.0	135150
G6PC3	114.6	100.0	100.0	612541
GATA2	115.0	100.0	99.0	614286
GDNF	183.5	100.0	100.0	171300
GFI1	105.7	100.0	100.0	613107;607847
GPC3	75.7	98.7	92.7	312870
GREM1	106.8	100.0	100.0	7
GRHL2	116.8	100.0	100.0	616029
HABP2	109.3	100.0	99.4	616535
HAX1	137.4	100.0	100.0	610738
HNF1A	163.2	100.0	99.9	144700

HOXB13	186.3	100.0	100.0	610997
IDH1	78.0	89.4	77.3	614569
IDH2	98.5	100.0	99.6	614569
IPMK	88.0	98.5	89.6	15
KIF1B	139.6	100.0	99.6	171300
KIT	136.2	100.0	99.6	601626;273300;154800;606764
KLLN	152.3	100.0	100.0	615107
KRAS	67.2	99.4	97.3	601626
LIG4	173.4	100.0	99.8	606593
LZTR1	143.6	100.0	99.7	615670
MAP2K1	92.3	99.5	96.3	615279
MAP2K2	124.2	98.5	94.1	615280
MAX	80.0	99.9	98.0	171300
MDH2	109.4	98.0	97.9	8
MEN1	132.0	100.0	99.5	131100
MET	151.3	99.9	99.3	605074
MITF	141.1	100.0	99.8	614456
MLH1	139.2	99.9	99.3	276300;609310;158320
MPL	125.8	100.0	99.8	601977;604498
MRE11	49.7	97.3	86.0	604391
MSH2	111.7	99.4	96.4	276300;120435;158320
MSH3	139.6	99.8	99.2	617100
MSH6	165.1	100.0	100.0	276300;614350;608089
MUC5B	85.5	82.9	72.7	178500
MUTYH	152.0	100.0	100.0	608456;132600
NBN	93.8	99.8	98.4	251260
NF1	106.2	92.5	89.4	193520;607785;162200;162210;601321
NF2	94.2	100.0	99.6	607174;101000;162091
NHP2	121.9	100.0	99.2	613987
NOP10	120.5	100.0	100.0	224230

NPM1	65.0	94.5	83.5	601626
NRAS	145.5	100.0	100.0	613224
NSD1	147.0	100.0	99.8	117550;601626
NTHL1	121.6	100.0	100.0	602656
PALB2	143.5	100.0	99.9	114480;610832;613348
PARN	127.3	99.9	99.5	616353
PAX5	105.6	99.2	96.0	167414
PDGFB	115.4	100.0	100.0	607174
PDGFRA	124.7	100.0	100.0	173490
PHOX2B	145.5	100.0	100.0	613013
PIK3CA	127.7	100.0	99.8	612918
PMS2	94.7	83.4	81.0	276300;614337
PMS2CL				9
POLD1	124.5	98.0	93.9	612591
POLE	126.9	99.9	99.4	615083
POLH	116.2	99.9	98.6	278750
POT1	97.7	99.9	98.5	615848
POU6F2	135.1	100.0	100.0	601583
PPM1D	170.2	100.0	99.6	114480
PRF1	138.1	91.2	90.6	603553;605027
PRKAR1A	79.4	98.6	92.6	255960;160980;610489
PRKN	82.1	79.9	78.1	608935
PRSS1	141.4	100.0	99.9	167800
PTCH1	110.2	99.9	98.4	109400
PTCH2	120.3	99.9	98.7	109400
PTEN	129.7	99.6	97.0	607174;605309;158350;153480;613028
PTPN11	78.3	98.6	90.7	163950
RAD50	102.0	97.5	91.1	613078
RAD51C	140.6	99.9	99.5	613390
RAD51D	140.9	100.0	99.4	614291

RAF1	108.3	100.0	99.9	611553
RB1	89.8	97.8	93.1	180200
RECQL	144.1	99.8	98.7	10
RECQL4	159.9	100.0	99.8	218600;268400;266280
REST	115.5	98.5	98.4	616806
RET	136.8	100.0	99.2	171400;171300;162300;155240
RHBDF2	105.1	99.9	98.9	148500
RINT1	158.0	99.8	98.2	11
RIT1	139.2	100.0	100.0	615355
RNF43	145.9	100.0	98.8	617108
RPL11	85.4	100.0	99.3	612562
RPL35A	75.4	96.4	84.6	612528
RPL5	34.7	85.0	67.7	612561
RPS10	91.8	98.8	91.8	613308
RPS17	38.2	87.0	68.9	612527
RPS19	76.7	99.9	96.6	105650
RPS20	58.1	97.9	88.8	-
RPS24	84.5	95.2	89.7	610629
RPS26	75.9	89.2	75.8	613309
RPS7	76.6	84.8	70.0	612563
RTEL1	131.1	99.7	97.7	615190
RUNX1	84.6	99.6	96.3	601626;601399
SAMD9	163.9	100.0	99.9	617053
SAMD9L	171.8	100.0	100.0	159550
SBDS	166.2	100.0	100.0	260400
SDHA	88.9	85.1	77.7	252011;614165
SDHAF2	127.7	95.6	94.6	601650
SDHB	114.8	100.0	99.9	115310171300;606864;606764;612359
SDHC	85.8	99.6	95.3	605373;606864;606764
SDHD	43.7	52.7	50.6	171300;606864;615106;114900;168000

SEMA4A	124.3	100.0	99.3	12
SFTPA1	155.5	100.0	100.0	178500
SFTPA2	141.1	100.0	100.0	178500
SH2B3	108.4	99.9	97.6	19
SHOC2	139.6	99.9	99.4	607721
SLX4	124.2	100.0	99.7	613951
SMAD4	108.9	100.0	99.9	175050;174900;139210
SMAD9	110.0	100.0	100.0	13
SMARCA4	150.9	100.0	99.4	613325
SMARCB1	179.1	100.0	99.9	609322;614608;162091
SMARCE1	66.5	94.4	84.2	607174
SOS1	102.0	99.6	97.4	610733
SPINK1	85.0	100.0	99.4	167800
SPRED1	146.5	99.8	98.8	611431
SQSTM1	117.8	99.9	99.2	602080
STK11	131.0	100.0	100.0	260350;175200
SUFU	132.8	100.0	99.9	155255;607174;109400
TERC				614743;127550
TERF2IP	128.4	100.0	99.6	18
TERT	144.1	99.7	97.6	613989;615134;614742
TINF2	177.1	100.0	100.0	613990;268130
TMEM127	112.0	100.0	99.8	171300
TNFRSF11A	131.0	96.1	95.2	602080
TP53	89.2	99.8	98.5	114480;202300;260350;151623;137800;259500;607107;114500;614740
TRIP13	127.4	100.0	99.9	617598
TSC1	112.5	99.6	98.2	191100
TSC2	140.5	100.0	99.9	613254
USB1	118.2	99.8	97.2	604173
VHL	169.6	100.0	98.3	193300;171300;263400
WAS	70.4	94.2	83.6	313900;300299

WRAP53	162.8	100.0	100.0	613988
WRN	124.8	99.7	98.8	277700
WT1	90.1	100.0	99.3	194070
XPA	74.7	99.7	98.2	278700
XPC	143.5	100.0	99.8	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