

WES HEREDITARY CANCER DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
A2ML1	130.8	100.0	99.7	-
ACD	135.2	100.0	98.2	616553
AIP	154.2	99.9	99.0	102200
ALK	128.8	99.4	98.1	613014
ANKRD26	81.0	88.7	76.8	188000
APC	159.0	99.9	98.9	175100
ARMC5	136.5	99.8	97.5	615954
ASXL1	159.8	99.1	97.7	614286
ATM	109.7	99.0	94.0	114480
ATR	138.3	99.4	96.9	614564
AXIN2	114.5	99.7	98.9	608615
BAP1	111.0	85.1	82.7	614327
BARD1	150.3	100.0	99.7	114480
BLM	116.3	99.4	96.5	210900
BMPR1A	98.0	99.7	94.2	174900
BRAF	74.4	87.6	77.2	613706
BRCA1	177.5	98.9	96.9	604370
BRCA2	102.7	99.0	97.4	612555
BRIP1	117.8	99.8	97.7	114480
BUB1	136.4	99.8	97.9	1
BUB1B	136.5	98.6	97.9	257300;176430
BUB3	146.1	98.6	97.8	2
CBL	129.8	96.9	95.7	613563
CDC73	102.7	99.8	97.7	145001
CDH1	125.3	99.1	98.4	114480;176807;176430

CDK4	128.0	100.0	99.6	609048
CDKN1A	145.9	100.0	100.0	-
CDKN1B	93.2	100.0	99.5	610755
CDKN1C	21.1	68.1	51.8	130650
CDKN2A	79.6	92.2	91.4	606719;155601;155755
CDKN2B	85.9	100.0	99.8	-
CDKN2C	153.6	100.0	100.0	-
CEBPA	46.7	75.5	65.1	601626
CHEK2	100.8	82.4	78.7	114480;176807;609265
CREBBP	123.5	99.4	96.7	180849
CTC1	119.0	100.0	99.8	612199
CTNNA1	125.6	99.9	99.0	4
CTR9	154.2	100.0	99.8	16
CYLD	119.9	98.1	93.0	132700;601606
DDB2	162.4	100.0	99.7	278740
DDX11	113.9	86.0	81.0	613398
DICER1	145.4	99.5	98.1	138800;601200;180295
DIS3L2	158.5	99.8	99.0	267000
DKC1	111.9	99.6	98.1	305000
DNAJC21	125.7	99.8	98.5	617052
EGFR	160.8	100.0	99.1	211980
ELANE	80.9	99.7	95.9	162800;202700
EPCAM	64.5	93.3	79.8	613244
ERCC1	76.6	100.0	97.3	610758
ERCC2	123.7	100.0	99.7	278730
ERCC3	113.2	99.9	98.9	610651
ERCC4	139.2	100.0	99.5	615272;610965;278760
ERCC5	139.8	100.0	99.4	278780
ERCC6	191.3	100.0	99.9	211980;133540;214150;600630
ESR2	133.7	100.0	99.5	17

ETV6	140.1	100.0	99.9	616216
EXT1	105.4	99.9	98.5	215300;133700
EXT2	163.5	99.9	99.1	133701
EZH2	139.5	99.8	97.6	277590
FAN1	143.9	100.0	99.9	6
FANCA	123.3	99.8	98.5	227650
FANCB	68.4	96.7	87.9	300514
FANCC	121.6	99.4	97.1	613899;227645
FANCD2	127.6	98.7	95.5	227646
FANCE	108.0	85.9	84.6	600901
FANCF	166.8	100.0	100.0	603467
FANCG	147.7	100.0	100.0	614082
FANCI	152.1	99.5	97.5	609053
FANCL	87.8	99.4	94.7	614083
FANCM	96.8	99.2	94.3	614087
FAS	272.1	100.0	99.3	601859
FH	146.4	91.7	87.6	150800;606812
FLCN	160.5	100.0	99.5	135150
G6PC3	123.7	100.0	100.0	612541
GATA2	119.6	99.9	98.5	614286
GDNF	185.9	99.9	98.8	171300
GFI1	83.1	99.0	92.9	613107;607847
GPC3	85.1	98.6	92.6	312870
GREM1	95.1	100.0	100.0	7
GRHL2	134.6	100.0	100.0	616029
HABP2	136.9	100.0	99.6	616535
HAX1	136.5	100.0	100.0	610738
HNF1A	156.7	100.0	99.4	144700
HOXB13	127.8	99.9	97.7	610997
IPMK	95.5	96.9	83.9	15

KIF1B	154.8	100.0	99.5	171300
KIT	153.0	100.0	99.7	601626;273300;154800;606764
KLLN	117.1	100.0	100.0	615107
KRAS	64.7	99.9	98.7	601626
LIG4	165.6	100.0	99.6	606593
LZTR1	134.0	100.0	99.4	615670
MAP2K1	92.3	99.8	95.6	615279
MAP2K2	107.9	97.6	89.2	615280
MAX	86.6	99.4	96.4	171300
MDH2	123.3	98.0	97.9	8
MEN1	123.2	99.5	96.3	131100
MET	184.7	100.0	99.6	605074
MITF	155.5	100.0	99.9	614456
MLH1	162.0	100.0	99.7	276300;609310;158320
MPL	136.7	99.6	97.5	601977;604498
MRE11	51.2	95.3	82.3	604391
MSH2	113.4	98.6	93.1	276300;120435;158320
MSH3	113.4	99.0	95.0	617100
MSH6	171.1	100.0	99.5	276300;614350;608089
MUC5B	98.5	87.7	82.4	178500
MUTYH	165.0	100.0	99.9	608456;132600
NBN	80.6	99.1	94.6	251260
NF1	125.9	92.3	89.3	193520;607785;162200;162210;601321
NF2	100.2	100.0	99.9	607174;101000;162091
NHP2	111.0	100.0	100.0	613987
NOP10	160.5	100.0	100.0	224230
NPM1	70.3	88.7	77.9	601626
NRAS	188.4	100.0	100.0	613224
NSD1	155.2	100.0	99.9	117550;601626
NTHL1	98.1	99.1	95.7	602656

OGG1	128.5	100.0	99.6	144700
PALB2	152.6	100.0	99.7	114480;610832;613348
PARN	128.4	99.9	98.0	616353
PAX5	118.8	98.3	95.4	167414
PDGFB	95.1	100.0	100.0	607174
PHOX2B	92.9	93.0	87.2	613013
PIK3CA	120.7	99.9	99.1	612918
PMS2	95.1	83.5	80.7	276300;614337
PMS2CL				9
POLD1	101.2	93.9	90.8	612591
POLE	144.1	100.0	99.5	615083
POLH	140.7	100.0	99.8	278750
POT1	90.7	99.6	96.0	615848
POU6F2	142.4	100.0	99.9	601583
PPM1D	166.7	100.0	99.8	114480
PRF1	122.5	91.2	90.8	603553;605027
PRKAR1A	90.7	99.1	93.9	255960;160980;610489
PRKN	98.6	79.6	78.8	608935
PRSS1	190.8	100.0	99.9	167800
PTCH1	114.6	98.4	95.9	109400
PTCH2	120.1	99.4	97.5	109400
PTEN	143.2	99.6	96.0	607174;605309;158350;153480;613028
PTPN11	103.1	97.9	92.5	163950
RAD50	99.0	92.6	86.2	613078
RAD51C	143.4	100.0	98.9	613390
RAD51D	153.9	100.0	99.5	614291
RAF1	127.3	100.0	99.7	611553
RB1	88.0	90.1	76.3	180200
RECQL	127.4	99.7	96.1	10
RECQL4	149.6	99.2	96.5	218600;268400;266280

REST	128.6	98.5	98.4	616806
RET	141.0	99.7	97.8	171400;171300;162300;155240
RHBDF2	97.7	99.5	97.0	148500
RINT1	171.0	99.7	97.5	11
RIT1	165.6	100.0	100.0	615355
RNF43	126.5	100.0	99.6	617108
RPL11	99.8	100.0	99.5	612562
RPL35A	83.9	99.2	91.0	612528
RPL5	43.8	81.8	69.3	612561
RPS10	140.1	99.8	97.1	613308
RPS17	52.4	85.0	73.7	612527
RPS19	82.5	99.7	95.5	105650
RPS20	74.7	99.7	94.1	-
RPS24	110.4	92.4	87.2	610629
RPS26	106.8	94.8	82.1	613309
RPS7	93.7	76.9	63.4	612563
RTEL1	110.9	99.2	95.1	615190
RUNX1	92.0	97.2	89.7	601626;601399
SAMD9	159.1	99.9	99.3	617053
SAMD9L	165.7	100.0	99.9	159550
SBDS	212.3	100.0	99.9	260400
SDHA	122.2	84.8	80.8	252011;614165
SDHAF2	144.3	94.7	94.3	601650
SDHB	120.3	100.0	99.3	115310171300;606864;606764;612359
SDHC	100.1	99.8	96.8	605373;606864;606764
SDHD	48.4	55.2	50.4	171300;606864;615106;114900;168000
SEMA4A	127.5	99.9	98.9	12
SFTPA1	175.5	99.8	98.3	178500
SFTPA2	167.9	100.0	99.1	178500
SH2B3	97.5	90.7	79.0	19

SHOC2	140.4	100.0	99.4	607721
SLX4	114.2	100.0	99.8	613951
SMAD4	125.5	100.0	100.0	175050;174900;139210
SMAD9	132.8	100.0	100.0	13
SMARCA4	143.8	100.0	99.5	613325
SMARCB1	214.3	100.0	100.0	609322;614608;162091
SMARCE1	73.6	96.5	86.8	607174
SOS1	94.3	96.7	90.3	610733
SPINK1	80.2	100.0	99.4	167800
SPRED1	164.3	98.7	96.7	611431
SQSTM1	109.1	98.6	94.5	602080
STK11	111.9	99.7	95.8	260350;175200
SUFU	122.6	99.9	99.0	155255;607174;109400
TERC				614743;127550
TERF2IP	116.7	100.0	97.6	18
TERT	138.3	95.3	92.0	613989;615134;614742
TINF2	184.0	100.0	100.0	613990;268130
TMEM127	107.4	98.2	94.7	171300
TNFRSF11A	146.3	93.3	91.4	602080
TP53	92.0	99.9	98.1	114480;202300;260350;151623;137800;259500;607107;114500;614740
TRIP13	141.2	100.0	100.0	617598
TSC1	128.8	99.8	98.8	191100
TSC2	131.2	100.0	99.0	613254
USB1	125.0	99.9	98.2	604173
VHL	119.7	92.6	85.3	193300;171300;263400
WAS	66.1	88.2	78.7	313900;300299
WRAP53	154.4	100.0	100.0	613988
WRN	123.6	98.3	94.6	277700
WT1	76.5	91.8	81.4	194070
XPA	52.9	98.5	88.9	278700

XPC	140.7	100.0	99.7	278720
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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