WES HEREDITARY CANCER DG 3.1

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
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	145.8	99.8	99.3	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	80.6	91.0	81.1	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
CD27	103.3	99.9	96.9	No OMIM phenotype
CD70	126.0	99.8	97.7	No OMIM phenotype
CDC73	136.4	100.0	99.4	145001
CDH1	126.7	99.2	99.1	114480;176807;176430
CDH23	187.8	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	66.8	88.0	77.8	130650
CDKN2A	96.9	92.3	92.1	155601;606719;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	609265;114480;176807
CREBBP	124.1	99.7	98.5	180849
CTC1	128.0	100.0	99.6	612199
CTLA4	162.1	100.0	100.0	No OMIM phenotype
CTNNA1	125.7	99.3	98.1	-
CTR9	168.6	100.0	99.9	-
CYLD	127.1	99.8	98.0	601606;132700
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	143.8	99.8	98.7	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	202700;162800
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	107.8	96.9	96.3	610651
ERCC4	141.7	100.0	99.9	278760;610965;615272
ERCC5	143.4	100.0	99.7	278780
ERCC6	197.0	100.0	100.0	600630;211980;133540;214150
ESR2	122.2	100.0	99.7	-
ETV6	185.3	100.0	99.9	616216
EXT1	114.3	99.9	98.4	133700;215300
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	118.4	97.2	96.6	227645;613899
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

FBXW7	171.4	99.9	99.2	No OMIM phenotype
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	607847;613107
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
IKZF1	188.0	99.3	99.3	No OMIM phenotype
IPMK	112.5	99.2	92.0	-
ITK	126.2	100.0	98.9	No OMIM phenotype
KIF1B	167.4	100.0	99.6	171300
KIT	163.2	100.0	99.6	606764;154800;601626;273300
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
MCM8	136.8	100.0	99.6	No OMIM phenotype
MCM9	153.0	99.9	99.8	No OMIM phenotype
MDH2	116.9	98.0	97.9	-
MEN1	132.8	96.9	94.8	131100
MET	178.5	100.0	99.5	605074
MITF	160.6	100.0	99.9	614456
MLH1	166.0	100.0	99.9	609310;276300;158320
MPL	140.2	100.0	99.5	601977;604498
MRE11	64.1	98.9	93.3	604391
MSH2	141.4	99.0	96.9	276300;158320;120435
MSH3	160.0	98.0	97.3	617100
MSH6	179.9	100.0	99.8	614350;608089;276300
MTAP	108.3	99.1	93.5	112250
MUTYH	154.3	100.0	100.0	132600;608456
NBN	109.6	99.9	98.6	251260
NF1	131.8	92.6	90.2	162200;193520;601321;607785;162210
NF2	119.7	100.0	99.9	101000;162091;607174
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	175.5	100.0	99.9	601626;117550
NTHL1	111.0	100.0	99.8	602656
PALB2	182.1	100.0	100.0	114480;610832;613348
PARN	134.2	81.2	81.1	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	151.3	98.0	97.8	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	147.8	95.2	95.2	601583
PPM1D	177.8	100.0	99.9	114480
PRF1	114.3	91.2	90.8	605027;603553
PRKAR1A	94.5	99.3	93.5	610489;160980;255960
PRKN	81.8	67.0	66.2	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;158350;153480;613028;605309
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	101.9	96.8	92.1	180200
RECQL4	149.8	99.8	98.1	266280;268400;218600
REST	119.0	98.5	98.2	616806
RET	139.5	99.9	99.1	155240;171300;171400;162300
RHBDF2	101.4	99.9	98.6	148500
RIT1	178.3	100.0	100.0	615355
RMRP				No OMIM phenotype
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	103.4	98.0	92.5	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	99.2	98.4	93.1	610629
RPS26	92.6	95.7	84.9	613309
RPS27	42.9	85.9	60.6	603702
RPS28	57.7	100.0	94.8	603685
RPS29	76.3	82.0	74.7	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	606764;606864;115310;171300
SDHC	107.5	100.0	99.3	605373;606764;606864
SDHD	48.5	54.0	51.6	171300;168000;114900;615106;606864
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	-
SH2D1A	122.6	97.2	94.0	No OMIM phenotype
SHOC2	164.2	99.9	99.4	607721
SLX4	130.9	100.0	99.8	613951
SMAD4	127.2	100.0	99.9	174900;175050;139210
SMAD9	118.3	100.0	99.9	-
SMARCA4	157.4	99.9	99.0	613325
SMARCB1	210.4	100.0	100.0	614608;162091;609322
SMARCE1	86.1	95.6	88.8	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	119.0	92.4	91.7	175200;260350
SUFU	153.1	100.0	100.0	155255;109400;607174
TERC				127550;614743
TERF2IP	112.6	99.9	97.8	-
TERT	132.4	96.2	94.5	615134;614742;613989
TG	137.4	100.0	99.4	274700
THPO	83.3	81.4	81.0	187950
TINF2	187.5	100.0	100.0	268130;613990
TMEM127	102.5	99.5	96.5	171300
TNFRSF11A	141.5	94.6	93.3	602080
TP53	113.9	99.9	97.7	114480;151623;260350;137800;202300;114500;614740;607107;259500
TRIM28	153.6	96.8	95.2	No OMIM phenotype
TRIM37	141.7	98.6	98.1	No OMIM phenotype
TRIP13	165.0	100.0	100.0	617598
TSC1	137.9	99.8	98.8	191100
TSC2	143.7	100.0	99.6	613254
USB1	139.6	100.0	99.4	604173

VHL	141.3	96.3	91.4	263400;171300;193300
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	81.1	97.3	95.4	194070
XPA	83.7	99.6	95.6	278700
XPC	162.6	100.0	100.0	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.
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 10x 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