

# WES INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION TO HEMATOLOGICAL MALIGNANCIES

## DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ABCB7	122.8	99.5	97.1	301310
ABCD4	135.3	99.8	97.7	614857
ACBD5	150.7	100.0	98.4	-
ACD	159.3	100.0	99.9	616553
AMN	86.5	92.5	82.9	261100
ANKRD26	82.9	94.6	88.5	188000
AP3B1	114.7	99.2	96.4	608233
ASXL1	128.2	99.8	98.9	614286
ATR	164.5	99.7	99.0	210600
BLM	113.6	99.3	97.7	210900
BRAF	63.9	89.4	77.6	613706;613707;115150
BRCA1	167.7	99.4	98.4	604370;617883;614320
BRCA2	97.7	99.1	98.2	612555;605724
BRIP1	129.1	99.4	98.5	114480;609054
CASP10	109.9	99.3	96.8	605027;603909
CBL	133.8	97.3	96.9	613563;607785
CEBPA	84.4	95.9	80.1	601626
CLPB	121.2	94.9	94.0	616271
CSF3R	107.9	99.8	98.5	617014;162830
CTC1	116.1	100.0	99.1	612199

CTLA4	128.1	100.0	100.0	616100
CUBN	107.3	99.2	97.1	261100
CXCR4	130.5	100.0	100.0	193670
DBF4	86.1	96.0	88.9	No OMIM phenotype
DDX41	158.2	100.0	100.0	616871
DHFR	43.9	88.9	76.3	613839
DICER1	148.8	99.5	98.5	180295;601200;138800;618272
DKC1	90.7	99.7	97.2	305000
DNAJC21	117.0	99.5	97.4	617052
EFL1	154.0	99.3	97.7	617941
ELANE	128.3	99.9	98.8	202700;162800
EPO	105.9	99.9	97.8	617907;617911
ERCC4	122.5	100.0	99.9	278760;610965;615272
ERCC6L2	135.5	99.6	98.6	615715
ETV6	177.8	100.0	99.3	616216
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
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	227650
FAS	337.2	100.0	99.6	601859
FASLG	95.5	100.0	99.1	601859
G6PC3	116.4	100.0	99.9	612541
GATA1	106.2	99.9	98.5	314050;300835;300367

GATA2	109.3	99.8	97.0	614172
GBA	179.6	100.0	100.0	230900;231005;230800;231000
GFI1	88.9	100.0	99.9	607847;613107
GP1BA	142.5	97.6	94.3	231200;153670;177820
GP1BB	41.3	77.8	66.9	231200
GRHL2	125.6	100.0	99.9	616029
HAVCR2	122.7	100.0	99.8	618398
HAX1	148.7	100.0	100.0	610738
HOXA11	77.4	97.1	88.3	605432
IKZF1	183.4	99.3	99.3	616873
IKZF5	149.4	100.0	100.0	619130
IVD	90.4	100.0	99.9	243500
JAGN1	86.4	100.0	100.0	616022
KLF1	97.5	100.0	99.6	613673
KRAS	68.4	99.0	97.8	609942;615278;614470
LAPTM5	101.2	96.9	91.6	No OMIM phenotype
LIG4	183.8	99.8	99.3	606593
MAD2L2	147.2	100.0	99.9	617243
MCM4	125.6	95.3	95.0	609981
MDM4	147.1	100.0	98.8	618849
MECOM	133.8	100.0	99.6	616738
MLH1	139.2	100.0	99.9	609310;276300
MPL	132.8	100.0	99.8	601977;604498
MSH2	116.8	98.5	94.5	120435;276300
MSH6	147.4	100.0	99.3	614350;276300
MYH9	132.2	99.9	98.9	155100
MYSM1	111.1	96.1	95.4	618116
NBEAL2	169.7	99.5	99.3	139090
NBN	96.9	99.2	97.8	609135;251260;613065
NF1	117.2	91.8	89.3	162200;607785

NFE2	107.5	100.0	100.0	No OMIM phenotype
NHP2	143.7	100.0	100.0	613987
NOP10	106.3	100.0	99.2	224230
NPAT	127.8	99.7	98.8	No OMIM phenotype
NPM1	78.3	95.3	84.9	-
NRAS	163.7	100.0	100.0	613224;614470
PALB2	150.5	100.0	99.9	114480;610832;613348
PARN	110.0	81.1	80.4	616353;616371
PAX5	100.5	98.8	95.4	615545
PMS2	100.9	83.9	81.6	614337;276300
POT1	105.9	99.5	98.5	615848
PRF1	121.1	91.2	90.1	605027;609135;603553
PTPN11	81.2	97.7	87.6	163950;607785;151100
RAD51	96.5	89.4	89.4	114480;617244
RAD51C	143.5	99.8	99.4	613390;613399
RBBP6	110.5	97.9	95.2	No OMIM phenotype
RBM8A	94.3	99.6	95.3	274000
RFWD3	110.6	100.0	99.4	No OMIM phenotype
RMRP				250250;250460;607095
RPL11	89.1	99.9	97.9	612562
RPL15	31.8	84.9	70.4	615550
RPL18	112.1	100.0	99.9	618310
RPL26	36.0	94.2	75.5	614900
RPL27	33.8	68.0	56.6	617408
RPL31	82.0	97.6	87.4	-
RPL35	61.0	90.1	79.1	618312
RPL35A	80.0	94.7	84.9	612528
RPL4	45.1	83.3	73.3	No OMIM phenotype
RPL5	32.6	81.9	59.7	612561
RPL9	85.3	98.6	88.0	-

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
RPS24	79.4	96.2	90.3	610629
RPS26	89.3	93.2	81.2	613309
RPS27	38.2	95.5	70.0	617409
RPS28	47.1	99.7	86.3	606164
RPS29	61.7	78.0	70.5	615909
RPS7	89.6	81.7	66.9	612563
RTEL1	130.0	99.7	97.2	615190;616373
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
SH2B3	113.3	99.0	94.7	133100;254450;187950
SH2D1A	105.1	97.8	92.9	308240
SLC19A2	95.1	100.0	98.5	249270
SLC25A38	98.1	97.4	93.3	205950
SLC37A4	118.5	99.8	97.6	232240;232220
SLC46A1	103.5	100.0	98.5	229050
SLX4	124.7	100.0	99.9	613951
SOS1	104.8	99.6	97.9	610733
SRP54	111.1	98.0	93.4	618752
SRP72	72.5	98.0	90.1	614675
STIM1	121.5	99.9	97.5	185070;612783
STN1	86.6	99.9	99.8	617341
TAZ	104.7	99.3	93.7	302060
TBXAS1	131.9	100.0	100.0	614158;231095
TCIRG1	118.4	98.5	93.4	259700

TERC				127550;614743
TERF2IP	115.7	100.0	99.9	No OMIM phenotype
TERT	135.5	97.0	94.8	613989;614742
TET2	163.0	100.0	100.0	619126;614286
THPO	74.2	81.4	78.7	187950
TINF2	160.3	100.0	100.0	268130;613990
TLR8	149.5	99.9	99.8	No OMIM phenotype
TP53	94.2	99.0	95.2	618165;151623
TSR2	70.3	99.9	98.3	300946
TYK2	133.6	100.0	99.3	511521
UBA1	134.8	99.2	97.3	301054
UBE2T	94.4	99.9	99.3	616435
USB1	131.2	100.0	98.8	604173
VPS45	129.0	97.8	95.1	615285
WAS	75.5	94.1	83.7	301000;313900;300299
WRAP53	164.2	100.0	100.0	613988
XRCC2	161.0	99.6	95.7	617247
YARS2	171.1	99.9	99.4	613561
ZCCHC8	115.0	99.7	98.0	618674

*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.*

*Coverage stated is using Agilent V5 chemistry, used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.*

*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.*