WES INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION TO HEMATOLOGICAL MALIGNANCIES DG 3.1

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ABCB7	148.3	99.5	98.2	301310
ABCD4	141.4	99.9	98.6	614857
ACBD5	181.1	100.0	99.2	-
ACD	148.6	100.0	99.9	616553
AMN	77.8	89.7	80.0	261100
ANKRD26	94.5	95.0	89.3	188000
ASXL1	145.8	99.8	99.3	614286
ATR	178.2	99.9	99.4	210600
BLM	133.4	99.8	98.3	210900
BRAF	80.6	91.0	81.1	613706;613707;115150
BRCA1	195.0	99.4	98.8	604370;617883;614320
BRCA2	119.2	99.8	98.5	612555;605724
BRIP1	151.0	99.9	99.0	114480;609054
CASP10	129.9	99.5	97.3	605027;603909
CBL	152.5	97.3	97.1	613563;607785
CEBPA	84.1	98.6	83.9	601626
CLPB	135.7	94.9	94.9	616271
CSF3R	108.5	99.6	98.2	617014;162830
CTC1	128.0	100.0	99.6	612199
CTLA4	162.1	100.0	100.0	616100

CUBN	129.7	99.7	98.3	261100
CXCR4	151.1	100.0	100.0	193670
DBF4	99.1	96.6	89.6	No OMIM phenotype
DDX41	161.7	100.0	100.0	616871
DHFR	51.8	92.1	78.9	613839
DICER1	179.1	99.8	99.0	180295;601200;138800;618272
DKC1	108.6	99.8	98.7	305000
DNAJC21	143.8	99.8	98.7	617052
EFL1	178.2	99.6	98.5	617941
ELANE	143.2	99.7	97.4	202700;162800
EPO	102.8	99.9	97.6	617907;617911
ERCC4	141.7	100.0	99.9	278760;610965;615272
ERCC6L2	158.4	99.9	99.2	615715
ETV6	185.3	100.0	99.9	616216
EZH2	171.4	100.0	99.5	277590
FANCA	124.7	100.0	99.4	227650
FANCB	88.6	98.6	94.1	300514
FANCC	118.4	97.2	96.6	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	227650
FAS	262.8	100.0	99.6	601859
FASLG	98.0	100.0	99.6	601859
G6PC3	113.2	100.0	99.9	612541
GATA1	108.4	99.8	98.4	314050;300835;300367
GATA2	119.1	100.0	98.3	614172

GBA	202.3	100.0	100.0	230900;231005;230800;231000
GFI1	88.3	100.0	99.2	607847;613107
GP1BA	169.1	98.6	95.9	231200;153670;177820
GP1BB	32.0	72.9	59.6	231200
GRHL2	139.4	100.0	100.0	616029
HAVCR2	141.9	100.0	100.0	618398
HAX1	166.6	100.0	100.0	610738
HOXA11	95.6	97.1	87.5	605432
IKZF1	188.0	99.3	99.3	616873
IKZF5	176.8	100.0	100.0	619130
IVD	101.1	100.0	100.0	243500
JAGN1	119.7	100.0	100.0	616022
KLF1	87.9	100.0	97.8	613673
KRAS	84.3	99.5	96.9	609942;615278;614470
LAPTM5	109.7	97.9	92.9	No OMIM phenotype
LIG4	222.9	100.0	99.9	606593
MAD2L2	151.7	100.0	99.9	617243
MDM4	182.7	99.9	99.0	618849
MECOM	157.3	100.0	99.9	616738
MLH1	166.0	100.0	99.9	609310;276300
MPL	140.2	100.0	99.5	601977;604498
MSH2	141.4	99.0	96.9	120435;276300
MSH6	179.9	100.0	99.8	614350;276300
MYH9	134.9	100.0	99.3	155100
MYSM1	130.4	96.4	95.5	618116
NBEAL2	166.4	99.4	99.4	139090
NBN	109.6	99.9	98.6	609135;2151260;613065
NF1	131.8	92.6	90.2	162200;607785
NHP2	146.1	100.0	100.0	613987
NOP10	147.4	100.0	99.8	224230

NPAT	144.2	99.8	98.7	No OMIM phenotype
NPM1	90.1	98.2	85.3	-
NRAS	185.6	100.0	100.0	613224;614470
PALB2	182.1	100.0	100.0	114480;610832;613348
PARN	134.2	81.2	81.1	616353;616371
PAX5	115.0	98.7	96.1	615545
PMS2	125.2	84.3	82.8	614337;276300
POT1	120.7	99.9	99.0	615848
PRF1	114.3	91.2	90.8	605027;609135;603553
PTPN11	100.3	99.1	93.7	163950;607785;151100
RAD51	113.0	89.4	89.4	114480;617244
RAD51C	162.8	100.0	99.8	613390;613399
RBBP6	137.3	97.8	95.9	No OMIM phenotype
RBM8A	109.3	99.8	97.9	274000
RFWD3	128.7	100.0	99.8	No OMIM phenotype
RPL11	111.0	100.0	100.0	612562
RPL15	35.2	86.8	78.0	615550
RPL18	106.9	100.0	100.0	618310
RPL26	42.3	97.2	84.4	614900
RPL27	33.7	73.6	56.5	617408
RPL31	102.6	99.3	94.6	-
RPL35	63.9	86.4	75.0	618312
RPL35A	94.5	97.1	88.7	612528
RPL4	55.1	87.6	78.3	No OMIM phenotype
RPL5	42.2	86.2	70.0	612561
RPL9	97.5	98.9	92.0	-
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

RPS24	99.2	98.4	93.1	610629
RPS26	92.6	95.7	84.9	613309
RPS27	42.9	85.9	60.6	617409
RPS28	57.7	100.0	94.8	606164
RPS29	76.3	82.0	74.7	615909
RPS7	112.0	80.0	68.7	612563
RTEL1	127.7	99.5	96.8	615190;616373
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
SH2B3	106.6	99.4	95.1	133100;254450;187950
SH2D1A	122.6	97.2	94.0	308240
SLC19A2	110.3	100.0	99.7	249270
SLC25A38	112.2	97.9	95.3	205950
SLC37A4	124.2	100.0	99.2	232240;232220
SLC46A1	116.3	99.9	98.5	229050
SLX4	130.9	100.0	99.8	613951
SOS1	123.6	99.8	98.4	610733
SRP54	133.2	99.5	96.5	618752
SRP72	81.8	97.6	89.7	614675
STIM1	126.2	99.8	98.0	185070;612783
STN1	102.9	100.0	100.0	617341
TAZ	101.2	99.1	95.5	302060
TBXAS1	151.1	100.0	100.0	614158;231095
TCIRG1	116.3	97.6	90.1	259700
TERC				127550;614743
TERF2IP	112.6	99.9	97.8	No OMIM phenotype
TERT	132.4	96.2	94.5	613989;614742
TET2	201.7	100.0	100.0	619126;614286

THPO	83.3	81.4	81.0	187950
TINF2	187.5	100.0	100.0	268130;613990
TP53	113.9	99.9	97.7	618165;151623
TSR2	87.3	100.0	100.0	300946
TYK2	133.3	99.9	99.0	511521
UBA1	142.9	99.4	98.2	301054
UBE2T	109.4	100.0	99.9	616435
USB1	139.6	100.0	99.4	604173
VPS45	157.5	99.2	95.7	615285
WAS	81.1	95.9	85.3	301000;313900;300299
WRAP53	187.5	100.0	100.0	613988
XRCC2	205.8	99.8	97.4	617247
YARS2	194.6	100.0	99.8	613561
ZCCHC8	133.1	99.8	98.7	618674

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