

# WES INHERITED BONE MARROW FAILURE DG 2.18

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ABCB7	148.9	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
ATR	178.2	99.9	99.4	210600
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
CSF3R	108.5	99.6	98.2	162830;617014
CTC1	128.0	100.0	99.6	612199
CTLA4	162.1	100.0	100.0	616100
CUBN	129.7	99.7	98.3	261100
DHFR	51.8	92.1	78.9	613839
DKC1	108.6	99.8	98.7	305000
DNAJC21	146.2	99.9	99.0	617052
EFL1	178.2	99.6	98.5	617941
ELANE	143.2	99.7	97.4	162800;202700
ERCC4	141.7	100.0	99.9	615272;610965;278760
ERCC6L2	141.4	100.0	99.4	615715
ETV6	185.3	100.0	99.9	616216
FANCA	124.7	100.0	99.4	227650
FANCB	88.6	98.6	94.1	300514
FANCC	121.7	99.9	99.3	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
G6PC3	113.2	100.0	99.9	612541
GATA1	108.4	99.8	98.4	300367;300835;314050
GATA2	119.1	100.0	98.3	614172
GBA	202.3	100.0	100.0	231005;231000;230900;230800
GFI1	88.3	100.0	99.2	613107;607847
GP1BA	169.1	98.6	95.9	153670;231200;177820
GP1BB	32.0	72.9	59.6	231200
GRHL2	139.4	100.0	100.0	616029
HAX1	166.6	100.0	100.0	610738
HOXA11	95.6	97.1	87.5	605432
IVD	101.1	100.0	100.0	243500
JAGN1	124.1	100.0	100.0	616022
KLF1	87.9	100.0	97.8	613673
LIG4	222.9	100.0	99.9	606593
MAD2L2	151.7	100.0	99.9	617243
MECOM	157.3	100.0	99.9	616738
MPL	140.2	100.0	99.5	601977;604498
MYH9	134.9	100.0	99.3	155100
MYSM1	135.3	100.0	99.1	618116
NBEAL2	166.7	99.4	99.3	139090
NHP2	146.1	100.0	100.0	613987
NOP10	147.4	100.0	99.8	224230
NPM1	90.1	98.2	85.3	-

PALB2	182.1	100.0	100.0	114480;610832;613348
PARN	165.2	100.0	99.9	616371;616353
POT1	120.7	99.9	99.0	615848
PRF1	114.3	91.2	90.8	609135;603553;605027
RAD51	113.0	89.4	89.4	114480;617244
RAD51C	162.8	100.0	99.8	613390;613399
RBM8A	109.3	99.8	97.9	274000
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	-
RPL35A	94.5	97.1	88.7	612528
RPL5	42.2	86.2	70.0	612561
RPL9	76.2	98.7	90.8	-
RPS10	109.9	97.7	91.7	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	98.4	98.5	93.4	610629
RPS26	92.6	95.7	84.9	613309
RPS27	39.4	89.3	60.9	617409
RPS28	57.7	100.0	94.8	606164
RPS29	113.4	100.0	98.2	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
SH2D1A	122.6	97.2	94.0	308240
SLC19A2	110.3	100.0	99.7	249270
SLC25A38	114.5	99.7	97.1	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
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
TBXAS1	151.1	100.0	100.0	231095;614158
TCIRG1	116.3	97.6	90.1	259700
TERC				614743;127550
TERT	132.4	96.2	94.5	613989
THPO	104.3	100.0	99.5	187950
TINF2	187.5	100.0	100.0	613990;268130
TSR2	87.3	100.0	100.0	300946
UBE2T	109.4	100.0	99.9	616435
USB1	139.6	100.0	99.4	604173
VPS45	155.6	99.2	95.6	615285
WAS	81.1	95.9	85.3	313900;300299;301000
WRAP53	187.5	100.0	100.0	613988
XRCC2	205.8	99.8	97.4	617247

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