

WES INHERITED BONE MARROW FAILURE DG 2.16

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCB7	126.2	99.9	98.6	301310
ABCD4	129.0	99.8	98.4	614857
ACBD5	145.1	99.6	98.0	-
ACD	159.6	100.0	100.0	616553
AMN	101.5	98.1	90.6	261100
ANKRD26	83.3	95.3	90.1	188000
ATR	144.6	99.8	98.6	210600
BRCA1	161.4	99.1	98.1	604370;617883;614320
BRCA2	106.2	99.6	98.7	612555;605724
BRIP1	125.8	99.7	98.8	114480;609054
CSF3R	105.4	99.7	98.6	162830;617014
CTC1	105.5	100.0	99.3	612199
CTLA4	141.0	100.0	100.0	616100
CUBN	103.2	99.6	97.6	261100
DHFR	50.0	94.1	83.1	613839
DKC1	91.2	99.8	97.7	305000
DNAJC21	128.1	99.9	99.5	617052
EFL1	150.5	99.5	98.1	617941
ELANE	141.5	100.0	99.3	162800;202700
ERCC4	132.0	100.0	99.8	615272;610965;278760
ERCC6L2	121.7	99.9	99.0	615715
ETV6	148.3	100.0	99.4	616216
FANCA	112.4	99.9	98.9	227650
FANCB	76.4	98.6	93.2	300514
FANCC	100.8	99.7	99.2	227645

FANCD2	115.6	99.1	96.6	227646
FANCE	118.2	96.6	89.9	600901
FANCF	244.4	100.0	100.0	603467
FANCG	140.7	100.0	99.8	614082
FANCI	136.2	99.9	98.9	609053
FANCL	105.8	99.7	98.0	614083
FANCM	100.6	99.3	97.1	227650
G6PC3	114.6	100.0	100.0	612541
GATA1	92.9	99.9	98.2	300367;300835;314050
GATA2	115.0	100.0	99.0	614172
GBA	169.8	100.0	100.0	231005;231000;230900;230800
GFI1	105.7	100.0	100.0	613107;607847
GP1BA	136.8	98.7	95.7	153670;231200;177820
GP1BB	68.6	94.5	83.1	231200
GRHL2	116.8	100.0	100.0	616029
HAX1	137.4	100.0	100.0	610738
HOXA11	88.3	100.0	98.0	605432
IVD	100.0	100.0	99.9	243500
JAGN1	118.5	100.0	100.0	616022
KLF1	115.3	100.0	99.9	613673
LIG4	173.4	100.0	99.8	606593
MAD2L2	139.1	100.0	99.8	617243
MECOM	131.2	100.0	99.6	616738
MPL	125.8	100.0	99.8	601977;604498
MYH9	128.5	99.6	98.5	155100
MYSM1	111.0	99.8	98.4	618116
NBEAL2	166.0	100.0	99.5	139090
NHP2	121.9	100.0	99.2	613987
NOP10	120.5	100.0	100.0	224230
PALB2	143.5	100.0	99.9	114480;610832;613348

PARN	127.3	99.9	99.5	616371;616353
POT1	97.7	99.9	98.5	615848
PRF1	138.1	91.2	90.6	609135;603553;605027
RAD51	100.6	89.4	89.4	114480;617244
RAD51C	140.6	99.9	99.5	613390;613399
RBM8A	87.4	99.8	97.4	274000
RPL11	85.4	100.0	99.3	612562
RPL15	32.0	87.2	72.1	615550
RPL18	89.3	100.0	99.5	-
RPL26	31.0	91.7	68.9	614900
RPL27	32.5	72.7	54.6	617408
RPL31	72.9	98.6	93.7	-
RPL35A	75.4	96.4	84.6	612528
RPL5	34.7	85.0	67.7	612561
RPL9	67.9	98.4	86.4	-
RPS10	91.8	98.8	91.8	613308
RPS15A	58.3	97.1	86.3	-
RPS17	38.2	87.0	68.9	612527
RPS19	76.7	99.9	96.6	105650
RPS24	84.5	95.2	89.7	610629
RPS26	75.9	89.2	75.8	613309
RPS27	34.4	89.5	57.5	617409
RPS28	54.1	99.7	95.0	606164
RPS29	90.8	98.7	94.6	615909
RPS7	76.6	84.8	70.0	612563
RTEL1	131.1	99.7	97.7	615190
RUNX1	84.6	99.6	96.3	601626;601399
SAMD9	163.9	100.0	99.9	617053
SAMD9L	171.8	100.0	100.0	159550
SBDS	166.2	100.0	100.0	260400

SH2D1A	108.9	97.8	92.4	308240
SLC19A2	101.3	100.0	99.6	249270
SLC25A38	94.5	99.1	95.2	205950
SLC37A4	114.3	100.0	99.6	232240;232220
SLC46A1	111.1	99.9	98.4	229050
SLX4	124.2	100.0	99.7	613951
SRP72	69.2	95.7	85.6	614675
STIM1	120.7	99.8	96.8	185070;612783
STN1	82.2	100.0	99.6	617341
TBXAS1	128.8	100.0	100.0	231095;614158
TCIRG1	131.4	99.2	96.6	259700
TERC				614743;127550
TERT	144.1	99.7	97.6	613989
THPO	97.3	100.0	99.7	187950
TINF2	177.1	100.0	100.0	613990;268130
TSR2	76.1	100.0	99.4	300946
UBE2T	91.1	100.0	99.3	616435
USB1	118.2	99.8	97.2	604173
VPS45	126.5	97.3	94.4	615285
WAS	70.4	94.2	83.6	313900;300299;301000
WRAP53	162.8	100.0	100.0	613988
XRCC2	171.8	99.8	96.5	617247

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 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