

WES INHERITED BONE MARROW FAILURE DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCB7				301310
ABCD4				614857
ACBD5				-
ACD				616553
ANKRD26				188000
ATR				210600
BRCA1				604370;617883;614320
BRCA2				612555;605724
BRIP1				114480;609054
CSF3R				162830;617014
CTC1				612199
DHFR				613839
DKC1				305000
DNAJC21				617052
EFL1				617941
ELANE				162800;202700
ERCC4				615272;610965;278760
ERCC6L2				615715
ETV6				616216
FANCA				227650
FANCB				300514
FANCC				227645
FANCD2				227646
FANCE				600901
FANCF				603467

FANCG			614082
FANCI			609053
FANCL			614083
FANCM			227650
G6PC3			612541
GATA1			300367;300835;314050
GATA2			614172
GBA			231005;231000;230900;230800
GFI1			613107;607847
GP1BA			153670;231200;177820
GP1BB			231200
GRHL2			616029
HAX1			610738
HOXA11			605432
IVD			243500
JAGN1			616022
KLF1			613673
LIG4			606593
MAD2L2			617243
MECOM			616738
MPL			601977;604498
MYH9			155100
NBEAL2			139090
NHP2			613987
NOP10			224230
PALB2			114480;610832;613348
PARN			616371;616353
POT1			615848
PRF1			609135;603553;605027
RAD51			114480;617244

RAD51C				613390;613399
RBM8A				274000
RPL11				612562
RPL15				615550
RPL18				-
RPL26				614900
RPL27				617408
RPL31				-
RPL35A				612528
RPL5				612561
RPL9				-
RPS10				613308
RPS15A				-
RPS17				612527
RPS19				105650
RPS24				610629
RPS26				613309
RPS27				617409
RPS28				606164
RPS29				615909
RPS7				612563
RTEL1				615190
RUNX1				601626;601399
SAMD9				617053
SAMD9L				159550
SBDS				260400
SH2D1A				308240
SLC19A2				249270
SLC25A38				205950
SLC37A4				232240;232220

SLC46A1				229050
SLX4				613951
SRP72				614675
STIM1				185070;612783
STN1				617341
TBXAS1				231095;614158
TCIRG1				259700
TERC				614743;127550
TERT				613989
THPO				187950
TINF2				613990;268130
TSR2				300946
UBE2T				616435
USB1				604173
VPS45				615285
WAS				313900;300299;301000
WRAP53				613988
XRCC2				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