MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Bone Marrow Failure Gene Panel

The following applies to BMFGP / Inherited Bone Marrow Failure Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select noncoding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from July 2025 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ABCB7	NM_004299.6	-	-
ACD	NM_001082486.2	-	-
ADA	NM_000022.4	chr20:g.43249076C>T (c.976-34G>A); chr20:g.43248503A>T (c.1079-15T>A)	-
ADA2	NM_001282225.2	-	-
AK2	NM_001625.4	-	-
ALAS2	NM_000032.5	chrX:g.55054636 to chrX:g.55054634 (c.-15-2188 to c.-15-2186); chrX:g.55054628_55054663del (c.-15-2214_-15-2179del)	-
ANKRD26	NM_014915.2	chr10:g.27389395 to chr10:g.27389368 (c.-140 to c.-113)	CNV analysis in exon 19 is not performed
AP3B1	NM_003664.4	-	-
AP3D1	NM_001261826.3	-	-
ARPC1B	NM_005720.4	-	-
BLOC1S6	NM_012388.3	-	-
BRCA1	NM_007294.4	-	-
BRCA2	NM_000059.3	-	-
BRIP1	NM_032043.3	-	-
C15orf41 (CDIN1)	NM_001130010.3	-	-
CD27	NM_001242.4	-	-
CDAN1	NM_138477.4	-	-
CEBPA	NM_004364.4	-	-
CLCN7	NM_001287.6	-	CNV analysis in exon 17 is not performed
CLPB	NM_030813.6	-	-
CSF3R	NM_000760.4	-	-
CTC1	NM_025099.6	-	-
CXCR2	NM_001557.4	-	-
CXCR4	NM_003467.3	-	-
CYCS	NM_018947.6	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
DDX41	NM_016222.4	-	-
DHFR	NM_000791.4	-	CNV analysis in exon 6 is not performed
DIAPH1	NM_005219.5	-	-
DKC1	NM_001363.5	chrX:g.153991099C>G (c.-142C>G)	-
DNAJC21	NM_001012339.3	-	-
DNMT3B	NM_006892.4	chr20:g.31395557G>A (c.2421-11G>A)	-
EFL1	NM_024580.6	-	-
ELANE	NM_001972.4	-	-
EPO	NM_000799.4	-	-
ERCC4	NM_005236.3	-	-
ERCC6L2	NM_020207.5	-	-
ETV6	NM_001987.5	-	-
FANCA	NM_000135.4	chr16:g.89864654G>T (c.893+920C>A); chr16:g.89849346T>C (c.1567-20A>G); chr16:g.89836805T>C (c.2223-138A>G); chr16:g.89836111T>C (c.2504+134A>G); chr16:g.89831215G>C (c.2778+83C>G); chr16:g.89816056A>C (c.3239+82T>G); chr16:g.89805128_89805135del (c.4261-19_4261-12del)	-
FANCB	NM_001018113.3	-	-
FANCC	NM_000136.3	-	-
FANCD2	NM_033084.5	chr3:g.10083186C>G (c.696-121C>G); chr3:g.10102127T>G (c.1766+40T>G); chr3:g.10106024T>G (c.1948-16T>G)	CNV Analysis in exons 13, 14, and 17 is not performed
FANCE	NM_021922.3	-	-
FANCF	NM_022725.4	-	-
FANCG	NM_004629.1	-	-
FANCI	NM_001113378.1	-	-
FANCL	NM_018062.3	-	-
FANCM	NM_020937.4	-	CNV analysis in exon 8 is not performed
FLI1	NM_002017.5	-	-
FYB1	NM_001465.6	-	Sequence variants and CNV in exon 5 will not be performed, CNV analysis in exon 18 will not be performed
G6PC3	NM_138387.3	-	-
GATA1	NM_002049.4	-	-
GATA2	NM_032638.5	chr3:g.128202114 to chr3:g128202177 (c.1017+526 to c.1017+589 corresponding to a highly conserved intronic region); chr3:g.128200787_128200806dup (c.1018-17_1020dup)	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
GFI1	NM_005263.5	-	-
GFI1B	NM_004188.7	-	-
GINS1	NM_021067.5	chr20:g.25388397A>G (c.-60A>G); chr20:g.25388409C>G (c.-48C>G)	-
GLA	NM_000169.2	chrX:g.100653945A>T (c.640-11T>A); chrX:g.100654735C>T (c.640-801G>A); chrX:g.100653097C>T (c.1000-10G>A)	-
GLRX5	NM_016417.3	-	-
GP1BA	NM_000173.7	-	-
GP1BB	NM_000407.5	chr22:g.19710933C>G (c.-160C>G)	-
GP9	NM_000174.4	-	-
HAX1	NM_006118.4	-	-
HOXA11	NM_005523.6	-	-
IKZF1	NM_001291845.2; NM_006060.6	-	CNV analysis in exons 4–8 is not performed
ITGA2B	NM_000419.5	chr17:g.42463181G>T (c.408+11C>A); chr17:g.42458507T>C (c.1211-78A>G); chr17:g.g.42457859_42457871del (c.1440-13_1440-1del); chr17:g.42455177A>T (c.2095-19T>A); chr17:g.42449567A>G (c.*165T>C)	-
ITGB3	NM_000212.2	-	-
JAGN1	NM_032492.4	-	-
KDM1A	NM_001009999.3	-	-
KIF23	NM_138555.4	-	-
KLF1	NM_006563.5	chr19:g.12998108G>A (c.-154C>T)	-
LIG4	NM_002312.3	-	-
LPIN2	NM_014646.2	-	-
LYST	NM_000081.4	-	-
MAD2L2	NM_001127325.1	-	-
MDM4	NM_002393.5	-	-
MECOM	NM_001105078.4	-	-
MPIG6B	NM_025260.3	-	-
MPL	NM_005373.3	-	-
MTHFD1	NM_005956.4	-	-
MYH9	NM_002473.5	-	-
MYSM1	NM_001085487.3	-	-
NAF1	NM_138386.3	-	-
NBEAL2	NM_015175.3	-	CNV analysis in exon 1 is not performed
NBN	NM_002485.4	-	CNV analysis in exon 16 is not performed

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
NHP2	NM_017838.3	-	-
NOP10	NM_018648.3	-	-
OSTM1	NM_014028.4	-	-
PALB2	NM_024675.4	chr16:g.23649285A>T (c.109-12T>A); chr16:g.23625426_23625431del (c.3114-16_3114-11del)	-
PARN	NM_002582.4	-	CNV analysis in exons 6 and 19 is not performed
PAX5	NM_016734.3	-	-
PGM3	NM_001199917.2	-	-
PIK3CD	NM_005026.5	-	-
POT1	NM_015450.3	-	CNV analysis in exon 5 is not performed
PRKACG	NM_002732.3	-	-
PUS1	NM_025215.6	-	-
RAB27A	NM_004580.5	-	-
RAC2	NM_002872.5	-	-
RAD50	NM_005732.4	-	-
RAD51	NM_002875.5	-	-
RAD51C	NM_058216.3	-	-
RBM8A	NM_005105.4	chr1:g.145507648G>T (c.-19G>T); chr1:g.145507646G>A (c.-21G>A); chr1:g.145507765G>C (c.67+32G>C); chr1:g.145508462C>A (c.206-13C>A); chr1:g.145509217C>G (c.*6C>G)	-
RECQL4	NM_004260.3	chr8:g.145739527_145739550del (c.1878+32_1879-27del)	-
RFWD3	NM_018124.4	-	-
RMRP (NME1)	NR_003051.3	5' UTR variants from n.-25 through the transcription initiation site	CNV analysis is not performed
RPA1	NM_002945.5	-	-
RPL11	NM_000975.5	-	-
RPL15	NM_002948.4	-	CNV analysis in exon 4 is not performed
RPL18	NM_000979.4	-	-
RPL26	NM_000987.5	-	-
RPL27	NM_000988.5	-	-
RPL35	NM_007209.4	-	-
RPL35A	NM_000996.4	-	-
RPL5	NM_000969.5	-	-
RPS10	NM_001014.5	-	-
RPS15A	NM_001019.5	-	-

Targeted Genes and Methodology Details for Inherited Bone Marrow Failure Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
RPS17	NM_001021.6	Whole gene deletion and duplication performed	CNV analysis in individual exons 1–5 is not performed
RPS19	NM_001022.4	-	-
RPS24	NM_033022.4	-	-
RPS26	NM_001029.5	-	-
RPS27	NM_001030.6	-	-
RPS28	NM_001031.5	-	-
RPS29	NM_001032.4	-	-
RPS7	NM_001011.4	-	-
RTEL1	NM_032957.5	-	-
RUNX1	NM_001754.4	-	-
SAMD9	NM_017654.4	-	-
SAMD9L	NM_152703.5	-	-
SBDS	NM_016038.4	-	-
SEC23B	NM_006363.6	-	-
SLC19A2	NM_006996.3	-	-
SLC25A38	NM_017875.4	-	-
SLC37A4	NM_001164277.1	-	-
SLC46A1	NM_080669.6	-	-
SLFN14	NM_001129820.2	-	-
SLX4	NM_032444.4	-	-
SMARCAL1	NM_014140.4	-	-
SMARCD2	NM_001098426.2	-	-
SNX10	NM_001199835.1	-	-
SRC	NM_005417.4	-	-
SRP54	NM_003136.4	-	-
SRP72	NM_006947.4	-	CNV analysis in exon 17 is not performed
STAT3	NM_139276.2	chr17:g.40478306G>A (c.1282-89C>T)	-
STIM1	NM_003156.3	-	-
STN1	NM_024928.5	-	-
STXBP2	NM_006949.4	chr19:g.7705756_7705763del (c.326-30_326-23del); chr19: g.7705763_7705770del (c.326-23_326-16del)	-
SYK	NM_003177.7	-	-
TAZ (TAFAZZIN)	NM_000116.5	chrX:g.153641699G>A (c.284+110G>A)	-
TCIRG1	NM_006019.4	chr11:g.67816893 to chr11:g.67816910 (c.1887+132 to c.1887+149)	-
TCN2	NM_000355.4	chr22:g.31011112A>T (c.581-176A>T)	-

Targeted Genes and Methodology Details for Inherited Bone Marrow Failure Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
TERC	NR_001566.1	chr3:g.169482906G>A (n.-58C>G); chr3:169482870G>A (n.-22C>T)	CNV analysis is not performed
TERT	NM_198253.3	-	-
THPO	NM_000460.4	chr3:g.184094078C>A (c.-31G>T); chr3:g.184094097del (c.-47del)	-
TINF2	NM_001099274.3	-	-
TNFRSF11A	NM_003839.4	-	-
TNFRSF13B	NM_012452.3	-	-
TNFSF11	NM_003701.4	-	-
TP53	NM_000546.5	chr17:g.7579601G>C (c.97-11C>G)	-
TRNT1	NM_182916.3	-	-
TSR2	NM_058163.3	-	-
TUBB1	NM_030773.4	-	-
UBE2T	NM_014176.4	-	-
USB1	NM_024598.4	-	-
VPS13B	NM_017890.4	chr8:g.100479619T>G (c.3446-23T>G)	-
VPS45	NM_007259.5	-	-
VWF	NM_000552.4	-	-
WAS	NM_000377.3	chrX:g.48547690_48547698delinsATCTGCAGACC (c.1339-19_1339-11delinsATCTGCAGACC)	Sequence and CNV analysis is only reported for exon 28 (can detect majority of Von Willibrand disease-associated variants)
WDR1	NM_017491.5	-	-
WIPF1	NM_001077269.1	-	-
WRAP53	NM_018081.2	-	-
XIAP	NM_001167.3	-	-
XRCC2	NM_005431.2	-	-
YARS2	NM_001040436.3	-	-
ZCCHC8	NM_017612.5	-	-