MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Osteogenesis Imperfecta and Bone Fragility Gene Panel

The following applies to OIBFG / Osteogenesis Imperfecta and Bone Fragility 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 non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants 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 November 2022 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
ALPL	NM_000478.6	-	-
ANO5	NM_213599.2	-	CNV may not be detected in exon 2.
BMP1	NM_006129.5	-	-
BMP1	NM_001199.4	-	CNV and sequence variants will be analyzed and reported in exon 16 only.
COL1A1	NM_000088.3	chr17:48272201C>T (c.1354-12G>A)	-
COL1A2	NM_000089.4	-	CNV may not be detected in exons 2-3.
CREB3L1	NM_052854.4	-	-
CRTAP	NM_006371.5	chr3:33160815C>G (c.472-1021C>G)	-
FKBP10	NM_021939.4	-	-
IFITM5	NM_001025295.3	chr11:299504G>A (c.-14C>T)	-
LRP5	NM_002335.4	-	-
MBTPS2	NM_015884.4	-	-
P3H1	NM_022356.4	chr1:43212925C>T (c.2055+18G>A)	-
P4HB	NM_000918.4	-	-
PLOD2	NM_182943.3	-	-
PLS3	NM_005032.7	chrX:114856534T>A (c.74-24T>A)	-
PPIB	NM_000942.4	-	-
SEC24D	NM_014822.4	-	-
SERPINF1	NM_002615.7	-	-
SERPINH1	NM_001235.4	-	-
SP7	NM_001173467.3	-	-
SPARC	NM_003118.4	-	-
TAPT1	NM_153365.3	-	-
TMEM38B	NM_018112.3	-	-
WNT1	NM_005430.4	-	-
XYLT2	NM_022167.4	-	-