MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Ehlers-Danlos Syndrome Gene Panel

The following applies to EDSGG / Ehlers-Danlos Syndrome 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, or 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
ADAMTS2	NM_014244.5	-	-
AEBP1	NM_001129.5	-	-
ATP7A	NM_000052.7	-	-
B3GALT6	NM_080605.4	-	-
B3GAT3	NM_012200.4	-	-
B4GALT7	NM_007255.3	-	-
CHST14	NM_130468.3	-	-
COL12A1	NM_004370.6	-	-
COL1A1	NM_000088.3	chr17:48272201C>T (c.1354-12G>A)	-
COL1A2	NM_000089.4	-	CNV may not be detected in exons 2–3.
COL3A1	NM_000090.3	-	-
COL5A1	NM_000093.5	-	-
COL5A2	NM_000393.5	-	CNV may not be detected in exon 3.
DSE	NM_013352.4	-	-
FKBP14	NM_017946.4	-	-
FLNA	NM_001456.3	-	-
PLOD1	NM_000302.4	-	-
PRDM5	NM_018699.3	-	-
SLC39A13	NM_152264.4	-	-
SPARC	NM_003118.4	-	-
TNXB	NM_019105.8	-	CNV analysis of exons 17–44 will not be performed.
ZNF469	NM_001367624.2	-	-

Effective Date	Version	Synopsis of Test Change
04/25/2024	V2	Added lack of CNV detection for *TNXB* exons 17–44