MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Comprehensive Marfan, Loeys-Dietz, Ehlers-Danlos, and Aortopathy Gene Panel

The following applies to CAORG / Comprehensive Marfan, Loeys-Dietz, Ehlers-Danlos, and Aortopathy 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 (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 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
ACTA2	NM_001613.4	-	-
ADAMTS10	NM_030957.4	-	-
ADAMTS17	NM_139057.4	-	-
ADAMTS2	NM_014244.5	-	-
AEBP1	NM_001129.5	-	-
ATP7A	NM_000052.7	-	-
B3GALT6	NM_080605.4	-	-
B3GAT3	NM_012200.4	-	-
B4GALT7	NM_007255.3	-	-
BGN	NM_001711.6	-	-
CBS	NM_000071.2	-	-
CHST14	NM_130468.3	-	-
COL1A1	NM_000088.3	chr17:g.48273743A>T (c.904-15T>A); chr17:g.48273742C>T (c.904-14G>A); chr17:g.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	chr9:g.137686903T>G (c.2701-25T>G); chr9:g.137726806T>A (c.5137-11T>A)	-
COL5A2	NM_000393.5	-	CNV may not be detected in exon 3
COL12A1	NM_004370.6	-	-
DSE	NM_013352.4	-	-
EFEMP2	NM_016938.5	-	-
FBN1	NM_000138.4	chr15:g.48818478G>A (c.863-26C>T); chr15:g.48802380T>C (c.1589-14A>G); chr15:g.48780451A>T (c.3209-13T>A); chr15:g.48721629T>C (c.6872-961A>G); chr15:g.48720692A>T (c.6872-24T>A); chr15:g.48713895A>T (c.7571-12T>A); chr15:g.48707358C>A (c.8051+375G>T)	-

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Targeted Genes and Methodology Details for Comprehensive Marfan, Loeys-Dietz, Ehlers-Danlos, and Aortopathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
FBN2	NM_001999.4	chr5:g.127670562A>C (c.3974-26T>G); chr5:g.127671284T>C (c.3725-15A>G)	-
FKBP14	NM_017946.4	-	-
FLNA	NM_001456.3	-	-
LOX	NM_002317.7	-	-
LTBP3	NM_001130144.2	-	-
MED12	NM_005120.3	-	-
MFAP5	NM_003480.4	-	-
MYH11	NM_001040113.2	-	-
MYLK	NM_053025.4	-	-
NOTCH1	NM_017617.5	-	-
PLOD1	NM_000302.4	-	-
PRDM5	NM_018699.3	-	-
PRKG1	NM_006258.4	-	-
SKI	NM_003036.4	-	-
SLC2A10	NM_030777.4	-	-
SLC39A13	NM_152264.4	-	-
SMAD2	NM_005901.6	chr18:g.45396947T>C (c.237-12A>G)	-
SMAD3	NM_005902.4	-	-
SMAD4	NM_005359.6	-	-
SMAD6	NM_005585.5	-	-
SPARC	NM_003118.4	-	-
TGFB2	NM_003238.6	-	-
TGFB3	NM_003239.4	-	-
TGFBR1	NM_004612.4	-	-
TGFBR2	NM_003242.6	-	-
TNXB	NM_019105.8	-	CNV analysis of exons 32–44 will not be performed
ZNF469	NM_001367624.2	-	-

Effective Date	Version	Synopsis of Test Change
4/25/2024	V2	Added lack of CNV detection for *TNXB* exons 17–44
6/4/2025	V3	• Updated lack of CNV detection for *TNXB* to exons 32–44 • Added Additional Evaluations for *COL1A1*, *COL5A1*, *FBN1*, *FBN2*, and *SMAD2*

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