MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Muscular Dystrophy Gene Panel

The following applies to MDYSP / Inherited Muscular Dystrophy 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 December 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
ACTA1	NM_001100.4	-	-
ANO5	NM_213599.2	-	CNV in exon 2 may not be detected or reported
B3GALNT2	NM_152490.5	-	-
B4GAT1	NM_006876.3	-	-
BAG3	NM_004281.3	-	-
BIN1	NM_139343.3	-	-
BVES	NM_007073.4	-	-
CAPN3	NM_000070.3	c.380-13T>A c.2184+21G>A c.2185-16A>G	-
CAV3	NM_033337.3	-	-
CAVIN1	NM_012232.6	-	-
CHKB	NM_005198.4	-	-
COL12A1	NM_004370.6	-	-
COL6A1	NM_001848.3	c.930+189C>T	-
COL6A2	NM_001849.4	-	-
COL6A3	NM_004369.4	-	-
CRPPA	NM_001101426.4	-	-
CRYAB	NM_001885.3	-	-
DAG1	NM_004393.6	-	-
DES	NM_001927.4	-	-
DMD	NM_004006.2	c.-54T>A c.31+36947G>A c.3408+2036A>G c.9225-648A>G c.9225-647A>G c.9225-287C>A c.9225-285A>G	-
DNAJB6	NM_058246.4	-	-
DNM2	NM_001005360.2	-	-

Targeted Genes and Methodology Details for Inherited Muscular Dystrophy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
DPM1	NM_003859.2	-	-
DPM2	NM_003863.3	-	-
DPM3	NM_153741.2	-	-
DYSF	NM_003494.4	c.1054-43_1059delinsA	-
EMD	NM_000117.3	-	-
FHL1	NM_001449.5	-	-
FKRP	NM_024301.5	c.-272G>A c.-253+4A>G	-
FKTN	NM_001079802.1	c.648-1243G>T c.*4375_*4376ins3062	-
FLNC	NM_001458.4	-	-
GAA	NM_000152.5	c.-32-18 to c.-1 c.1076-22T>G c.2647-20T>G	-
GMPPB	NM_013334.3	-	-
GNE	NM_001128227.3	-	-
GOSR2	NM_004287.4	-	CNV in exon 2 may not be detected or reported
HNRNPA1	NM_031157.4	-	-
HNRNPA2B1	NM_031243.3	-	-
HNRNPDL	NM_031372.3	-	-
INPP5K	NM_016532.4	-	-
ITGA7	NM_002206.3	-	-
JAG2	NM_002226.5	-	Sequence variants in exon 1 may not be detected or reported, CNV in exon 1 will not be detected or reported
LAMA2	NM_000426.3	c.3556-13T>A	-
LARGE1	NM_004737.6	-	-
LDB3	NM_001080116.1	-	-
LMNA	NM_170707.4	c.1698+13C>A	-
MATR3	NM_199189.2	-	Duplication analysis for CNV will not be performed
MSTO1	NM_018116.3	-	Sequence variants and CNV in exons 1–7, 13 and 14 will not be detected or reported
MYH7	NM_000257.4	-	CNV in exon 27 may not be detected or reported
MYOT	NM_006790.3	-	-
PLEC	NM_000445.5	-	-
PLEC	NM_201378.4	-	-
POGLUT1	NM_152305.3	-	CNV in exon 10 may not be detected or reported

Targeted Genes and Methodology Details for Inherited Muscular Dystrophy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
POMGNT1	NM_017739.3	-	-
POMGNT2	NM_032806.6	-	-
POMK	NM_032237.5	-	-
POMT1	NM_007171.3	-	-
POMT2	NM_013382.5	c.1333-14G>A	-
RXYLT1	NM_014254.3	-	-
SELENON	NM_020451.3	c.-71 to c.-11	CNV in exon 3 may not be detected or reported
SGCA	NM_000023.4	-	-
SGCB	NM_000232.4	-	-
SGCD	NM_000337.5	-	-
SGCG	NM_000231.2	-	-
SQSTM1	NM_003900.5	-	-
SUN1	NM_001130965.3	-	-
SUN2	NM_015374.3	-	-
SYNE1	NM_033071.3	c.15705-12A>G	-
TCAP	NM_003673.4	-	-
TIA1	NM_022173.4	-	-
TMEM43	NM_024334.2	-	-
TNPO3	NM_012470.3	-	-
TOR1AIP1	NM_001267578.1	-	-
TRAPPC11	NM_021942.6	-	-
TRIM32	NM_012210.3	-	-
TTN	NM_133378.4	-	Sequence variants and CNV in exons 153–155 will not be detected or reported
VCP	NM_007126.5	-	-