MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Limb-Girdle Muscular Dystrophy and Congenital Myasthenic Syndrome Gene Panel

The following applies to LGCMP / Inherited Limb-Girdle Muscular Dystrophy and Congenital Myasthenic 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 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
AGRN	NM_198576.4		
ALG14	NM_144988.4		
ALG2	NM_033087.4		
ANO5	NM_213599.2	-	CNV in exon 2 may not be detected or reported
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		
CHAT	NM_020549.4		
CHRNA1	NM_000079.4		
CHRNB1	NM_000747.3		
CHRND	NM_000751.3		
CHRNE	NM_000080.4	c.501-16G>A c.-94G>A c.-95G>A c.-96C>T	-
COL13A1	NM_001130103.2	-	CNV in exons 3 and 33 may not be detected or reported
COL6A1	NM_001848.3	c.930+189C>T	-
COL6A2	NM_001849.4		
COL6A3	NM_004369.4		
COLQ	NM_005677.4		
CRPPA	NM_001101426.4		
DAG1	NM_004393.6		
DES	NM_001927.4		
DNAJB6	NM_058246.4		
DNM2	NM_001005360.2		
DOK7	NM_173660.5	c.54+25_55-38del	-
DPAGT1	NM_001382.4		
DPM3	NM_153741.2		
DYSF	NM_003494.4	c.1054-43_1059delinsA	-
FKRP	NM_024301.5	c.-272G>A c.-253+4A>G	-

Targeted Genes and Methodology Details for Inherited Limb-Girdle Muscular Dystrophy and Congenital Myasthenic Syndrome Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
FKTN	NM_001079802.1	c.648-1243G>T c.*4375_*4376ins3062	-
GAA	NM_000152.5	c.-32-18 to c.-11 c.1076-22T>G c.2647-20T>G	-
GFPT1	NM_001244710.1	-	CNV in exon 5 may not be detected or reported
GMPPB	NM_013334.3		
HNRNPDL	NM_031372.3		
LAMA2	NM_000426.3	c.3556-13T>A	-
LAMB2	NM_002292.4		
LMNA	NM_170707.4	c.1698+13C>A	-
LRP4	NM_002334.4		
MUSK	NM_005592.4		
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
POMGNT1	NM_017739.3		
POMGNT2	NM_032806.6		
POMK	NM_032237.5		
POMT1	NM_007171.3		
POMT2	NM_013382.5	c.1333-14G>A	-
PREPL	NM_006036.4		
RAPSN	NM_005055.5	c.-210A>G c.-199C>G	-
SCN4A	NM_000334.4		
SGCA	NM_000023.4		
SGCB	NM_000232.4		
SGCD	NM_000337.5		
SGCG	NM_000231.2		
SLC18A3	NM_003055.3		
SLC25A1	NM_005984.5		
SLC5A7	NM_021815.5		
SYT2	NM_177402.5		
TCAP	NM_003673.4		
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
VAMP1	NM_014231.5		
VCP	NM_007126.5		