



The following applies to MUPAN / Comprehensive Neuromuscular 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 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
<i>ABHD5</i>	NM_016006.6	-	-
<i>ACAD9</i>	NM_014049.5	-	-
<i>ACADM</i>	NM_000016.5	c.388-19T>A c.388-14A>G c.600-18G>A	-
<i>ACADS</i>	NM_000017.4	-	-
<i>ACADVL</i>	NM_000018.4	c.1183-15A>G	-
<i>ACTA1</i>	NM_001100.4	-	-
<i>ADSS1</i>	NM_199165.2	-	-
<i>AGK</i>	NM_018238.4	-	-
<i>AGL</i>	NM_000642.3	c.4260-12A>G	-
<i>AGRN</i>	NM_198576.4	-	-
<i>ALDOA</i>	NM_000034.3	-	-
<i>ALG14</i>	NM_144988.4	-	-
<i>ALG2</i>	NM_033087.4	-	-
<i>ANO5</i>	NM_213599.2	-	CNV in exon 2 may not be detected or reported
<i>ASAH1</i>	NM_177924.5	-	CNV in exon 4 may not be detected or reported
<i>ASCC1</i>	NM_001198800.3	-	-
<i>ATP1A2</i>	NM_000702.4	-	-
<i>ATP2A1</i>	NM_173201.4	-	-
<i>B3GALNT2</i>	NM_152490.5	-	-
<i>B4GAT1</i>	NM_006876.3	-	-
<i>BAG3</i>	NM_004281.3	-	-
<i>BIN1</i>	NM_139343.3	-	-
<i>BVES</i>	NM_007073.4	-	-
<i>CACNA1S</i>	NM_000069.3	-	-
<i>CAPN3</i>	NM_000070.3	c.380-13T>A c.2184+21G>A c.2185-16A>G	-
<i>CASQ1</i>	NM_001231.5	-	-

Targeted Genes and Methodology Details for Comprehensive Neuromuscular Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CAV3</i>	NM_033337.3	-	-
<i>CAVIN1</i>	NM_012232.6	-	-
<i>CFL2</i>	NM_021914.7	-	-
<i>CHAT</i>	NM_020549.4	-	-
<i>CHCHD10</i>	NM_213720.3	-	-
<i>CHKB</i>	NM_005198.4	-	-
<i>CHRNA1</i>	NM_000079.4	-	-
<i>CHRNA1</i>	NM_000079.4	-	-
<i>CHRNB1</i>	NM_000747.3	-	-
<i>CHRNA1</i>	NM_000079.4	-	-
<i>CHRNA1</i>	NM_000079.4	-	-
<i>CHRNA1</i>	NM_000079.4	c.501-16G>A c.-94G>A c.-95G>A c.-96C>T	-
<i>CLCN1</i>	NM_000083.3	-	-
<i>COL12A1</i>	NM_004370.6	-	-
<i>COL13A1</i>	NM_001130103.2	-	CNV in exons 3 and 33 may not be detected or reported
<i>COL6A1</i>	NM_001848.3	c.930+189C>T	-
<i>COL6A2</i>	NM_001849.4	-	-
<i>COL6A3</i>	NM_004369.4	-	-
<i>COLQ</i>	NM_005677.4	-	-
<i>COQ2</i>	NM_015697.8	-	-
<i>COQ4</i>	NM_016035.5	-	-
<i>COQ6</i>	NM_182476.3	-	-
<i>COQ8A</i>	NM_020247.5	-	-
<i>COQ9</i>	NM_020312.4	-	-
<i>CPT2</i>	NM_000098.3	-	-
<i>CRPPA</i>	NM_001101426.4	-	-
<i>CRYAB</i>	NM_001885.3	-	-
<i>CTDP1</i>	NM_004715.4	c.863+389C>T	-
<i>DAG1</i>	NM_004393.6	-	-
<i>DES</i>	NM_001927.4	-	-
<i>DGUOK</i>	NM_080916.3	c.444-62C>A c.444-11C>G	CNV in exon 13 may not be detected or reported
<i>DMD</i>	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	-

Targeted Genes and Methodology Details for Comprehensive Neuromuscular Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>DNA2</i>	NM_001080449.3	-	-
<i>DNAJB6</i>	NM_058246.4	-	-
<i>DNM2</i>	NM_001005360.2	-	-
<i>DOK7</i>	NM_173660.5	c.54+25_55-38del	-
<i>DPAGT1</i>	NM_001382.4	-	-
<i>DPM1</i>	NM_003859.2	-	-
<i>DPM2</i>	NM_003863.3	-	-
<i>DPM3</i>	NM_153741.2	-	-
<i>DYSF</i>	NM_003494.4	c.1054-43_1059delinsA	-
<i>EMD</i>	NM_000117.3	-	-
<i>ENO3</i>	NM_053013.4	-	-
<i>ETFA</i>	NM_000126.4	-	CNV in exon 12 may not be detected or reported
<i>ETFB</i>	NM_001985.3	-	-
<i>ETFDH</i>	NM_004453.4	c.-75A>G	-
<i>FAM111B</i>	NM_198947.4	-	-
<i>FBXL4</i>	NM_012160.4	-	-
<i>FDX2</i>	NM_001031734.4	-	-
<i>FHL1</i>	NM_001449.5	-	-
<i>FKBP14</i>	NM_017946.4	-	-
<i>FKRP</i>	NM_024301.5	c.-272G>A c.-253+4A>G	
<i>FKTN</i>	NM_001079802.1	c.648-1243G>T c.*4375_*4376ins3062	
<i>FLAD1</i>	NM_025207.5	-	-
<i>FLNC</i>	NM_001458.4	-	-
<i>GAA</i>	NM_000152.5	c.-32-18 to c.-11 c.1076-22T>G c.2647-20T>G	
<i>GBE1</i>	NM_000158.4	c.2053-3358_2053-3350delinsTGTTTTTACATGACAGGT	-
<i>GFER</i>	NM_005262.3	c.259-25_259-24del	-
<i>GFPT1</i>	NM_001244710.1	-	CNV in exon 5 may not be detected or reported
<i>GMPPB</i>	NM_013334.3	-	-
<i>GNE</i>	NM_001128227.3	-	-
<i>GOSR2</i>	NM_004287.4	-	CNV in exon 2 may not be detected or reported
<i>GYG1</i>	NM_004130.3	-	-
<i>GYS1</i>	NM_002103.5	-	-

Targeted Genes and Methodology Details for Comprehensive Neuromuscular Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>HADHA</i>	NM_000182.5	-	CNV in exon 14 may not be detected or reported
<i>HADHB</i>	NM_000183.3	-	-
<i>HNRNPA1</i>	NM_031157.4	-	-
<i>HNRNPA2B1</i>	NM_031243.3	-	-
<i>HNRNPDL</i>	NM_031372.3	-	-
<i>HRAS</i>	NM_005343.4	-	-
<i>INPP5K</i>	NM_016532.4	-	-
<i>ISCU</i>	NM_213595.3	c.343+382G>C	-
<i>ITGA7</i>	NM_002206.3	-	-
<i>JAG2</i>	NM_002226.5	-	Sequence variants in exon 1 may not be detected or reported, CNV in exon 1 will not be detected or reported
<i>KBTBD13</i>	NM_001101362.2	-	-
<i>KCNJ2</i>	NM_000891.3	-	-
<i>KLHL40</i>	NM_152393.4	-	-
<i>KLHL41</i>	NM_006063.3	-	-
<i>KY</i>	NM_178554.6	-	-
<i>LAMA2</i>	NM_000426.3	c.3556-13T>A	-
<i>LAMB2</i>	NM_002292.4	-	-
<i>LAMP2</i>	NM_013995.2	-	-
<i>LARGE1</i>	NM_004737.6	-	-
<i>LDB3</i>	NM_001080116.1	-	-
<i>LDHA</i>	NM_005566.4	-	-
<i>LMNA</i>	NM_170707.4	c.1698+13C>A	-
<i>LMOD3</i>	NM_198271.4	-	-
<i>LPIN1</i>	NM_145693.4	-	-
<i>LRP4</i>	NM_002334.4	-	-
<i>MAP3K20</i>	NM_016653.3	-	-
<i>MATR3</i>	NM_199189.2	-	Duplication analysis for CNV will not be performed
<i>MB</i>	NM_005368.3	-	-
<i>MEGF10</i>	NM_032446.3	-	-
<i>MGME1</i>	NM_052865.4	-	-
<i>MICU1</i>	NM_006077.3	-	-
<i>MRPS25</i>	NM_022497.5	-	-
<i>MSTO1</i>	NM_018116.3	-	Sequence variants and CNV in exons 1–7, 13 and 14 will not be detected or reported
<i>MTM1</i>	NM_000252.2	-	-

Targeted Genes and Methodology Details for Comprehensive Neuromuscular Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>MTO1</i>	NM_012123.4	-	-
<i>MUSK</i>	NM_005592.4	-	-
<i>MYBPC1</i>	NM_002465.4	-	-
<i>MYH2</i>	NM_017534.6	-	-
<i>MYH7</i>	NM_000257.4	-	CNV in exon 27 may not be detected or reported
<i>MYMK</i>	NM_001080483.3	-	-
<i>MYOT</i>	NM_006790.3	-	-
<i>MYPN</i>	NM_032578.3	-	-
<i>NEB</i>	NM_001271208.2	-	Sequence variants and CNV in exons 82–105 will not be detected or reported
<i>OPA1</i>	NM_015560.2	c.625-5459G>A	-
<i>ORAI1</i>	NM_032790.3	-	-
<i>PAX7</i>	NM_002584.3	-	-
<i>PDSS1</i>	NM_014317.5	-	CNV in exons 2–3 may not be detected or reported
<i>PDSS2</i>	NM_020381.4	-	CNV in exon 7 may not be detected or reported
<i>PFKM</i>	NM_000289.6	-	-
<i>PGAM2</i>	NM_000290.4	-	-
<i>PGK1</i>	NM_000291.4	-	-
<i>PGM1</i>	NM_002633.3	c.1145-222G>T c.1600-523G>A	-
<i>PHKA1</i>	NM_002637.4	-	-
<i>PLEC</i>	NM_000445.5	-	-
<i>PLEC</i>	NM_201378.4	-	-
<i>PNPLA2</i>	NM_020376.4	-	-
<i>PNPLA8</i>	NM_015723.5	-	-
<i>POGLUT1</i>	NM_152305.3	-	CNV in exon 10 may not be detected or reported
<i>POLG</i>	NM_002693.2	-	-
<i>POLG2</i>	NM_007215.4	-	-
<i>POMGNT1</i>	NM_017739.3	-	-
<i>POMGNT2</i>	NM_032806.6	-	-
<i>POMK</i>	NM_032237.5	-	-
<i>POMT1</i>	NM_007171.3	-	-
<i>POMT2</i>	NM_013382.5	c.1333-14G>A	-
<i>PREPL</i>	NM_006036.4	-	-
<i>PRKAG2</i>	NM_016203.4	-	CNV in exon 13 may not be detected or reported

Targeted Genes and Methodology Details for Comprehensive Neuromuscular Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>PUS1</i>	NM_025215.6	-	-
<i>PYGM</i>	NM_005609.4	c.425-26A>G	-
<i>PYROXD1</i>	NM_024854.5	-	-
<i>RAPSN</i>	NM_005055.5	c.-210A>G c.-199C>G	
<i>RBCK1</i>	NM_031229.4	-	-
<i>RNASEH1</i>	NM_002936.5	-	-
<i>RRM2B</i>	NM_015713.5	-	-
<i>RXYLT1</i>	NM_014254.3	-	-
<i>RYR1</i>	NM_000540.3	-	-
<i>SCN4A</i>	NM_000334.4	-	-
<i>SDHA</i>	NM_004168.4	-	-
<i>SELENON</i>	NM_020451.3	c.-71 to c.-11	CNV in exon 3 may not be detected or reported
<i>SGCA</i>	NM_000023.4	-	-
<i>SGCB</i>	NM_000232.4	-	-
<i>SGCD</i>	NM_000337.5	-	-
<i>SGCG</i>	NM_000231.2	-	-
<i>SIL1</i>	NM_022464.5	-	-
<i>SLC18A3</i>	NM_003055.3	-	-
<i>SLC22A5</i>	NM_003060.4	c.-149G>A c.394-16T>A	
<i>SLC25A1</i>	NM_005984.5	-	-
<i>SLC25A20</i>	NM_000387.6	-	-
<i>SLC25A3</i>	NM_005888.3	-	-
<i>SLC25A4</i>	NM_001151.4	-	-
<i>SLC25A42</i>	NM_178526.5	-	-
<i>SLC5A7</i>	NM_021815.5	-	-
<i>SMN1</i>	NM_022874.2	g.27134T>G (NG_008691.1) Provided only upon request	Analyzed for the presence of exon 7 CNV only; analysis for other sequence variants and CNV will not be performed
<i>SMN2</i>	NM_022876.2	-	Analyzed for the presence of exon 7 CNV only; analysis for other sequence variants and CNV will not be performed
<i>SPEG</i>	NM_005876.5	-	-
<i>SQSTM1</i>	NM_003900.5	-	-
<i>STAC3</i>	NM_145064.3	-	-
<i>STIM1</i>	NM_003156.3	-	-
<i>SUCLA2</i>	NM_003850.2	-	-
<i>SUCLG1</i>	NM_003849.4	-	-

Targeted Genes and Methodology Details for Comprehensive Neuromuscular Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>SUN1</i>	NM_001130965.3	-	-
<i>SUN2</i>	NM_015374.3	-	-
<i>SYNE1</i>	NM_033071.3	c.15705-12A>G	-
<i>SYT2</i>	NM_177402.5	-	-
<i>TANGO2</i>	NM_152906.7	-	-
<i>TAZ</i> (<i>TAFAZZIN</i>)	NM_000116.5	c.284+110G>A	-
<i>TBCD</i>	NM_005993.5	c.1564-12C>G	-
<i>TCAP</i>	NM_003673.4	-	-
<i>TFG</i>	NM_006070.6	-	-
<i>TIA1</i>	NM_022173.4	-	-
<i>TK2</i>	NM_004614.5	-	-
<i>TMEM43</i>	NM_024334.2	-	-
<i>TMEM65</i>	NM_194291.2	-	-
<i>TNNT1</i>	NM_003283.6	-	-
<i>TNPO3</i>	NM_012470.3	-	-
<i>TOR1AIP1</i>	NM_001267578.1	-	-
<i>TPM2</i>	NM_003289.3	-	-
<i>TPM3</i>	NM_152263.4	-	-
<i>TRAPPC11</i>	NM_021942.6	-	-
<i>TRIM32</i>	NM_012210.3	-	-
<i>TRIP4</i>	NM_016213.5	-	-
<i>TSFM</i>	NM_001172696.2	-	CNV in exon 5 may not be detected or reported
<i>TTC19</i>	NM_017775.4	-	-
<i>TTN</i>	NM_133378.4	-	Sequence variants and CNV in exons 153–155 will not be detected or reported
<i>TWINK</i>	NM_021830.5	-	-
<i>UBA1</i>	NM_003334.4	-	-
<i>VAMP1</i>	NM_014231.5	-	-
<i>VCP</i>	NM_007126.5	-	-
<i>VMA21</i>	NM_001017980.3	c.54-27A>C/T c.54-16_54-8del	
<i>YARS2</i>	NM_001040436.3	-	-