MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Rhabdomyolysis and Metabolic Myopathy Gene Panel

The following applies to RABMP / Inherited Rhabdomyolysis and Metabolic Myopathy 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
ABHD5	NM_016006.6	-	-
ACAD9	NM_014049.5	-	-
ACADM	NM_000016.5	c.388-19T>A c.388-14A>G c.600-18G>A	-
ACADS	NM_000017.4	-	-
ACADVL	NM_000018.4	c.1183-15A>G	-
AGK	NM_018238.4	-	-
AGL	NM_000642.3	c.4260-12A>G	-
ALDOA	NM_000034.3	-	-
ANO5	NM_213599.2	-	CNV in exon 2 may not be detected or reported
ATP2A1	NM_173201.4	-	-
CASQ1	NM_001231.5	-	-
CAVIN1	NM_012232.6	-	-
CHCHD10	NM_213720.3	-	-
COQ2	NM_015697.8	-	-
COQ4	NM_016035.5	-	-
COQ6	NM_182476.3	-	-
COQ8A	NM_020247.5	-	-
COQ9	NM_020312.4	-	-
CPT2	NM_000098.3	-	-
CTDP1	NM_004715.4	c.863+389C>T	-
DGUOK	NM_080916.3	c.444-62C>A c.444-11C>G	-
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	-

Targeted Genes and Methodology Details for Inherited Rhabdomyolysis and Metabolic Myopathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
DNA2	NM_001080449.3	-	-
DYSF	NM_003494.4	c.1054-43_1059delinsA	-
ENO3	NM_053013.4	-	-
ETFA	NM_000126.4	-	CNV in exon 12 may not be detected or reported
ETFB	NM_001985.3	-	-
ETFDH	NM_004453.4	c.-75A>G	-
FBXL4	NM_012160.4	-	-
FDX2	NM_001031734.4	-	-
FKRP	NM_024301.5	c.-272G>A c.-253+4A>G	
FKTN	NM_001079802.1	c.648-1243G>T c.*4375_*4376ins3062	
FLAD1	NM_025207.5	-	-
GAA	NM_000152.5	c.-32-18 to c.-11 c.1076-22T>G c.2647-20T>G	
GBE1	NM_000158.4	c.2053-3358_2053-3350delinsTGTTTTTTACATGACAGGT	-
GFER	NM_005262.3	c.259-25_259-24del	-
GYG1	NM_004130.3	-	-
GYS1	NM_002103.5	-	-
HADHA	NM_000182.5	-	CNV in exon 14 may not be detected or reported
HADHB	NM_000183.3	-	-
ISCU	NM_213595.3	c.343+382G>C	-
LAMP2	NM_013995.2	-	-
LDHA	NM_005566.4	-	-
LPIN1	NM_145693.4	-	-
MGME1	NM_052865.4	-	-
MRPS25	NM_022497.5	-	-
MSTO1	NM_018116.3	-	Sequence variants and CNV in exons 1–7, 13 and 14 will not be detected or reported
OPA1	NM_015560.2	c.625-5459G>A	-
PDSS1	NM_014317.5	-	CNV in exons 2–3 may not be detected or reported
PDSS2	NM_020381.4	-	CNV in exon 7 may not be detected or reported
PFKM	NM_000289.6	-	-
PGAM2	NM_000290.4	-	-
PGK1	NM_000291.4	-	-
PGM1	NM_002633.3	c.1145-222G>T c.1600-523G>A	

Targeted Genes and Methodology Details for Inherited Rhabdomyolysis and Metabolic Myopathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PHKA1	NM_002637.4	-	-
PNPLA2	NM_020376.4	-	-
PNPLA8	NM_015723.5	-	-
POLG	NM_002693.2	-	-
POLG2	NM_007215.4	-	-
PRKAG2	NM_016203.4	-	CNV in exon 13 may not be detected or reported
PUS1	NM_025215.6	-	-
PYGM	NM_005609.4	c.425-26A>G	-
RBCK1	NM_031229.4	-	-
RNASEH1	NM_002936.5	-	-
RRM2B	NM_015713.5	-	-
RYR1	NM_000540.3	-	-
SCN4A	NM_000334.4	-	-
SDHA	NM_004168.4	-	-
SLC22A5	NM_003060.4	c.-149G>A c.394-16T>A	
SLC25A20	NM_000387.6	-	-
SLC25A4	NM_001151.4	-	-
SLC25A42	NM_178526.5	-	-
SUCLA2	NM_003850.2	-	-
SUCLG1	NM_003849.4	-	-
TANGO2	NM_152906.7	-	-
TK2	NM_004614.5	-	-
TMEM65	NM_194291.2	-	-
TRIM32	NM_012210.3	-	-
TSFM	NM_001172696.2	-	CNV in exon 5 may not be detected or reported
TTC19	NM_017775.4	-	-
TWNK	NM_021830.5	-	-
VMA21	NM_001017980.3	c.54-27A>C/T c.54-16_54-8del	
YARS2	NM_001040436.3	-	-