Reference transcripts based on build GRCh37 (hg19) interrogated by Neuromuscular Genetic Panels

Motor Neuron Disease Panel	
Gene	**GenBank Accession Number**
ALS2	NM_020919
ANG	NM_001145
CHMP2B	NM_014043
ERBB4	NM_005235
FIG4	NM_014845
FUS	NM_004960
HNRNPA1	NM_031157
OPTN	NM_021980
PFN1	NM_005022
SETX	NM_015046
SIGMAR1	NM_005866
SOD1	NM_000454
SQSTM1	NM_003900
TARDBP	NM_007375
UBQLN2	NM_013444
VAPB	NM_004738
VCP	NM_007126

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

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Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Muscular Dystrophy Panel	
Gene	**GenBank Accession Number**
ACTA1	NM_001100
ANO5	NM_213599
B3GALNT2	NM_152490
B4GAT1	NM_006876
BAG3	NM_004281
BIN1	NM_139343
BVES	NM_007073
CAPN3	NM_000070
CAV3	NM_033337
CAVIN1	NM_012232
CHKB	NM_005198
COL12A1	NM_004370
COL6A1	NM_001848
COL6A2	NM_001849
COL6A3	NM_004369
CRYAB	NM_001885
DAG1	NM_004393
DES	NM_001927
DMD	NM_004006
DNAJB6	NM_058246
DNM2	NM_001005360
DOLK	NM_014908
DPM1	NM_003859
DPM2	NM_003863
DPM3	NM_153741
DYSF	NM_003494
EMD	NM_000117
FAM111B	NM_198947
FHL1	NM_001449
FKRP	NM_024301
FKTN	NM_001079802
FLNC	NM_001458
GGPS1	NM_001037277
GMPPA	NM_205847
GMPPB	NM_021971
GNE	NM_005476
GOSR2	NM_004287
HNRNPA1	NM_031157
HNRNPA2B1	NM_031243
HNRNPDL	NM_031372
ISPD	NM_001101426
ITGA7	NM_002206
LAMA2	NM_000426
LARGE1	NM_004737
LDB3	NM_001080116

Muscular Dystrophy Panel	
Gene	**GenBank Accession Number**
LMNA	NM_170707
LPIN1	NM_145693
MATR3	NM_199189
MYH2	NM_017534
MYH7	NM_000257
MYOT	NM_006790
NEB	NM_004543
PLEC	NM_000445
POMGNT1	NM_017739
POMGNT2	NM_032806
POMK	NM_032237
POMT1	NM_007171
POMT2	NM_013382
SELENON	NM_020451
SGCA	NM_000023
SGCB	NM_000232
SGCD	NM_000337
SGCG	NM_000231
SMCHD1	NM_015295
SQSTM1	NM_003900
SYNE1	NM_033071
TCAP	NM_003673
TIA1	NM_022173
TMEM43	NM_024334
TMEM5	NM_014254
TNPO3	NM_012470
TRAPPC11	NM_021942
TRIM32	NM_012210
TRIM54	NM_032546
TRIM63	NM_032588
TTN	NM_133378
VCP	NM_007126

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the gene TTN that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Additionally, NGS is used to test for the presence of large deletions and/or duplications in the ANO5, DMD, and LARGE1 genes.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Myofibrillar Myopathy Panel	
Gene	**GenBank Accession Number**
ACTA1	NM_001100
BAG3	NM_004281
CRYAB	NM_001885
DES	NM_001927
DNAJB6	NM_058246
FHL1	NM_001449
FLNC	NM_001458
LDB3	NM_001080116
LMNA	NM_170707
MYOT	NM_006790
SELENON	NM_020451
TTN	NM_133378

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the gene TTN that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Congenital Myopathy Panel	
Gene	**GenBank Accession Number**
ACTA1	NM_001100
ADGRG6	NM_198569
BIN1	NM_139343
CCDC78	NM_001031737
CFL2	NM_021914
CNTN1	NM_001843
COL12A1	NM_004370
COL6A1	NM_001848
COL6A2	NM_001849
COL6A3	NM_004369
DNM2	NM_001005360
HRAS	NM_005343
KBTBD13	NM_001101362
KLHL40	NM_152393
KLHL41	NM_006063
KY	NM_178554
MEGF10	NM_032446
MTM1	NM_000252
MYF6	NM_002469
MYH2	NM_017534
MYH7	NM_000257
MYO18B	NM_032608
NEB	NM_004543
ORAI1	NM_032790
RYR1	NM_000540
SBDS	NM_016038
SELENON	NM_020451
SPEG	NM_005876
SRPK3	NM_014370
STAC3	NM_145064
STIM1	NM_003156
TNNT1	NM_003283
TPM2	NM_003289
TPM3	NM_152263
TRDN	NM_006073
TTN	NM_133378

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the gene TTN that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Congenital Myasthenic Syndromes	
Gene	**GenBank Accession Number**
AGRN	NM_198576
ALG14	NM_144988
ALG2	NM_033087
BIN1	NM_139343
CHAT	NM_020549
CHRNA1	NM_001039523
CHRNB1	NM_000747
CHRND	NM_000751
CHRNE	NM_000080
COLQ	NM_005677
DNM2	NM_001005360
DOK7	NM_173660
DPAGT1	NM_001382
GAA	NM_000152
GFPT1	NM_002056
GMPPB	NM_021971
LAMB2	NM_002292
LRP4	NM_002334
MUSK	NM_005592
PLEC	NM_000445
PREPL	NM_006036
RAPSN	NM_005055
SCN4A	NM_000334
SNAP25	NM_003081 and NM_130811
SYT2	NM_177402

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

Regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence. Therefore, all reported alterations detected by NGS are confirmed by an independent reference method based on laboratory developed criteria. However, this does not rule out the possibility of a false-negative result in these regions.

Additionally, NGS is used to test for the presence of large deletions and/or duplications in the COLQ and RAPSN genes.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Metabolic Myopathy Panel	
Gene	**GenBank Accession Number**
ABHD5	NM_016006
ACAD9	NM_014049
ACADL	NM_001608
ACADM	NM_000016
ACADS	NM_000017
ACADVL	NM_000018
AGL	NM_000642
COQ2	NM_015697
COQ4	NM_016035
COQ6	NM_182476
COQ8A	NM_020247
COQ9	NM_020312
CPT1B	NM_004377
CPT2	NM_000098
ENO3	NM_053013
ETFA	NM_000126
ETFB	NM_001985
ETFDH	NM_004453
GBE1	NM_000158
GYG1	NM_004130
GYS1	NM_002103
HADHA	NM_000182
HADHB	NM_000183
LAMP2	NM_002294
LDHA	NM_005566
LPIN1	NM_145693
NHLRC1	NM_198586
PDSS1	NM_014317
PDSS2	NM_020381
PFKM	NM_000289
PGAM2	NM_000290
PGK1	NM_000291
PGM1	NM_002633
PHKA1	NM_002637
PNPLA2	NM_020376
PRKAG2	NM_016203
PYGM	NM_005609
RBCK1	NM_031229
SLC22A5	NM_003060
SLC25A20	NM_000387
VMA21	NM_001017980

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

Regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence. Therefore, all reported alterations detected by NGS are confirmed by an independent reference method based on laboratory developed criteria. However, this does not rule out the possibility of a false-negative result in these regions.

Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Emery-Dreifuss Panel	
Gene	**GenBank Accession Number**
EMD	NM_000117
FHL1	NM_001449
LMNA	NM_170707
MYOT	NM_006790
SYNE1	NM_033071

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

Regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence. Therefore, all reported alterations detected by NGS are confirmed by an independent reference method based on laboratory developed criteria. However, this does not rule out the possibility of a false-negative result in these regions.

Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Distal Myopathy Panel	
Gene	**GenBank Accession Number**
ACTA1	NM_001100
ANO5	NM_213599
BAG3	NM_004281
BIN1	NM_139343
CAV3	NM_033337
CRYAB	NM_001885
DES	NM_001927
DNAJB6	NM_058246
DNM2	NM_001005360
DYSF	NM_003494
FHL1	NM_001449
FLNC	NM_001458
GNE	NM_005476
HNRNPA1	NM_031157
HNRNPA2B1	NM_031243
LDB3	NM_001080116
LMNA	NM_170707
MATR3	NM_199189
MYH2	NM_017534
MYH7	NM_000257
MYOT	NM_006790
NEB	NM_004543
SELENON	NM_020451
SQSTM1	NM_003900
TIA1	NM_022173
TTN	NM_133378
VCP	NM_007126

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the gene TTN that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Additionally, NGS is used to test for the presence of large deletions and/or duplications in the ANO5 gene.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Skeletal Muscle Channelopathy Panel	
Gene	**GenBank Accession Number**
CACNA1S	NM_000069
CLCN1	NM_000083
KCNE3	NM_005472
KCNJ18	NM_001194958
KCNJ2	NM_000891
SCN4A	NM_000334

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

Regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence. Therefore, all reported alterations detected by NGS are confirmed by an independent reference method based on laboratory developed criteria. However, this does not rule out the possibility of a false-negative result in these regions.

Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Myopathy Expanded Panel	
Gene	**GenBank Accession Number**
ABHD5	NM_016006
ACAD9	NM_014049
ACADL	NM_001608
ACADM	NM_000016
ACADS	NM_000017
ACADVL	NM_000018
ACTA1	NM_001100
ADGRG6	NM_198569
AGL	NM_000642
ANO5	NM_213599
B3GALNT2	NM_152490
B4GAT1	NM_006876
BAG3	NM_004281
BIN1	NM_139343
BVES	NM_007073
CAPN3	NM_000070
CAV3	NM_033337
CAVIN1	NM_012232
CCDC78	NM_001031737
CFL2	NM_021914
CHKB	NM_005198
CNTN1	NM_001843
COL12A1	NM_004370
COL6A1	NM_001848
COL6A2	NM_001849
COL6A3	NM_004369
COQ2	NM_015697
COQ4	NM_016035
COQ6	NM_182476
COQ8A	NM_020247
COQ9	NM_020312
CPT1B	NM_004377
CPT2	NM_000098
CRYAB	NM_001885
DAG1	NM_004393
DES	NM_001927
DMD	NM_004006
DNAJB6	NM_058246
DNM2	NM_001005360
DOLK	NM_014908
DPM1	NM_003859
DPM2	NM_003863
DPM3	NM_153741
DYSF	NM_003494
EMD	NM_000117

Myopathy Expanded Panel	
Gene	**GenBank Accession Number**
ENO3	NM_053013
ETFA	NM_000126
ETFB	NM_001985
ETFDH	NM_004453
FAM111B	NM_198947
FHL1	NM_001449
FKRP	NM_024301
FKTN	NM_001079802
FLNC	NM_001458
GBE1	NM_000158
GGPS1	NM_001037277
GMPPA	NM_205847
GMPPB	NM_021971
GNE	NM_005476
GOSR2	NM_004287
GYG1	NM_004130
GYS1	NM_002103
HADHA	NM_000182
HADHB	NM_000183
HNRNPA1	NM_031157
HNRNPA2B1	NM_031243
HNRNPDL	NM_031372
HRAS	NM_005343
ISPD	NM_001101426
ITGA7	NM_002206
KBTBD13	NM_001101362
KLHL40	NM_152393
KLHL41	NM_006063
KY	NM_178554
LAMA2	NM_000426
LAMP2	NM_002294
LARGE1	NM_004737
LDB3	NM_001080116
LDHA	NM_005566
LMNA	NM_170707
LPIN1	NM_145693
MATR3	NM_199189
MEGF10	NM_032446
MTM1	NM_000252
MYF6	NM_002469
MYH2	NM_017534
MYH7	NM_000257
MYO18B	NM_032608
MYOT	NM_006790

Myopathy Expanded Panel	
Gene	**GenBank Accession Number**
NEB	NM_004543
NHLRC1	NM_198586
ORAI1	NM_032790
PDSS1	NM_014317
PDSS2	NM_020381
PFKM	NM_000289
PGAM2	NM_000290
PGK1	NM_000291
PGM1	NM_002633
PHKA1	NM_002637
PLEC	NM_000445
PNPLA2	NM_020376
POMGNT1	NM_017739
POMGNT2	NM_032806
POMK	NM_032237
POMT1	NM_007171
POMT2	NM_013382
PRKAG2	NM_016203
PYGM	NM_005609
RBCK1	NM_031229
RYR1	NM_000540
SBDS	NM_016038
SELENON	NM_020451
SGCA	NM_000023
SGCB	NM_000232
SGCD	NM_000337
SGCG	NM_000231
SLC22A5	NM_003060
SLC25A20	NM_000387
SMCHD1	NM_015295
SPEG	NM_005876
SQSTM1	NM_003900
SRPK3	NM_014370
STAC3	NM_145064
STIM1	NM_003156
SYNE1	NM_033071
TCAP	NM_003673
TIA1	NM_022173
TMEM43	NM_024334
TMEM5	NM_014254

Myopathy Expanded Panel	
Gene	**GenBank Accession Number**
TNNT1	NM_003283
TNPO3	NM_012470
TPM2	NM_003289
TPM3	NM_152263
TRAPPC11	NM_021942
TRDN	NM_006073
TRIM32	NM_012210
TRIM54	NM_032546
TRIM63	NM_032588
TTN	NM_133378
VCP	NM_007126
VMA21	NM_001017980

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Additionally, NGS is used to test for the presence of large deletions and/or duplications in the ANO5, DMD, and LARGE1 genes.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Distal Weakness Expanded Panel	
Gene	**GenBank Accession Number**
AAAS	NM_015665
AARS	NM_001605
ABCA1	NM_005502
ABCD1	NM_000033
ACTA1	NM_001100
ADCY6	NM_015270
AIFM1	NM_004208
AMACR	NM_014324
ANO5	NM_213599
AP1S1	NM_001283
AP4B1	NM_006594
AP4E1	NM_007347
AP4M1	NM_004722
AP4S1	NM_007077
AP5Z1	NM_014855
APOA1	NM_000039
APTX	NM_175073
ARHGEF10	NM_014629
ARSA	NM_000487
ATL1	NM_015915
ATM	NM_000051
ATP7A	NM_000052
B2M	NM_004048
B4GALNT1	NM_001478
BAG3	NM_004281
BCKDHB	NM_183050
BICD2	NM_001003800
BIN1	NM_139343
BSCL2	NM_032667
C12orf65	NM_152269
CAV3	NM_033337
CCT5	NM_012073
CLCF1	NM_013246
CNTNAP1	NM_003632
COX10	NM_001303
CPOX	NM_000097
CRLF1	NM_004750
CRYAB	NM_001885
CTDP1	NM_004715
CTSA	NM_000308
CYP27A1	NM_000784

Distal Weakness Expanded Panel	
Gene	**GenBank Accession Number**
CYP2U1	NM_183075
CYP7B1	NM_004820
DARS2	NM_018122
DCAF8	NM_015726
DCTN1	NM_004082
DDHD1	NM_001160147
DDHD2	NM_015214
DES	NM_001927
DGUOK	NM_080916
DHH	NM_021044
DHTKD1	NM_018706
DNAJB2	NM_001039550
DNAJB6	NM_058246
DNM2	NM_001005360
DNMT1	NM_001130823
DST	NM_015548 and NM_001723
DYNC1H1	NM_001376
DYSF	NM_003494
EGR2	NM_000399
ERBB3	NM_001982
ERCC6	NM_000124
ERCC8	NM_000082
ERLIN2	NM_007175
FA2H	NM_024306
FAH	NM_000137
FAM126A	NM_032581
FAM134B	NM_001034850
FBLN5	NM_006329
FBXO38	NM_030793
FGD4	NM_139241
FGF14	NM_004115
FHL1	NM_001449
FIG4	NM_014845
FLNC	NM_001458
FLVCR1	NM_014053
FMR1	NM_002024
GALC	NM_000153
GAN	NM_022041
GARS	NM_002047
GBA2	NM_020944
GBE1	NM_000158

Distal Weakness Expanded Panel	
Gene	**GenBank Accession Number**
GDAP1	NM_018972
GJB1	NM_000166
GJB3	NM_024009
GJC2	NM_020435
GLA	NM_000169
GNB4	NM_021629
GNE	NM_005476
GSN	NM_000177
HADHA	NM_000182
HADHB	NM_000183
HARS	NM_002109
HINT1	NM_005340
HK1	NM_000188
HMBS	NM_000190
HNRNPA1	NM_031157
HNRNPA2B1	NM_031243
HSPB1	NM_001540
HSPB3	NM_006308
HSPB8	NM_014365
HSPD1	NM_002156
IGHMBP2	NM_002180
IKBKAP	NM_003640
INF2	NM_022489
KARS	NM_001130089
KIF1A	NM_004321
KIF1B	NM_015074
KIF5A	NM_004984
L1CAM	NM_000425
LAMA2	NM_000426
LDB3	NM_001080116
LITAF	NM_004862
LMNA	NM_170707
LRSAM1	NM_138361
LYST	NM_000081
MAF	NM_005360
MARS	NM_004990
MATR3	NM_199189
MED25	NM_030973
MFN2	NM_014874
MMACHC	NM_015506
MPV17	NM_002437

Distal Weakness Expanded Panel	
Gene	**GenBank Accession Number**
MPZ	NM_000530
MTMR2	NM_016156
MTTP	NM_000253
MYH14	NM_024729
MYH2	NM_017534
MYH7	NM_000257
MYOT	NM_006790
NAGA	NM_000262
NAGLU	NM_000263
NDRG1	NM_006096
NEB	NM_004543
NEFL	NM_006158
NF2	NM_000268
NGF	NM_002506
NIPA1	NM_144599
NTRK1	NM_002529
OAT	NM_000274
OPA1	NM_015560
PANK2	NM_153638
PDHA1	NM_000284
PDK3	NM_001142386
PDYN	NM_024411
PEX10	NM_153818
PEX7	NM_000288
PHYH	NM_006214
PLA2G6	NM_003560
PLEKHG5	NM_198681
PLOD1	NM_000302
PLP1	NM_000533
PMM2	NM_000303
PMP2	NM_002677
PMP22	NM_000304
PNKP	NM_007254
PNPLA6	NM_006702
POLG	NM_002693
PPOX	NM_000309
PRNP	NM_000311
PRPS1	NM_002764
PRX	NM_181882
RAB7A	NM_004637
REEP1	NM_022912

Distal Weakness Expanded Panel	
Gene	**GenBank Accession Number**
RRM2B	NM_015713
RTN2	NM_005619
SACS	NM_014363
SBF1	NM_002972
SBF2	NM_030962
SCN10A	NM_006514
SCN11A	NM_014139
SCN9A	NM_002977
SCO2	NM_005138
SCP2	NM_002979
SELENON	NM_020451
SETX	NM_015046
SH3TC2	NM_024577
SLC12A6	NM_133647
SLC16A2	NM_006517
SLC25A19	NM_021734
SLC25A46	NM_138773
SLC33A1	NM_004733
SLC52A2	NM_024531
SLC5A7	NM_021815
SNAP29	NM_004782
SOD1	NM_000454
SOX10	NM_006941
SPAST	NM_014946
SPG11	NM_025137
SPG20	NM_015087
SPG21	NM_016630
SPG7	NM_003119
SPTLC1	NM_006415
SPTLC2	NM_004863
SQSTM1	NM_003900
SURF1	NM_003172
TDP1	NM_018319
TECPR2	NM_014844
TFG	NM_006070
TIA1	NM_022173
TRIM2	NM_015271
TRPA1	NM_007332
TRPV4	NM_021625
TTN	NM_133378

Distal Weakness Expanded Panel	
Gene	**GenBank Accession Number**
TTPA	NM_000370
TTR	NM_000371
TUBB3	NM_006086
TWNK	NM_021830
TYMP	NM_001953
VCP	NM_007126
VPS37A	NM_152415
WASHC5	NM_014846
WNK1	NM_018979 and NM_213655
XPA	NM_000380
XPC	NM_004628
YARS	NM_003680
ZFYVE26	NM_015346

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the genes CRLF1, DNMT1, GJC2, INF2, MAF, PNKP, and TTN that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Additionally, NGS is used to test for the presence of large deletions and/or duplications in the AN05, GDAP1, GLA, MFN2, MPZ, MTTP, PMP22, PNKP, POLG, and SPG7 genes.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)

Rhabdomyolysis and Myopathy Panel	
Gene	**GenBank Accession Number**
ACAD9	NM_014049
ACADL	NM_001608
ACADM	NM_000016
ACADVL	NM_000018
AGL	NM_000642
ANO5	NM_213599
CPT2	NM_000098
CTDP1	NM_004715
DGUOK	NM_080916
DMD	NM_004006
DYSF	NM_003494
ENO3	NM_053013
FKRP	NM_024301
FKTN	NM_001079802
GAA	NM_000152
GYS1	NM_002103
HADHA	NM_000182
HADHB	NM_000183
LPIN1	NM_145693
OPA1	NM_015560
PFKM	NM_000289
PGAM2	NM_000290
PGK1	NM_000291
PGM1	NM_002633
PHKA1	NM_002637
POLG	NM_002693
PYGM	NM_005609
RRM2B	NM_015713
RYR1	NM_000540
SLC22A5	NM_003060
TWNK	NM_021830

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

Regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence. Therefore, all reported alterations detected by NGS are confirmed by an independent reference method based on laboratory developed criteria. However, this does not rule out the possibility of a false-negative result in these regions.

Additionally, NGS is used to test for the presence of large deletions and/or duplications in the ANO5, DMD, and POLG genes.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate.(Unpublished Mayo method)