MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for the Nuclear Mitochondrial Disorders Custom Gene Panel

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed. NGS and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]
AARS2	NM_020745.4
ABAT	NM_020686.6
ABCB7	NM_004299.6
ACACA	NM_198839.2
ACAD9	NM_014049.5
ACO2	NM_001098.3
AFG3L2	NM_006796.3
AGK	NM_018238.4
AIFM1	NM_004208.4
ALDH3A2	NM_000382.3
APOPT1 (COA8)	NM_032374.4
APTX	NM_175073.2
ATP5F1A	NM_001001937.1
ATP5F1E	NM_006886.4
ATPAF2	NM_145691.4
AUH	NM_001698.2
BCS1L	NM_004328.5
BOLA3	NM_212552.3
C12orf65 (MTRFR)	NM_152269.5
CA5A	NM_001739.2
CARS2	NM_024537.4
CHAT	NM_020549.4
CHCHD10	NM_213720.3
CLPP	NM_006012.4
COA5	NM_001008215.3
COA6	NM_001012985.2
COA8 (APOPT1)	NM_032374.4
COASY	NM_025233.7
COQ2	NM_015697.8
COQ4	NM_016035.5
COQ6	NM_182476.3
COQ7	NM_016138.5
COQ8A	NM_020247.5
COQ8B	NM_024876.4
COQ9	NM_020312.4

Gene	Reference Transcript[a]
COX10[b]	NM_001303.4
COX14	NM_032901.4
COX15[b]	NM_004376.7
COX20	NM_198076.6
COX4I1	NM_001861.6
COX4I2	NM_032609.3
COX6A1	NM_004373.4
COX6A2	NM_005205.4
COX6B1	NM_001863.5
COX7B	NM_001866.3
COX8A	NM_004074.3
CPT1C[b]	NM_001136052.2
CYC1	NM_001916.5
D2HGDH	NM_152783.5
DARS2	NM_018122.5
DGUOK	NM_080916.3
DLAT	NM_001931.5
DLD	NM_000108.5
DNA2	NM_001080449.3
DNAJC19	NM_145261.4
DNM1L	NM_012062.5
EARS2	NM_001083614.2
ELAC2	NM_018127.7
ETFA[b]	NM_000126.4
ETFB	NM_001985.3
ETFDH	NM_004453.4
ETHE1	NM_014297.5
FARS2	NM_006567.5
FASTKD2	NM_014929.3
FBXL4	NM_012160.4
FDX2	NM_001031734.4
FDXR	NM_024417.5
FH	NM_000143.3
FOXRED1	NM_017547.4
FXN	NM_000144.5

Targeted Genes and Methodology Details for the Nuclear Mitochondrial Disorders Custom Gene Panel (continued)

Gene	Reference Transcript[a]
GAMT	NM_000156.6
GARS1	NM_002047.4
GCDH	NM_000159.4
GDAP1	NM_018972.4
GFER	NM_005262.3
GFM1	NM_024996.5
GFM2[b]	NM_032380.5
GLYCTK	NM_145262.4
GPT2	NM_133443.4
GTPBP3	NM_133644.4
HARS2	NM_012208.4
HIBCH	NM_014362.4
HK1	NM_000188.2
HSPD1[b]	NM_002156.5
IARS2	NM_018060.4
IBA57	NM_001010867.4
IDH2	NM_002168.3
INF2	NM_022489.4
ISCU	NM_213595.3
L2HGDH[b]	NM_024884.3
LARS2	NM_015340.4
LIAS	NM_006859.4
LRPPRC[b]	NM_133259.4
LYRM4	NM_020408.5
LYRM7[b]	NM_181705.4
MARS2	NM_138395.4
MFF[b]	NM_020194.5
MGME1	NM_052865.4
MICU1	NM_006077.3
MPC1[b]	NM_016098.4
MPV17	NM_002437.5
MRPL3	NM_007208.4
MRPL44	NM_022915.5
MRPS16	NM_016065.4
MRPS2	NM_016034.5
MRPS22	NM_020191.4
MRPS7	NM_015971.4
MSTO1[b]	NM_018116.3
MTFMT	NM_139242.4
MTO1	NM_012123.4
MTPAP	NM_018109.3

Gene	Reference Transcript[a]
MTRFR (C12orf65)	NM_152269.5
NARS2[b]	NM_024678.6
NBAS	NM_015909.4
NDUFA1	NM_004541.4
NDUFA10	NM_004544.4
NDUFA11	NM_175614.5
NDUFA12	NM_018838.5
NDUFA13	NM_015965.7
NDUFA2	NM_002488.4
NDUFA4	NM_002489.4
NDUFA9	NM_005002.5
NDUFAF1	NM_016013.4
NDUFAF2	NM_174889.5
NDUFAF3	NM_199069.2
NDUFAF4	NM_014165.4
NDUFAF5	NM_024120.5
NDUFAF6	NM_152416.4
NDUFB3	NM_002491.3
NDUFB9	NM_005005.3
NDUFS1	NM_005006.7
NDUFS2	NM_004550.4
NDUFS3	NM_004551.3
NDUFS4	NM_002495.4
NDUFS6	NM_004553.5
NDUFS7	NM_024407.5
NDUFS8	NM_002496.4
NDUFV1	NM_007103.4
NDUFV2	NM_021074.5
NFU1	NM_001002755.3
NR2F1[b]	NM_005654.6
NUBPL	NM_025152.3
OGDH	NM_002541.4
OPA1	NM_015560.2
OPA3	NM_025136.4
OXCT1[b]	NM_000436.4
PANK2	NM_153638.3
PARS2	NM_152268.4
PC	NM_000920.4
PCK2	NM_004563.4
PDHA1	NM_000284.4
PDHB	NM_000925.4

Targeted Genes and Methodology Details for the Nuclear Mitochondrial Disorders Custom Gene Panel (continued)

Gene	Reference Transcript[a]
PDHX	NM_003477.3
PDP1	NM_018444.4
PDSS1[b]	NM_014317.5
PDSS2[b]	NM_020381.4
PET100	NM_001171155.2
PNKD	NM_015488.5
PNPT1	NM_033109.5
POLG	NM_002693.2
POLG2[b]	NM_007215.4
PTRH2	NM_016077.4
PUS1	NM_025215.6
QARS1	NM_005051.3
RARS1	NM_002887.4
RARS2	NM_020320.5
RMND1	NM_017909.4
RNASEH1	NM_002936.5
RRM2B	NM_015713.5
RTN4IP1	NM_032730.5
SACS	NM_014363.6
SARS2	NM_017827.4
SCO1	NM_004589.4
SCO2	NM_005138.2
SDHAF1	NM_001042631.2
SERAC1[b]	NM_032861.4
SFXN4	NM_213649.2
SLC19A3	NM_025243.4
SLC25A1	NM_005984.5
SLC25A12	NM_003705.5
SLC25A19	NM_021734.4
SLC25A22	NM_024698.6
SLC25A26	NM_173471.3
SLC25A3	NM_005888.3
SLC25A4	NM_001151.4
SLC25A42	NM_178526.5
SLC25A46	NM_138773.4
SLC52A2	NM_024531.5

Gene	Reference Transcript[a]
SLC9A6	NM_006359.3
SOD1	NM_000454.4
SPG7	NM_003119.4
SUCLA2	NM_003850.2
SUCLG1	NM_003849.4
SUGCT[b]	NM_024728.2
SURF1	NM_003172.4
TACO1	NM_016360.4
TAFAZZIN (TAZ)	NM_000116.5
TARS2	NM_025150.5
TAZ (TAFAZZIN)	NM_000116.5
TFAM	NM_003201.3
TIMM8A	NM_004085.4
TK2	NM_004614.5
TMEM126A	NM_032273.4
TMEM126B	NM_018480.5
TMEM70	NM_017866.6
TOP3A	NM_004618.5
TPK1	NM_022445.4
TRIT1	NM_017646.6
TRMT10C	NM_017819.4
TRMU	NM_018006.5
TRNT1	NM_182916.3
TSFM[b]	NM_001172696.2
TTC19	NM_017775.4
TUFM	NM_003321.5
TWNK	NM_021830.5
TYMP	NM_001953.5
UQCC2	NM_032340.4
UQCRB	NM_006294.4
UQCRC2	NM_003366.4
UQCRQ	NM_014402.5
VARS2	NM_001167734.1
WDR45	NM_007075.3
XPNPEP3	NM_022098.4
YARS2	NM_001040436.3

[a] Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

[b] There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.

Targeted Genes and Methodology Details for the Nuclear Mitochondrial Disorders Custom Gene Panel (continued)

Available Nuclear Mitochondrial Disorders Panels

Test ID	Test Name	Genes
NMITO	Nuclear Mitochondrial Gene Panel by Next-Generation Sequencing (NGS)	*AARS2, ABAT, ABCB7, ACACA, ACAD9, ACO2, AFG3L2, AGK, AIFM1, ALDH3A2, APOPT1 (COA8), APTX, ATP5F1A, ATP5F1E, ATPAF2, AUH, BCS1L, BOLA3, C12orf65 (MTRFR), CA5A, CARS2, CHAT, CHCHD10, CLPP, COA5, COA6, COA8 (APOPT1), COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX6A1, COX6A2, COX6B1, COX7B, COX8A, CPT1C, CYC1, D2HGDH, DARS2, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FDXR, FH, FOXRED1, FXN, GAMT, GARS1, GCDH, GDAP1, GFER, GFM1, GFM2, GLYCTK, GPT2, GTPBP3, HARS2, HIBCH, HK1, HSPD1, IARS2, IBA57, IDH2, INF2, ISCU, L2HGDH, LARS2, LIAS, LRPPRC, LYRM4, LYRM7, MARS2, MFF, MGME1, MICU1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS7, MSTO1, MTFMT, MTO1, MTPAP, MTRFR (C12orf65), NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NR2F1, NUBPL, OGDH, OPA1, OPA3, OXCT1, PANK2, PARS2, PC, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PNKD, PNPT1, POLG, POLG2, PTRH2, PUS1, QARS1, RARS1, RARS2, RMND1, RNASEH1, RRM2B, RTN4IP1, SACS, SARS2, SCO1, SCO2, SDHAF1, SERAC1, SFXN4, SLC19A3, SLC25A1, SLC25A12, SLC25A19, SLC25A22, SLC25A26, SLC25A3, SLC25A4, SLC25A42, SLC25A46, SLC52A2, SLC9A6, SOD1, SPG7, SUCLA2, SUCLG1, SUGCT, SURF1, TACO1, TAFAZZIN (TAZ), TARS2, TAZ (TAFAZZIN), TFAM, TIMM8A, TK2, TMEM126A, TMEM126B, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCRB, UQCRC2, UQCRQ, VARS2, WDR45, XPNPEP3, YARS2*
CMITO	Combined Mitochondrial Analysis, Mitochondrial Full Genome and Nuclear Gene Panel	*AARS2, ABAT, ABCB7, ACACA, ACAD9, ACO2, AFG3L2, AGK, AIFM1, ALDH3A2, APOPT1 (COA8), APTX, ATP5F1A, ATP5F1E, ATPAF2, AUH, BCS1L, BOLA3, C12orf65 (MTRFR), CA5A, CARS2, CHAT, CHCHD10, CLPP, COA5, COA6, COA8 (APOPT1), COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX6A1, COX6A2, COX6B1, COX7B, COX8A, CPT1C, CYC1, D2HGDH, DARS2, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FDXR, FH, FOXRED1, FXN, GAMT, GARS1, GCDH, GDAP1, GFER, GFM1, GFM2, GLYCTK, GPT2, GTPBP3, HARS2, HIBCH, HK1, HSPD1, IARS2, IBA57, IDH2, INF2, ISCU, L2HGDH, LARS2, LIAS, LRPPRC, LYRM4, LYRM7, MARS2, MFF, MGME1, MICU1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS7, MSTO1, MTFMT, MTO1, MTPAP, MTRFR (C12orf65), NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NR2F1, NUBPL, OGDH, OPA1, OPA3, OXCT1, PANK2, PARS2, PC, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PNKD, PNPT1, POLG, POLG2, PTRH2, PUS1, QARS1, RARS1, RARS2, RMND1, RNASEH1, RRM2B, RTN4IP1, SACS, SARS2, SCO1, SCO2, SDHAF1, SERAC1, SFXN4, SLC19A3, SLC25A1, SLC25A12, SLC25A19, SLC25A22, SLC25A26, SLC25A3, SLC25A4, SLC25A42, SLC25A46, SLC52A2, SLC9A6, SOD1, SPG7, SUCLA2, SUCLG1, SUGCT, SURF1, TACO1, TAFAZZIN (TAZ), TARS2, TAZ (TAFAZZIN), TFAM, TIMM8A, TK2, TMEM126A, TMEM126B, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCRB, UQCRC2, UQCRQ, VARS2, WDR45, XPNPEP3, YARS2 and mitochondrial genome*