

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

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

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

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

Gene	Reference Transcript ^a
<i>SLC9A6</i>	NM_006359.3
<i>SOD1</i>	NM_000454.4
<i>SPG7</i>	NM_003119.4
<i>SUCLA2</i>	NM_003850.2
<i>SUCLG1</i>	NM_003849.4
<i>SUGCT^b</i>	NM_024728.2
<i>SURF1</i>	NM_003172.4
<i>TACO1</i>	NM_016360.4
<i>TAFAZZIN (TAZ)</i>	NM_000116.5
<i>TARS2</i>	NM_025150.5
<i>TAZ (TAFAZZIN)</i>	NM_000116.5
<i>TFAM</i>	NM_003201.3
<i>TIMM8A</i>	NM_004085.4
<i>TK2</i>	NM_004614.5
<i>TMEM126A</i>	NM_032273.4
<i>TMEM126B</i>	NM_018480.5
<i>TMEM70</i>	NM_017866.6
<i>TOP3A</i>	NM_004618.5
<i>TPK1</i>	NM_022445.4
<i>TRIT1</i>	NM_017646.6
<i>TRMT10C</i>	NM_017819.4
<i>TRMU</i>	NM_018006.5
<i>TRNT1</i>	NM_182916.3
<i>TSFM^b</i>	NM_001172696.2
<i>TTC19</i>	NM_017775.4
<i>TUFM</i>	NM_003321.5
<i>TWNK</i>	NM_021830.5
<i>TYMP</i>	NM_001953.5
<i>UQCC2</i>	NM_032340.4
<i>UQCRB</i>	NM_006294.4
<i>UQCRC2</i>	NM_003366.4
<i>UQCRQ</i>	NM_014402.5
<i>VAR2</i>	NM_001167734.1
<i>WDR45</i>	NM_007075.3
<i>XPNPEP3</i>	NM_022098.4
<i>YARS2</i>	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, DNMI1, 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, UQCRCQ, 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, DNMI1, 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, UQCRCQ, VARS2, WDR45, XPNPEP3, YARS2 and mitochondrial genome