

## Targeted Genes and Methodology Details for Combined Mitochondrial Full Genome and Nuclear Gene Panel, Varies

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 <sup>a</sup>
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 <sup>a</sup>
COX10 <sup>b</sup>	NM_001303.4
COX14	NM_032901.4
COX15 <sup>b</sup>	NM_004376.7
COX20	NM_198076.6
COX4I1	NM_001861.6
COX412	NM_032609.3
COX6A1	NM_004373.4
COX6A2	NM_005205.4
COX6B1	NM_001863.5
COX7B	NM_001866.3
COX8A	NM_004074.3
CPT1C <sup>b</sup>	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 <sup>b</sup>	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 Combined Mitochondrial Full Genome and Nuclear Gene Panel, Varies (continued)

Gene	Reference Transcript <sup>a</sup>
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 <sup>b</sup>	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 <sup>b</sup>	NM_002156.5
IARS2	NM_018060.4
IBA57	NM_001010867.4
IDH2	NM_002168.3
INF2	NM_022489.4
ISCU	NM_213595.3
L2HGDH <sup>b</sup>	NM_024884.3
LARS2	NM_015340.4
LIAS	NM_006859.4
LRPPRC <sup>b</sup>	NM_133259.4
LYRM4	NM_020408.5
LYRM7 <sup>⁵</sup>	NM_181705.4
MARS2	NM_138395.4
MFF <sup>b</sup>	NM_020194.5
MGME1	NM_052865.4
MICU1	NM_006077.3
MPC1 <sup>b</sup>	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
MSTO1⁵	NM_018116.3
MTFMT	NM_139242.4
MTO1	NM_012123.4
MTPAP	NM_018109.3

MTRFR (C120rf65)         NM_152269.5           NARS2°         NM_024678.6           NBAS         NM_015909.4           NDUFA1         NM_004541.4           NDUFA10         NM_004544.4           NDUFA11         NM_175614.5           NDUFA12         NM_01838.5           NDUFA13         NM_015965.7           NDUFA2         NM_002488.4           NDUFA4         NM_002489.4           NDUFA4         NM_005002.5           NDUFA5         NM_016013.4           NDUFAF         NM_174889.5           NDUFAFA         NM_174889.5           NDUFAFA         NM_199069.2           NDUFAFA         NM_199069.2           NDUFAFA         NM_014165.4           NDUFAFA         NM_024120.5           NDUFAFA         NM_024120.5           NDUFAFA         NM_024120.5           NDUFAFA         NM_024120.5           NDUFAFA         NM_024120.5           NDUFAFA         NM_002491.3           NDUFAFA         NM_002491.3           NDUFAFA         NM_005006.7           NDUFSB         NM_004550.4           NDUFSB         NM_004551.3           NDUFSA         NM_004553.5      <	Gene	Reference Transcript <sup>a</sup>
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           NDUFA2         NM_002489.4           NDUFA9         NM_005002.5           NDUFA9         NM_016013.4           NDUFA9         NM_174889.5           NDUFA1         NM_199069.2           NDUFA2         NM_174889.5           NDUFA3         NM_199069.2           NDUFA4         NM_014165.4           NDUFA5         NM_024120.5           NDUFA6         NM_152416.4           NDUFA6         NM_152416.4           NDUFA8         NM_005006.7           NDUFS3         NM_005006.7           NDUFS4         NM_004550.4           NDUFS3         NM_004551.3           NDUFS4         NM_004553.5           NDUFS6         NM_004553.5           NDUFS8         NM_002496.4           NDUFS8         NM_0021074.5           NPU1         NM_007103.4           NPUFY1         NM_0075054.6           NUBPL	MTRFR (C12orf65)	NM_152269.5
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_05002.5           NDUFAF1         NM_016013.4           NDUFAF2         NM_174889.5           NDUFAF3         NM_199069.2           NDUFAF4         NM_014165.4           NDUFAF5         NM_024120.5           NDUFAF6         NM_152416.4           NDUFAB3         NM_002491.3           NDUFB3         NM_005006.7           NDUFS3         NM_004550.4           NDUFS3         NM_004550.4           NDUFS3         NM_004551.3           NDUFS4         NM_004553.5           NDUFS5         NM_004553.5           NDUFS6         NM_004553.5           NDUFS8         NM_002496.4           NDUFS8         NM_0021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_005654.6           NUBPL         NM_005664.6           <	NARS2 <sup>b</sup>	NM_024678.6
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           NDUFAB3         NM_002491.3           NDUFB3         NM_005006.7           NDUFS3         NM_004550.4           NDUFS4         NM_004550.4           NDUFS3         NM_004551.3           NDUFS4         NM_002495.4           NDUFS6         NM_004553.5           NDUFS7         NM_024407.5           NDUFS8         NM_002496.4           NDUFS8         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_005664.6           NUBPL         NM_005664.6           NUBPL         NM_005664.6 <t< td=""><td>NBAS</td><td>NM_015909.4</td></t<>	NBAS	NM_015909.4
NDUFA11         NM_175614.5           NDUFA12         NM_018838.5           NDUFA13         NM_015965.7           NDUFA2         NM_002488.4           NDUFA4         NM_005002.5           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           NDUFB3         NM_005006.7           NDUFB9         NM_005006.7           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_001002755.3           NR2F1b         NM_001002755.3           NR2F1b         NM_00250654.6           NUBPL         NM_0025136.4           OPA1         NM_005660.2	NDUFA1	NM_004541.4
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_005006.7           NDUFS1         NM_005006.7           NDUFS2         NM_004550.4           NDUFS3         NM_004550.4           NDUFS4         NM_002495.4           NDUFS6         NM_004553.5           NDUFS7         NM_024407.5           NDUFS8         NM_002496.4           NDUFV1         NM_001002755.3           NR2F1b         NM_001002755.3           NR2F1b         NM_002541.4           OPA1         NM_002541.4           OPA1         NM_002541.4           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PARS2         NM_153638.3	NDUFA10	NM_004544.4
NDUFA13         NM_015965.7           NDUFA2         NM_002488.4           NDUFA4         NM_002025.5           NDUFA9         NM_016013.4           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_004553.5           NDUFS5         NM_004553.5           NDUFS6         NM_002496.4           NDUFS7         NM_002496.4           NDUFV1         NM_007103.4           NDUFV2         NM_001002755.3           NR2F1 <sup>b</sup> NM_001002755.3           NR2F1 <sup>b</sup> NM_005654.6           NUBPL         NM_005654.6           NUBPL         NM_005664.4           NDUF3         NM_004563.4           PANK2         NM_004563.4           PARS2         NM_15268.4	NDUFA11	NM_175614.5
NDUFA2	NDUFA12	NM_018838.5
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           NDUFS5         NM_024407.5           NDUFS6         NM_002496.4           NDUFS8         NM_002496.4           NDUFV1         NM_007103.4           NDUFV2         NM_001002755.3           NR2F1 <sup>0</sup> NM_001002755.3           NR2F1 <sup>0</sup> NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_004563.4           PANK2         NM_00436.4           PANK2         NM_153638.3           PARS2         NM_004563.4           PDHA1         NM_004563.4           P	NDUFA13	NM_015965.7
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_005006.7           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_021074.5           NPU1         NM_001002755.3           NR2F1 <sup>b</sup> NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_00563.4           NPARS2         NM_153638.3           PARS2         NM_15268.4           PC         NM_004563.4           PDHA1         NM_004563.4           NDUFS2	NDUFA2	NM_002488.4
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           NDUFS8         NM_0021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_005654.6           NUBPL         NM_005664.6           NUBPL <td>NDUFA4</td> <td>NM_002489.4</td>	NDUFA4	NM_002489.4
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           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_025156.2           OPA1         NM_025136.4           OYCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_15268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFA9	NM_005002.5
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           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_0025136.4           OYA1         NM_00436.4           PANK2         NM_153638.3           PARS2         NM_15268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFAF1	NM_016013.4
NDUFAF4         NM_014165.4           NDUFAF5         NM_024120.5           NDUFAF6         NM_152416.4           NDUFB3         NM_002491.3           NDUFB9         NM_005006.7           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 <sup>b</sup> NM_005654.6           NUBPL         NM_005654.6           NUBPL         NM_0025152.3           OGDH         NM_002541.4           OPA1         NM_00560.2           OPA3         NM_0025136.4           OXCT1 <sup>b</sup> NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFAF2	NM_174889.5
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           NDUFS5         NM_024407.5           NDUFS8         NM_002496.4           NDUFS8         NM_007103.4           NDUFV2         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFAF3	NM_199069.2
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_001002755.3           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_15268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFAF4	NM_014165.4
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           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_025136.4           OPA1         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFAF5	NM_024120.5
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           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_025136.4           OPA1         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFAF6	NM_152416.4
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           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFB3	NM_002491.3
NDUFS2         NM_004550.4           NDUFS3         NM_004551.3           NDUFS6         NM_002495.4           NDUFS7         NM_024407.5           NDUFS8         NM_002496.4           NDUFV1         NM_007103.4           NDUFV2         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_0004563.4           PDHA1         NM_000284.4	NDUFB9	NM_005005.3
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           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFS1	NM_005006.7
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           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_015560.2           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFS2	NM_004550.4
NDUFS6         NM_004553.5           NDUFS7         NM_0024407.5           NDUFS8         NM_002496.4           NDUFV1         NM_007103.4           NDUFV2         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFS3	NM_004551.3
NDUFS7         NM_024407.5           NDUFS8         NM_002496.4           NDUFV1         NM_007103.4           NDUFV2         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFS4	NM_002495.4
NDUFS8         NM_002496.4           NDUFV1         NM_007103.4           NDUFV2         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFS6	NM_004553.5
NDUFV1         NM_007103.4           NDUFV2         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFS7	NM_024407.5
NDUFV2         NM_021074.5           NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFS8	NM_002496.4
NFU1         NM_001002755.3           NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFV1	NM_007103.4
NR2F1b         NM_005654.6           NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NDUFV2	NM_021074.5
NUBPL         NM_025152.3           OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NFU1	NM_001002755.3
OGDH         NM_002541.4           OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NR2F1 <sup>b</sup>	NM_005654.6
OPA1         NM_015560.2           OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	NUBPL	NM_025152.3
OPA3         NM_025136.4           OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	OGDH	NM_002541.4
OXCT1b         NM_000436.4           PANK2         NM_153638.3           PARS2         NM_152268.4           PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	OPA1	NM_015560.2
PANK2 NM_153638.3  PARS2 NM_152268.4  PC NM_000920.4  PCK2 NM_004563.4  PDHA1 NM_000284.4	OPA3	NM_025136.4
PARS2 NM_152268.4  PC NM_000920.4  PCK2 NM_004563.4  PDHA1 NM_000284.4	OXCT1 <sup>b</sup>	NM_000436.4
PC         NM_000920.4           PCK2         NM_004563.4           PDHA1         NM_000284.4	PANK2	NM_153638.3
PCK2 NM_004563.4 PDHA1 NM_000284.4	PARS2	NM_152268.4
PDHA1 NM_000284.4	PC	NM_000920.4
	PCK2	NM_004563.4
PDHB NM_000925.4	PDHA1	NM_000284.4
	PDHB	NM_000925.4

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## Targeted Genes and Methodology Details for Combined Mitochondrial Full Genome and Nuclear Gene Panel, Varies (continued)

Gene	Reference Transcript <sup>a</sup>
PDHX	NM_003477.3
PDP1	NM_018444.4
PDSS1 <sup>b</sup>	NM_014317.5
PDSS2 <sup>b</sup>	NM_020381.4
PET100	NM_001171155.2
PNKD	NM_015488.5
PNPT1	NM_033109.5
POLG	NM_002693.2
POLG2 <sup>b</sup>	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 <sup>b</sup>	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 <sup>a</sup>
SLC9A6	NM_006359.3
SOD1	NM 000454.4
SPG7	NM_003119.4
SUCLA2	NM_003850.2
SUCLG1	NM 003849.4
SUGCT <sup>b</sup>	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 <sup>b</sup>	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

<sup>&</sup>lt;sup>a</sup> 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.

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

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<sup>&</sup>lt;sup>b</sup> There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.