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]	Gene	Reference Transcript[a]
AARS2	NM_020745.4	COX10[b]	NM_001303.4
ABAT	NM_020686.6	COX14	NM_032901.4
ABCB7	NM_004299.6	COX15[b]	NM_004376.7
ACACA	NM_198839.2	COX20	NM_198076.6
ACAD9	NM_014049.5	COX4I1	NM_001861.6
ACO2	NM_001098.3	COX4I2	NM_032609.3
AFG3L2	NM_006796.3	COX6A1	NM_004373.4
AGK	NM_018238.4	COX6A2	NM_005205.4
AIFM1	NM_004208.4	COX6B1	NM_001863.5
ALDH3A2	NM_000382.3	COX7B	NM_001866.3
APOPT1 (COA8)	NM_032374.4	COX8A	NM_004074.3
APTX	NM_175073.2	CPT1C[b]	NM_001136052.2
ATP5F1A	NM_001001937.1	CYC1	NM_001916.5
ATP5F1E	NM_006886.4	D2HGDH	NM_152783.5
ATPAF2	NM_145691.4	DARS2	NM_018122.5
AUH	NM_001698.2	DGUOK	NM_080916.3
BCS1L	NM_004328.5	DLAT	NM_001931.5
BOLA3	NM_212552.3	DLD	NM_000108.5
C12orf65 (MTRFR)	NM_152269.5	DNA2	NM_001080449.3
CA5A	NM_001739.2	DNAJC19	NM_145261.4
CARS2	NM_024537.4	DNM1L	NM_012062.5
CHAT	NM_020549.4	EARS2	NM_001083614.2
CHCHD10	NM_213720.3	ELAC2	NM_018127.7
CLPP	NM_006012.4	ETFA[b]	NM_000126.4
COA5	NM_001008215.3	ETFB	NM_001985.3
COA6	NM_001012985.2	ETFDH	NM_004453.4
COA8 (APOPT1)	NM_032374.4	ETHE1	NM_014297.5
COASY	NM_025233.7	FARS2	NM_006567.5
COQ2	NM_015697.8	FASTKD2	NM_014929.3
COQ4	NM_016035.5	FBXL4	NM_012160.4
COQ6	NM_182476.3	FDX2	NM_001031734.4
COQ7	NM_016138.5	FDXR	NM_024417.5
COQ8A	NM_020247.5	FH	NM_000143.3
COQ8B	NM_024876.4	FOXRED1	NM_017547.4
COQ9	NM_020312.4	FXN	NM_000144.5

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

Gene	Reference Transcript[a]	Gene	Reference Transcript[a]
GAMT	NM_000156.6	MTRFR (C12orf65)	NM_152269.5
GARS1	NM_002047.4	NARS2[b]	NM_024678.6
GCDH	NM_000159.4	NBAS	NM_015909.4
GDAP1	NM_018972.4	NDUFA1	NM_004541.4
GFER	NM_005262.3	NDUFA10	NM_004544.4
GFM1	NM_024996.5	NDUFA11	NM_175614.5
GFM2[b]	NM_032380.5	NDUFA12	NM_018838.5
GLYCTK	NM_145262.4	NDUFA13	NM_015965.7
GPT2	NM_133443.4	NDUFA2	NM_002488.4
GTPBP3	NM_133644.4	NDUFA4	NM_002489.4
HARS2	NM_012208.4	NDUFA9	NM_005002.5
HIBCH	NM_014362.4	NDUFAF1	NM_016013.4
HK1	NM_000188.2	NDUFAF2	NM_174889.5
HSPD1[b]	NM_002156.5	NDUFAF3	NM_199069.2
IARS2	NM_018060.4	NDUFAF4	NM_014165.4
IBA57	NM_001010867.4	NDUFAF5	NM_024120.5
IDH2	NM_002168.3	NDUFAF6	NM_152416.4
INF2	NM_022489.4	NDUFB3	NM_002491.3
ISCU	NM_213595.3	NDUFB9	NM_005005.3
L2HGDH[b]	NM_024884.3	NDUFS1	NM_005006.7
LARS2	NM_015340.4	NDUFS2	NM_004550.4
LIAS	NM_006859.4	NDUFS3	NM_004551.3
LRPPRC[b]	NM_133259.4	NDUFS4	NM_002495.4
LYRM4	NM_020408.5	NDUFS6	NM_004553.5
LYRM7[b]	NM_181705.4	NDUFS7	NM_024407.5
MARS2	NM_138395.4	NDUFS8	NM_002496.4
MFF[b]	NM_020194.5	NDUFV1	NM_007103.4
MGME1	NM_052865.4	NDUFV2	NM_021074.5
MICU1	NM_006077.3	NFU1	NM_001002755.3
MPC1[b]	NM_016098.4	NR2F1[b]	NM_005654.6
MPV17	NM_002437.5	NUBPL	NM_025152.3
MRPL3	NM_007208.4	OGDH	NM_002541.4
MRPL44	NM_022915.5	OPA1	NM_015560.2
MRPS16	NM_016065.4	OPA3	NM_025136.4
MRPS2	NM_016034.5	OXCT1[b]	NM_000436.4
MRPS22	NM_020191.4	PANK2	NM_153638.3
MRPS7	NM_015971.4	PARS2	NM_152268.4
MSTO1[b]	NM_018116.3	PC	NM_000920.4
MTFMT	NM_139242.4	PCK2	NM_004563.4
MTO1	NM_012123.4	PDHA1	NM_000284.4
MTPAP	NM_018109.3	PDHB	NM_000925.4

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

Gene	Reference Transcript[a]	Gene	Reference Transcript[a]
PDHX	NM_003477.3	SLC9A6	NM_006359.3
PDP1	NM_018444.4	SOD1	NM_000454.4
PDSS1[b]	NM_014317.5	SPG7	NM_003119.4
PDSS2[b]	NM_020381.4	SUCLA2	NM_003850.2
PET100	NM_001171155.2	SUCLG1	NM_003849.4
PNKD	NM_015488.5	SUGCT[b]	NM_024728.2
PNPT1	NM_033109.5	SURF1	NM_003172.4
POLG	NM_002693.2	TACO1	NM_016360.4
POLG2[b]	NM_007215.4	TAFAZZIN (TAZ)	NM_000116.5
PTRH2	NM_016077.4	TARS2	NM_025150.5
PUS1	NM_025215.6	TAZ (TAFAZZIN)	NM_000116.5
QARS1	NM_005051.3	TFAM	NM_003201.3
RARS1	NM_002887.4	TIMM8A	NM_004085.4
RARS2	NM_020320.5	TK2	NM_004614.5
RMND1	NM_017909.4	TMEM126A	NM_032273.4
RNASEH1	NM_002936.5	TMEM126B	NM_018480.5
RRM2B	NM_015713.5	TMEM70	NM_017866.6
RTN4IP1	NM_032730.5	TOP3A	NM_004618.5
SACS	NM_014363.6	TPK1	NM_022445.4
SARS2	NM_017827.4	TRIT1	NM_017646.6
SCO1	NM_004589.4	TRMT10C	NM_017819.4
SCO2	NM_005138.2	TRMU	NM_018006.5
SDHAF1	NM_001042631.2	TRNT1	NM_182916.3
SERAC1[b]	NM_032861.4	TSFM[b]	NM_001172696.2
SFXN4	NM_213649.2	TTC19	NM_017775.4
SLC19A3	NM_025243.4	TUFM	NM_003321.5
SLC25A1	NM_005984.5	TWNK	NM_021830.5
SLC25A12	NM_003705.5	TYMP	NM_001953.5
SLC25A19	NM_021734.4	UQCC2	NM_032340.4
SLC25A22	NM_024698.6	UQCRB	NM_006294.4
SLC25A26	NM_173471.3	UQCRC2	NM_003366.4
SLC25A3	NM_005888.3	UQCRQ	NM_014402.5
SLC25A4	NM_001151.4	VARS2	NM_001167734.1
SLC25A42	NM_178526.5	WDR45	NM_007075.3
SLC25A46	NM_138773.4	XPNPEP3	NM_022098.4
SLC52A2	NM_024531.5	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.