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]
ACAA2	NM_006111.3	FLAD1	NM_025207.5
ACACA	NM_198839.2	GLUD1	NM_005271.5
ACAD8	NM_014384.2	HADH	NM_005327.5
ACAD9	NM_014049.5	HADHA[b]	NM_000182.5
ACADL	NM_001608.4	HADHB	NM_000183.3
ACADM	NM_000016.5	HMGCL	NM_000191.3
ACADS	NM_000017.4	HMGCS2	NM_005518.4
ACADSB	NM_001609.4	HSD17B10	NM_004493.3
ACADVL	NM_000018.4	LPIN1	NM_145693.4
ACAT1	NM_000019.4	MLYCD	NM_012213.3
ACAT2	NM_005891.3	NADK2	NM_001085411.3
ACOT9	NM_001037171.2	OPA1	NM_015560.2
ALDH5A1	NM_001080.3	PPARG	NM_015869.4
CPT1A	NM_001876.4	SLC22A5	NM_003060.4
CPT2	NM_000098.3	SLC25A20	NM_000387.6
DECR1	NM_001359.2	SLC25A29	NM_001039355.3
ECHS1	NM_004092.4	SLC25A32	NM_030780.5
ECI1	NM_001919.4	SLC52A1	NM_001104577.1
ETFA[b]	NM_000126.4	SLC52A2	NM_024531.5
ETFB	NM_001985.3	SLC52A3	NM_033409.4
ETFDH	NM_004453.4	TANGO2	NM_152906.7
ETHE1	NM_014297.5	TAZ	NM_000116.5

[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.