MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Congenital Lactic Acidosis 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]
ACAD9	NM_014049.5
AGK	NM_018238.4
DLD	NM_000108.5
ECHS1	NM_004092.4
FBXL4	NM_012160.4
FLAD1	NM_025207.5
FOXRED1	NM_017547.4
GFER	NM_005262.3
HADHA[b]	NM_000182.5
HADHB	NM_000183.3
HLCS	NM_000411.8
MRPL3	NM_007208.4
MRPS22	NM_020191.4
NDUFB11	NM_019056.6

Gene	Reference Transcript[a]
NDUFS4	NM_002495.4
OGDH	NM_002541.4
PC	NM_000920.4
PDHA1	NM_000284.4
PDHX	NM_003477.3
PDP1	NM_018444.4
SLC19A2	NM_006996.3
SLC19A3	NM_025243.4
SLC25A19	NM_021734.4
SUCLG1	NM_003849.4
TMEM70	NM_017866.6
TPK1	NM_022445.4
UQCRC2	NM_003366.4
VARS2	NM_001167734.1

[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.