The following applies to CLADP / Congenital Lactic Acidosis Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 30 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from February 2021 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest to providers as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]	Gene	Reference Transcript[a]
ACAD9	NM_014049.5	NDUFS4	NM_002495.4
AGK	NM_018238.4	OGDH	NM_002541.4
DLD	NM_000108.5	PC	NM_000920.4
ECHS1	NM_004092.4	PDHA1	NM_000284.4
FBXL4	NM_012160.4	PDHX	NM_003477.3
FLAD1	NM_025207.5	PDP1	NM_018444.4
FOXRED1	NM_017547.4	SLC19A2	NM_006996.3
GFER	NM_005262.3	SLC19A3	NM_025243.4
HADHA[b]	NM_000182.5	SLC25A19	NM_021734.4
HADHB	NM_000183.3	SUCLG1	NM_003849.4
HLCS	NM_000411.8	TMEM70	NM_017866.6
MRPL3	NM_007208.4	TPK1	NM_022445.4
MRPS22	NM_020191.4	UQCRC2	NM_003366.4
NDUFB11	NM_019056.6	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.