

Targeted Genes and Methodology Details for Glutaric Aciduria Type II Gene Panel

The following applies to GA2P / Glutaric Aciduria Type II Gene 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
ETFA ^b	NM_000126.4
ETFB	NM_001985.3
ETFDH	NM_004453.4
FLAD1	NM_025207.5
SLC52A1	NM_001104577.1
SLC52A2	NM_024531.5
SLC52A3	NM_033409.4
TANG02	NM_152906.7

^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.

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

^b There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.