MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Glutaric Aciduria Type II Gene 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]
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
TANGO2	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.

[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.