MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for 3-Methylglutaconic Aciduria 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]
AGK	NM_018238.4
ATP5F1E	NM_006886.4
ATPAF2	NM_145691.4
AUH	NM_001698.2
CLPB	NM_030813.6
CPS1	NM_001875.5
DNAJC19	NM_145261.4
GFER	NM_005262.3
HMGCL	NM_000191.3

Gene	Reference Transcript[a]
HTRA2	NM_013247.4
OPA3	NM_025136.4
POLG	NM_002693.2
SERAC1[b]	NM_032861.4
SUCLA2	NM_003850.2
TAZ	NM_000116.5
TIMM50	NM_001001563.5
TMEM70	NM_017866.6

[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.