MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for 3-Methylglutaconic Aciduria Panel

The following applies to 3MGAP / 3-Methylglutaconic Aciduria 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 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]
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.