# MAYO CLINIC LABORATORIES | *Targeted Genes and Methodology Details for Methylmalonic Aciduria Gene Panel*

The following applies to MMAGP/Methylmalonic Aciduria 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 noncoding 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] | Gene | Reference Transcript[a] |
|---|---|---|---|
| ABCD4 | NM_005050.4 | MMADHC | NM_015702.3 |
| ACSF3 | NM_174917.5 | MTHFR | NM_005957.5 |
| ALDH6A1 | NM_005589.4 | MTR | NM_000254.2 |
| AMN | NM_030943.3 | MTRR | NM_002454.3 |
| CD320 | NM_016579.4 | MMUT | NM_000255.4 |
| CUBN | NM_001081.4 | PRDX1 | NM_002574.3 |
| CBLIF | NM_005142.3 | SUCLA2 | NM_003850.2 |
| HCFC1 | NM_005334.3 | SUCLG1 | NM_003849.4 |
| LMBRD1[b] | NM_018368.4 | TCN1 | NM_001062.4 |
| MCEE | NM_032601.4 | TCN2 | NM_000355.4 |
| MMAA | NM_172250.3 | THAP11 | NM_020457.3 |
| MMAB | NM_052845.4 | ZNF143 | NM_003442.6 |
| MMACHC | NM_015506.3 | | |

[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.