MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Methylmalonic Aciduria 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]
ABCD4	NM_005050.4
ACSF3	NM_174917.5
ALDH6A1	NM_005589.4
AMN	NM_030943.3
CD320	NM_016579.4
CUBN	NM_001081.4
CBLIF	NM_005142.3
HCFC1	NM_005334.3
LMBRD1[b]	NM_018368.4
MCEE	NM_032601.4
MMAA	NM_172250.3
MMAB	NM_052845.4
MMACHC	NM_015506.3

Gene	Reference Transcript[a]
MMADHC	NM_015702.3
MTHFR	NM_005957.5
MTR	NM_000254.2
MTRR	NM_002454.3
MMUT	NM_000255.4
PRDX1	NM_002574.3
SUCLA2	NM_003850.2
SUCLG1	NM_003849.4
TCN1	NM_001062.4
TCN2	NM_000355.4
THAP11	NM_020457.3
ZNF143	NM_003442.6

[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.