MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Maple Syrup Urine Disease 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]
BCKDHA	NM_000709.4
BCKDHB	NM_183050.4
BCKDK	NM_005881.4
DBT	NM_001918.4
DLD	NM_000108.5
PPM1K	NM_152542.5

[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.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.