MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Urea Cycle Disorders 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]
ALDH18A1	NM_002860.4
ARG1	NM_000045.4
ARG2	NM_001172.4
ASL	NM_000048.4
ASS1	NM_000050.4
CA5A	NM_001739.2
CPS1	NM_001875.5
GLUD1	NM_005271.5

Gene	Reference Transcript[a]
GLUL	NM_002065.6
NAGS	NM_153006.3
OAT	NM_000274.4
OTC	NM_000531.6
SLC25A13	NM_014251.3
SLC25A15	NM_014252.4
SLC7A7	NM_001126106.2
UMPS	NM_000373.4

[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.