MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Glycogen Storage 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]
AGL	NM_000642.3
ALDOA	NM_000034.3
ENO3	NM_001976.5
EPM2A	NM_005670.4
FBP1	NM_000507.4
G6PC	NM_000151.4
GAA	NM_000152.5
GBE1	NM_000158.4
GYG1	NM_004130.3
GYS1	NM_002103.5
GYS2	NM_021957.4
LAMP2	NM_002294.3
LDHA	NM_005566.4
NHLRC1	NM_198586.3

Gene	Reference Transcript[a]
PFKM	NM_000289.6
PGAM2	NM_000290.4
PGK1[b]	NM_000291.4
PGM1	NM_002633.3
PHKA1[b]	NM_002637.4
PHKA2[b]	NM_000292.3
PHKB[b]	NM_000293.3
PHKG2	NM_000294.3
PRKAG2[b]	NM_016203.4
PYGL	NM_002863.5
PYGM	NM_005609.4
RBCK1	NM_031229.4
SLC2A2	NM_000340.2
SLC37A4	ENST00000545985.1

[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.