Targeted Genes and Methodology Details for Glycogen Storage Disease Gene Panel

The following applies to GSDGP / Glycogen Storage Disease 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 non-coding 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]
AGL	NM_000642.3	PFKM	NM_000289.6
ALDOA	NM_000034.3	PGAM2	NM_000290.4
ENO3	NM_001976.5	PGK1[b]	NM_000291.4
EPM2A	NM_005670.4	PGM1	NM_002633.3
FBP1	NM_000507.4	PHKA1[b]	NM_002637.4
G6PC	NM_000151.4	PHKA2[b]	NM_000292.3
GAA	NM_000152.5	PHKB[b]	NM_000293.3
GBE1	NM_000158.4	PHKG2	NM_000294.3
GYG1	NM_004130.3	PRKAG2[b]	NM_016203.4
GYS1	NM_002103.5	PYGL	NM_002863.5
GYS2	NM_021957.4	PYGM	NM_005609.4
LAMP2	NM_002294.3	RBCK1	NM_031229.4
LDHA	NM_005566.4	SLC2A2	NM_000340.2
NHLRC1	NM_198586.3	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.