

HOTLINE: Effective September 16, 2021

**New Test**  
Available Now

**3001627**

**Glycogen Storage Disorders Panel, Sequencing**

**GSD NGS**



Patient History for Glycogen Storage Disorders Testing



Additional Technical Information

**Methodology:** Massively Parallel Sequencing  
**Performed:** Varies  
**Reported:** 3-6 weeks

**Specimen Required:** Collect: Lavender or Pink (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 3 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens; saliva; buccal brush or swab; FFPE tissue.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

**Reference Interval:** By report

**Interpretive Data:**

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

**Note:** Genes tested: *ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3,\* FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, OXCT1,\* PFKM,\* PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4.*

\*One or more exons are not covered by sequencing for the indicated gene; see Additional Technical Information.

**CPT Code(s):** 81403, 81404, 81405, 81406, 81407, 81479

New York DOH approval pending. Call for status update.

**HOTLINE NOTE:** Refer to the Test Mix Addendum for interface build information.