

Glycogen Storage Disease Type Ia Testing via *G6PC* Exon Sequencing (Test #221)

Brief Description: Glycogen Storage Disease Type Ia (GSDIa, also known as Von Gierke Disease) is caused by a deficiency of glucose-6-phosphatase, resulting in glycogen accumulation in the liver, kidney, and intestinal mucosa. Symptoms appear in the first year of life with severe fasting hypoglycemia, massive hepatomegaly, hyperlipidemia, hyperuricemia, lactic acidosis, and growth retardation. Patients with GSDIa may also display sweating, irritability, and muscle weakness. For further information see <http://www.agsdus.org/>.

Genetics: GSDIa is inherited in an autosomal recessive manner. Incidence is roughly 1 in 100,000 births with a carrier rate of about 1 in 150. Lei et al. (Science 262:580-583, 1993) identified mutations in the *G6PC* (glucose-6-phosphatase catalytic subunit) gene as the cause of GSDIa. The 12.6 kb *G6PC* gene with 5 exons encodes the key enzyme in homeostatic regulation of blood glucose levels.

Approximately 100 different mutations have been identified in the *G6PC* gene (Shieh et al. J Biol Chem 277:5047-5053, 2002; Froissart and Maire Orphanet Encyclopedia 2002 (<http://www.orpha.net/data/patho/GB/uk-glycogenosis1.pdf>); Human Gene Mutation Database (<http://www.hgmd.org/>)). This particular test involves DNA sequencing of all 5 *G6PC* exons.

Indications for Test: Candidates for this test are patients with symptoms consistent with GSDIa and the family members of patients with known mutations. In addition to this test, PreventionGenetics also offers sequencing of selected single exons of the *G6PC* gene.

Sensitivity of Test: Based on results from the literature, we estimate that our full gene sequencing test will detect likely causative mutations in nearly all patients with GSDIa.

Turn Around Time: Maximum of 40 days, although many tests are completed in less than 2 weeks.

SPECIMEN REQUIREMENTS: See page 4 of the Requisition Form.

Sequential exon sequencing of the <i>G6PC</i> gene	\$440
Molec Diag, Ascertainment	83890
Molec Diag, Isolation	83891
Molecular Diag, Amplif x 5	83898
Mutat Id By Seq, Single Seg x 5	83904
Molecular Diag, Separation	83894
Interpretation And Report	83912

Single exon sequencing for the presence of previously identified mutations in the *G6PG* gene is also available for \$190.

Accreditation Info. CLIA ID #: 52D1027685 (expires 1/18/13) (CAP#: 7185561, AU ID: 1407125 expires 12/20/12)

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