

## Glycogen Storage Disease Type 1a Testing via Single *G6PC* Exon Sequencing

**Brief Description:** Glycogen Storage Disease Type 1a (GSD1a, 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 GSD1a may also display sweating, irritability, and muscle weakness.

**Genetics:** GSD1a 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 GSD1a. 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 a selected single *G6PC* exon.

**Indications for Test:** Candidates for this test are family members of patients with known mutations.

**Sensitivity of Test:** Sensitivity is specified by the particular biological relationship between the patient with the known mutation and his/her relative. For example, a causative mutation will be found in nearly all of the parents of an affected child and, on average, in two-thirds of unaffected siblings of an affected child.

### SPECIMEN REQUIREMENTS

Collect 2-5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube). 5 ml is the preferred volume.

Whole blood collected in Na Heparin is acceptable but not preferred.

Ship whole blood specimens at ambient temperature. Do not freeze blood tube.

During hot summer months, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tubes. In winter, include an unfrozen ice pack to help moderate extremes in temperature. The DNA in whole blood is stable for at least 48 hours at 21°C, 5-7 days at 4°C.

<b>Single exon sequencing of the <i>G6PC</i> gene</b>		<b>\$230.00</b>
<b>Molec Diag, Ascertainment</b>	<b>83890</b>	
<b>Molec Diag, Isolation</b>	<b>83891</b>	
<b>Molecular Diag, Amplif</b>	<b>83898</b>	
<b>Mutat Id By Seq, Single Seg</b>	<b>83904</b>	
<b>Molecular Diag, Separation</b>	<b>83894</b>	
<b>Interpretation And Report</b>	<b>83912</b>	

**Accreditation Info. CLIA ID #: 52D1027685** (expires 1/18/07) (CAP#: 7185561, AU ID: 1407125 expires 12/20/06)

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