

## Glycogen Storage Disease Type 0 Testing Via Glycogen Synthase 2 (*GYS2*) Gene Sequencing (Test #220)

**Brief Description:** Glycogen storage disease type 0 (GSD0), or liver glycogen synthase deficiency, (OMIM 240600) is a form of fasting hypoglycemia presenting in infancy or early childhood and accompanied by high blood ketones and low alanine and lactate concentrations. Although feeding relieves symptoms, it often results in hyperglycemia and hyperlactatemia. Glycogen synthase activity is low or immeasurable in liver. Most cases can be managed by appropriate diet. Glycogen storage disease type 0 is rare, but has probably been substantially underdiagnosed (see Weinstein et al. Mol Genet Metab 87:284-288, 2006). It has been recently reported that about 10% of children with ketotic hypoglycemia have mutations in both copies of the *GYS2* gene. For further information see <http://www.agsdus.org/>.

**Genetics:** Orho et al. (J. Clin. Invest. 102:507-515, 1998) determined that GSD0 is caused by molecular defects in the *GYS2* gene on chromosome 12 encoding the glycogen synthase enzyme present in liver. At least most cases appear to result from recessive inheritance.

**Description of This Particular Test:** Since mutations in the *GYS2* gene have been reported throughout the coding region, and since no common mutations have yet been identified, our test involves full sequencing of all 16 exons in this gene.

**Indications for Test:** Candidates for this test are patients with symptoms consistent with GSD0 and the family members of patients who have known mutations.

**Sensitivity of Test:** Based on literature reports, we estimate that our full gene sequencing test will detect at least one likely causative mutation in nearly all patients with true liver glycogen synthase deficiency. As noted above, it has been reported that about 10% of children with ketotic hypoglycemia have mutations in *GYS2*.

**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.

**Exon sequencing of the *GYS2* gene**

**\$940**

<b>Molec Diag, Ascertainment</b>	<b>83890</b>	<b>\$30</b>
<b>Molec Diag, Isolation</b>	<b>83891</b>	<b>\$40</b>
<b>Molecular Diag, Amplif x 16</b>	<b>83898</b>	<b>\$300</b>
<b>Mutat Id By Seq, Single Seg x 16</b>	<b>83904</b>	<b>\$440</b>
<b>Molecular Diag, Separation</b>	<b>83894</b>	<b>\$50</b>
<b>Interpretation and Report</b>	<b>83912</b>	<b>\$80</b>

**Single exon sequencing for the presence of previously identified mutations in the *GYS2* gene is also available for \$230.**

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

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