PreventionGenetics

Disease Prevention Through Genetic Testing

Glycogen Storage Disease Type 0 (GSD-0) Testing Via Glycogen Synthase 2 (*GYS2*) Gene Sequencing

Glycogen storage disease type 0 (GSD-0), or liver glycogen synthase deficiency, 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.

Orho et al. (J. Clin. Invest. 102:507-515, 1998) determined that GSD-0 is caused by molecular defects in the *GYS2* gene on chromosome 12. At least most cases appear to result from recessive inheritance.

Glycogen storage disease type 0 is relatively rare, but has probably been substantially underdiagnosed. It was recently reported that about 10% of children with ketotic hypoglycemia have mutations in both copies of the *GYS2* gene.

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.

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.
- Only one blood tube is required for multiple tests.
- Ship whole blood specimens at room temperature.
- Do not freeze blood.
- During hot weather, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tube.
- In cold weather, 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.

CPT Codes and Cost

Sequence analysis of *GYS2* gene			**$940.00**
Molec Diag, Ascertainment	**83890**	**x1**	
Molec Diag, Isolation	**83891**	**x1**	
Molecular Diag, Amplif	**83898**	**x 16**	
Mutat Id By Seq, Seg	**83904**	**x 16**	
Molecular Diag, Separation	**83894**	**x1**	
Interpretation And Report	**83912**	**x1**	

Single exon sequencing for known mutations is also available for $230.

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

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