

**Glycogen Storage Disease Type Ib Testing
 via *SLC37A4* (*G6PT1*) Exon Sequencing (Test #222)**

Brief Description: Glycogen Storage Disease Type Ib (GSDIb) is caused by a deficiency of glucose-6-phosphate translocase. Symptoms, which appear in the first year of life, are mostly similar to GSDIa and include severe fasting hypoglycemia, massive hepatomegaly, and hyperlactatemia. In addition, patients with GSDIb may also display neutropenia with abnormal monocytes leading to frequent bacterial and fungal infections. Type Ib patients may also have oral and intestinal mucosal ulcerations and inflammatory intestinal disease suggestive of Crohn’s disease. For further information see <http://www.agsdus.org/>.

Genetics: GSDIb is inherited in an autosomal recessive manner. Veiga-da-Cunha et al. (Am J Hum Genet 63:976-983, 1998) identified mutations in the *SLC37A4* (solute carrier family 37, member 4) gene (also known as the *G6PT1* gene) as the cause of GSDIb. It appears that most or all cases of GSD types 1c and 1d are allelic with GSDIb and are caused by mutations in *SLC37A4* (Veiga-da-Cunha et al. Eur J Hum Genet 7:717-723, 1999).

Approximately 80 different mutations have been identified in *SLC37A4* (Froissart and Maire Orphanet Encyclopedia 2002 (<http://www.orpha.net/data/patho/GB/uk-glycogenosis1.pdf>); Human Gene Mutation Database (<http://www.hgmd.org/>)). The mutations are distributed throughout the length of the gene. Causative mutations are missense, nonsense, splicing, frameshift, and rarely, large deletions.

Description of This Particular Test: This test involves DNA sequencing of all 8 coding exons of *SLC37A4* plus about 50 bp of flanking non-coding DNA on each side.

Indications for Test: Candidates for this test are patients with symptoms consistent with GSDIb and the family members of patients who have known mutations. In addition to this test, PreventionGenetics also offers sequencing of selected single exons of the *SLC37A4* gene.

Sensitivity of Test: Based on literature reports, we estimate that our full gene sequencing test will detect likely causative mutations in nearly all patients with GSDIb. Rarely (in perhaps 3% of patients), only one of the two mutations will be detected.

Turn Around Time: Maximum of 40 days, although many tests are completed in less than 2-3 weeks.

SPECIMEN REQUIREMENTS: See page 4 of the Requisition Form.

Exon sequencing of the *SLC37A4* gene

\$540

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| Molec Diag, Ascertainment | 83890 | \$ 30 |
| Molec Diag, Isolation | 83891 | \$ 40 |
| Molecular Diag, Amplif x 8 | 83898 | \$140 |
| Mutat Id By Seq, Single Seg x 8 | 83904 | \$200 |
| Molecular Diag, Separation | 83894 | \$ 40 |
| Interpretation And Report | 83912 | \$ 90 |

Single exon sequencing for the presence of previously identified mutations in the *SLC37A4* 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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