

## Fanconi-Bickel Syndrome via *SLC2A2* Gene Sequencing (Test #170)

**Brief Description of Disorder:** Fanconi-Bickel Syndrome (OMIM 227810) (also sometimes known as Glycogen Storage Disease, Type XI) is a rare disorder of monosaccharide transport. The disease is usually diagnosed in infancy and is characterized by hepatomegaly, glucose and galactose intolerance, fasting hypoglycemia, short stature, proximal renal tubular dysfunction, and often rickets. See Santer et al. (Hum Genet 110:21-29, 2002) for other references and additional information.

**Genetics:** Fanconi-Bickel Syndrome is inherited in an autosomal recessive manner. All causative genetic defects reported to date have been within the *SLC2A2* (*GLUT2*) gene on chromosome 3q26. About 40 likely causative mutations in this gene have been reported to date (see Santer et al. 2002 and Human Gene Mutation Database (<http://www.hgmd.org/>)). The mutations are located throughout the length of the gene. No mutations are predominant. Although some missense mutations at conserved amino acids have been reported, approximately 75% of the causative mutations have been nonsense, frameshift, or obvious splicing defects.

**Description of This Particular Test:** This test involves bidirectional DNA sequencing of the coding regions of all 11 exons of *SLC2A2* plus about 50 bp of flanking non-coding DNA on each side. We will sequence any single exon or pair of exons in family members of patients with known mutations, and to confirm research results (\$190-340).

**Reference Sequences:** Genomic: NC\_000003.10 mRNA: NM\_000340.1 protein: NP\_000331.1

**Indications for Test:** Patients with symptoms consistent with Fanconi-Bickel Syndrome are candidates for this test.

**Sensitivity of Test:** Santer et al. (2002) reported finding two likely causative mutations in 41 out of 49 Fanconi-Bickel patients (84%). Occasionally, only one of the two mutations in a patient may 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.

**Price: Sequencing of complete coding regions of *SLC2A2* Gene** **\$ 650**

**CPT Codes:**

Sample Ascertainment	83890	\$ 30	DNA Isolation	83891	\$ 40
Amplification x10	83898	\$ 180	Sequencing x10	83904	\$ 280
Separation	83894	\$ 40	Interpretation/Report	83912	\$ 80

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

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