

## Glycogen Storage Disease Type II Testing via GAA Exon Sequencing (Test #223)

**Brief Description:** Glycogen Storage Disease Type II (GSDII, also known as Pompe Disease) (OMIM 232300) is caused by defects in the lysosomal degradation of glycogen. Symptoms consist primarily of weakness in muscles, including cardiac and respiratory muscles. Symptoms can arise during infancy, childhood or adult life. Disease severity also varies broadly. The infantile-onset form often involves heart enlargement and is usually fatal. For further information see <http://www.agsdus.org/>.

**Genetics:** GSDII is inherited in an autosomal recessive manner. Mutations within the GAA gene encoding the acid alpha-glucosidase enzyme are the only known cause of the disease. Over 100 different mutations have been reported in GAA (see Hermans et al. Hum Mut 23:47-56, 2004; [www.pompecenter.nl](http://www.pompecenter.nl); and Human Gene Mutation Database (<http://www.hgmd.org/>)). The mutations are distributed throughout the length of the gene. Missense and nonsense mutations predominate. Some correlations have been made between specific mutations and disease severity. Although founder mutations are known in some genetically isolated populations, in the overall American population no mutations are predominant.

**Description of This Particular Test:** This test involves DNA sequencing from genomic DNA of all 19 coding exons of GAA plus about 50 bp of flanking non-coding DNA on each side. The c.-45T>G mutation sometimes found in late onset patients is covered. The exon 18 deletion mutation common in some European populations is also covered. Although PreventionGenetics clinical test reports focus on the likely causative sequence variants, we do report all deviations from the reference sequences.

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

**Sensitivity of Test:** Through gene sequencing, Hermans et al. were able to detect at least one likely causative mutation in every one of the 29 GSDII patients in their collection. Two likely causative mutations were identified in 24 out of the 29 patients.

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

**Specimen Requirements:** See page 4 of the Requisition Form.

**Price: Sequencing of GAA Gene** **\$ 990**

**CPT Codes:**

Sample Ascertainment	83890	\$ 30	DNA Isolation	83891	\$ 40
Amplification x16	83898	\$ 280	Sequencing x18	83904	\$ 470
Separation	83894	\$ 60	Interpretation/Report	83912	\$ 110

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

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