Urine Glucose Tetrasaccharide Quantitation (HEX4)

Test Code: BHEX4
Turnaround time: 10 days
CPT Codes: 82542 x1, 82570 x1

Condition Description

Pompe disease, also called glycogen storage disease type II (GSD-II), is an autosomal recessive disorder due to a deficiency of the lysosomal enzyme acid alpha-1,4-glucosidase (abbreviated GAA). The function of the GAA enzyme, also known as acid maltase, is to breakdown glycogen in the lysosome. Absent or reduced GAA activity results in accumulation of glycogen within the lysosome, particularly in muscle cells. GSD-II is divided into two forms; an infantile form and a juvenile/adult onset form. In individuals with the infantile form of Pompe disease there is less than 1% of normal enzymatic activity, whereas in the juvenile/adult onset form there is some residual enzymatic activity. In Pompe disease, affected infants are severely hypotonic and have cardiomegaly. In addition, patients may have an enlarged tongue. The disease is usually fatal within the first year of life due cardiorespiratory failure. The clinical presentation in the juvenile/adult onset form (onset after 12 months of age) is much more variable than in the Infantile form of Pompe disease. In this later onset form of the disease, patients generally suffer from slowly progressive proximal muscle weakness with progressive respiratory insufficiency. Unlike the infantile form, in the later onset form there is usually not cardiomegaly or cardiomyopathy.

Pompe disease can be suspected based on clinical findings, or more recently, abnormal newborn screening results. Diagnostic testing for Pompe disease usually involves enzyme analysis, in either leukocytes or dried blood spots, followed by molecular testing to confirm these results. Urine glucose tetrasaccharide levels can be measured to assist in diagnosis and to monitor the response to treatment, once initiated.

Indications

Quantification of glucose tetrasaccharide may be indicated for individuals with a confirmed or suspected diagnosis of Pompe disease.

Methodology

Liquid chromatography - tandem mass spectrometry

Detection

Results should be correlated with clinical findings and the results of enzyme or molecular testing for Pompe disease.

Reference Range

Age specific reference ranges are included in each report.

Specimen Requirements

Type: DNA, Isolated

Specimen Requirements:
- Microtainer
- 3µg
- Isolation using the Perkin Elmer™Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:
- Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)

Specimen Requirements:
- EDTA (Purple Top)
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml
- Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:
- Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Related Tests

DW: Pompe disease: Acid alpha-glucosidase activity, leukocytes
AN: Pompe disease: GAA sequencing
NF: Pompe disease: GAA gene deletion / duplication

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