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**Lab Dept:** Urine/Stool

**Test Name:** **HEX4 (GLUCOSE TETRASACCHARIDE, URINE FOR POMPE DISEASE)**

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***General Information***

**Lab Order Codes:** HEX4

**Synonyms:** Urine Glucose Tetrasaccharide (Hex4) for Pompe Disease; Pompe Disease; HEX4, Urine; Acid Maltase Deficiency; GSD Type II

**CPT Codes:** 82570 – Creatinine; other source  
83789 – Mass spectrometry and tandem mass spectrometry, analyte not elsewhere specified; quantitative, each specimen

**Test Includes:** Hex4 concentrations are measured relative to creatinine and reported as a normalized Hex4 in mmol/mol creatinine.

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***Logistics***

**Test Indications:** Useful for the diagnosis of Pompe disease and to monitor patients on enzyme replacement therapy.

As a monitoring tool, urine Hex4 can be used as an indirect measure of the degree of skeletal muscle glycogen clearance in patients with Pompe disease receiving enzyme replacement therapy.

**Lab Testing Sections:** Urine/Stool - Sendouts

**Referred to:** Duke University School of Medicine

**Phone Numbers:** MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:** Monday - Thursday

**Turnaround Time:** 10 – 14 days

**Special Instructions:** Provide the date and time of last meal for the patient.

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***Specimen***

**Specimen Type:** Urine

**Container:** Plastic leakproof container (No preservative)

**Draw Volume:** 1 mL (Absolute Minimum: 0.25 mL)

**Processed Volume:** Same as Draw Volume

**Collection:** Random urine collection, void or catheterized

**Special Processing:** Lab Staff: A well-mixed aliquot should be placed into a screw-capped round bottom plastic vial. Freeze and ship on dry ice. Specimens can only be shipped Monday – Thursday overnight. There are no Saturday deliveries. Include the Duke Biochemical Genetics Request Form with the specimen.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimens

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***Interpretive***

**Reference Range:**

<b>Age Specific Hex4 Normalized Control Ranges</b>	
<b>Age</b>	<b>Result (mmol/mol creatinine)</b>
<6 months:	<19
6 months – 1 year:	<14
>1 year:	<4

**Critical Values:**

N/A

**Limitations:**

Biochemical test results depend in part on the clinical and dietary status at the time of specimen collection. A normal or non-diagnostic test result does not rule out the possibility of an underlying metabolic disorder, including that for which the test was requested.

**Methodology:**

Liquid Chromatography – Tandem Mass Spectrometry (LC-MS/MS)

**References:**

[Duke University Hospital Biochemical Genetics Laboratory](#) June 2013  
 Phone: 909-549-0445 Fax: 919-549-0709