
Lab Dept: Urine/Stool

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

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.

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: Daily, 24 hours

Turnaround Time: 10 – 14 days

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

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 – Thurs. There are no Saturday deliveries. If a specimen is collected on Friday, or the weekend, store frozen and ship on Monday or the next business day if a holiday falls on Monday. Include the Duke Biochemical Genetics Request Form with the specimen.

Patient Preparation: None

Sample Rejection: Mislabeled or unlabeled specimens

Interpretive

Reference Range:

Age Specific Hex4 Normalized Control Ranges	
Age	Result (mmol/mol creatinine)
<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](#) March 2020
Phone: 909-549-0445 Fax: 919-549-0709

Updated: 3/20/2020: Updated test availability and lab processing for shipping info.