Lab Dept:	**Urine/Stool**
Test Name:	**LYSOSOMAL STORAGE DISORDERS SCREEN, URINE**

General Information

Lab Order Codes:	LYSDU
Synonyms:	Lysosomal Disorders Screen Urine
CPT Codes:	83789 – Ceramide Trihex/Sulfatide, Urine 84377 – Oligosachharide Screen, Urine 83864 – Mucopolysaccharides, acid, quantitative 84275 – Sialic Acid
Test Includes:	This is a general urine screening test for a broad array of lysosomal storage and related disorders. Lysosomal storage disorders are a group of genetic diseases characterized by the accumulation of substrates in the cells and tissues of affected individuals. There is a significant phenotypic overlap between lysosomal storage disorders making diagnosis a challenge. In many cases, accumulating analytes spill out into bodily fluids and can be detected in urine; therefore, the first step in diagnostic workup includes urine analyses for metabolites associated with specific lysosomal storage disorders. The recognition of disease specific metabolites in the screening profile can help secure a diagnosis. Targeted follow-up testing can and should be performed using enzymatic or molecular assays.

Logistics

Test Indications:	This test is the recommended screening test for the initial workup of a suspected lysosomal storage disorder (LSD) when the patient's clinical features are not suggestive of any particular LSD.
Lab Testing Sections:	Urine/Stool - Sendouts
Referred to:	Mayo Clinic Laboratories (MML Test: LSDS)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours

Turnaround Time:	8-14 days
Special Instructions:	Early morning random urine specimen preferred.

Specimen

Specimen Type:	Urine, random Early morning collection preferred
Container:	Plastic leakproof container (No preservative). Mayo urine container T313
Draw Volume:	Submit entire random urine collection
Processed Volume:	12 mL (Minimum: 3.5 mL) urine
Collection:	A random urine sample may be obtained by voiding into a urine cup. Make sure all specimens submitted to the laboratory are properly labeled with the patient's name, medical record number and date of birth. Early morning collection is preferred.
Special Processing:	Lab staff: Mix urine specimen well before aliquot is taken. Aliquot 12 mL (Minimum: 3.5 mL) urine. Store and ship refrigerated. Forward promptly. Specimen stable refrigerated (preferred) for 15 days, frozen for 90 days.
Patient Preparation:	Do not administer low-molecular weight heparin before specimen collection. Baby wipes or wipes containing soaps or lotions should not be used before specimen collection because these may interfere with results.
Sample Rejection:	Mislabeled or unlabeled specimens; ambient specimens.

Interpretive

Reference Range:	An interpretive report will be provided. See reference lab website for further details.
Critical Values:	N/A
Limitations:	Specific enzymatic or molecular assays should be used to confirm positive results. In rare instances, a normal excretion of ceramide trihexosides may be seen in individuals who are carriers of, or affected with Fabry disease.

	Not all lysosomal storage disorders are detectable through urine screening.
Methodology:	CTS02, OLI02: Matrix-Assisted Laser Desorption/Ionization Time-of-Flight Mass Spectrometry (MALDI-TOF MS) MPS02: Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS) BG721: Medical Interpretation
References:	Mayo Clinical Laboratories November 2024
Updates:	12/10/2024: Added Sialic acid, updated CPT codes, updated optimal and minimum specimen volume, updated turnaround times, added patient preparation, updated methodology, added specimen stability.