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

General Information

Lab Order Codes:	LYSDU
Synonyms:	N/A
CPT Codes:	82542 – Ceramide Trihex/Sulfatide, Urine 82542 – Mucopolysachharides MPS, Qualitative, Urine 83864 – Mucopolysaccharadies (MPS), Quantitative, Urine 84377 – Oligosachharide Screen, Urine
Test Includes:	This is a general urine screening test for a broad array of lysosomal storage and related disorders. Not all lysosomal storage disorders are detectable by this method. 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:	Screening patients suspected of having a lysosomal storage disorder.
Lab Testing Sections:	Urine/Stool - Sendouts
Referred to:	Mayo Medical Laboratories (MML Test: LYSDU)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	2 – 3 weeks

Special Instructions:	Early morning random urine specimen preferred.

Specimen

Specimen Type:	Urine, random Early morning collection preferred
Container:	Plastic leakproof container (No preservative).
Draw Volume:	Submit entire random urine collection
Processed Volume:	10 mL (Minimum: 3 mL) urine
Collection:	A random urine sample may be obtained by voiding into a urine cup. Specimens need to be frozen as soon as possible after collection. Make sure all specimens submitted to the laboratory are properly labeled with the patient's name, medical record number and date of birth.
Special Processing:	Lab staff: Mix urine specimen well before aliquot is taken. Aliquot 10 mL (Minimum: 3 mL) urine. Store in freezer. Ship frozen. Forward promptly.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	An interpretive report will be provided. Abnormal results are not sufficient to conclusively establish a diagnosis of a particular disease. Follow-up testing is recommended to confirm a diagnosis. When abnormal results are detected with characteristic patterns, a detailed interpretation is given, including an overview of the results and their significance, a correlation to available clinical information, elements of differential diagnosis, recommendations for additional biochemical testing and in vitro confirmatory studies (enzyme assay and molecular test).
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. Occasionally, an abnormal value for glycosaminoglycans (GAG) will be

obtained on a specimen that yields a normal liquid chromatography-tandem mass spectrometry (LC-MS/MS) pattern. This situation can occur as an artifact when a patient is treated with low-moleular-weight heparin. Other known causes are sample contamination with acrylic polymers used in disposable diapers and several clinical situations associated with excessive connective tissue destruction, bladder disease, or bone disease.

Not all lysosomal storage disorders are detectable through urine screening.

Methodology: Liquid Chromatography-Tandem Mass Spectrometry (GC-MS)
Spectrophotometry (SP)

References:

Mayo Medical Laboratories Web Page June 2017