Lab Dept: Chemistry

Test Name: LYSOSOMAL ENZYMES

**General Information**

Lab Order Codes: LYE

Synonyms: N/A

CPT Codes:
- 82657 – Enzyme activity in blood cells, cultured cells, or tissue, not elsewhere specified; nonradioactive substrate, each specimen
- 82658 – Enzyme activity in blood cells, cultured cells, or tissue, not elsewhere specified; radioactive substrate, each specimen

Test Includes: The majority of patients are initially screened by enzyme assay, which is the most efficient method to arrive at a definitive diagnosis.

**Logistics**

Test Indications: Lysosomal storage diseases are genetic disorders resulting from mutations in genes coding for proteins involved in the degradation of normal body compounds, including lipids, proteins and complex carbohydrates. While most lysosomal disorders are caused by defects in genes coding for enzymes, some are caused by defects in genes coding for transport proteins, activator proteins, protective proteins or enzymes involved in the processing of lysosomal enzymes or substrates. The lack of one of these proteins leads to the accumulation of undegraded compounds which can disrupt the normal functioning of the cells involved. Most of these disorders are autosomal recessively inherited, however a few are X-linked recessively inherited, such as Fabry disease and Hunter syndrome (MPS II).

Lab Testing Sections: Chemistry - Sendouts

Referred to: Jefferson Medical College, Dr. David Wenger's Lab

Phone Numbers:
- MIN Lab: 612-813-6280
- STP Lab: 651-220-6550

Test Availability: Monday – Wednesday (anytime), Thursday before Noon

Turnaround Time: Approximately 1 week from the time of arrival at the reference lab

Special Instructions: Specimen must arrive in the lab within 4 hours of draw. See Test Availability for restricted draw times. Please include a Clinical History form.
**Specimen**

Specimen Type: Whole blood

Container: Green top (Li or Na Heparin) tube  
Alternate: Lavender (EDTA) tube

Draw Volume: 4 -6 mL (Minimum: 2 mL) blood

Processed Volume: Same as Draw Volume

Collection: Routine venipuncture

Special Processing: Lab Staff: Do Not centrifuge. Specimen should remain in original collection container and be kept at room temperature. Promptly send specimen Monday – Wednesday (anytime) and Thursday by Noon only. Ship Federal Express at ambient temperatures. Fax health history to 1-215-955-9554. Never draw or ship on Friday.

Patient Preparation: None

Sample Rejection: Mislabeled or unlabeled specimens; improper specimen handling; frozen specimens

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**Interpretive**

Reference Range: Interpretive report

Critical Values: N/A

Limitations: N/A

Methodology: Spectrophotometric Enzyme Measurement

References: Jefferson Medical College, Philadelphia, PA, Dr. David Wenger’s Laboratory December 2017

Updates: 12/20/2017: Added alternate collection container and volume requirements.