
Lab Dept: Chemistry

Test Name: LYSOSOMAL ENZYMES

General Information

Lab Order Codes: LYE

Synonyms: Leukocyte Lysosomal Enzyme Screen

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: Tests for individual patients are selected by the reference lab based upon clinical features, previous laboratory and/or radiologic findings, family history, and physician suggestions. Further studies may be indicated by the results of our preliminary testing. See [reference lab's list](#) for diseases included in screening.

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: Lysosomal Diseases Testing Laboratory at Thomas Jefferson University

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: **Collection Monday through Thursday only**, to arrive overnight within 24 hours of collection by Friday. **Never collect Friday - Sunday.**

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

Special Instructions: [Restricted draw times](#). A [Clinical History](#) form is required.

Specimen

Specimen Type: Whole blood

Container: Lavender (EDTA) tube
Alternate: Green top (Li or Na Heparin no gel) 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 ship via Federal Express First Overnight at ambient temperatures Monday – Thursday ONLY. Insulate specimen in a styro box to stabilize the ambient temperature.

Specimen is stable for 24 hours. Consult with the reference laboratory at 215-955-1666 for approval to send specimens that would be received past 24 hours from collection.

See [website](#) for shipping information and required paperwork.

Never draw or ship on Friday - Sunday.

Patient Preparation: None

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

Interpretive

Reference Range: Interpretive report

Critical Values: N/A

Limitations: N/A

Methodology: Spectrophotometric Enzyme Measurement

References: [Lysosomal Diseases Testing Laboratory at Thomas Jefferson University](#)
March 2024

Updates:

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

3/7/2024: Updated broken links to reference lab and added additional links to details. Clarified collection restriction timing.