
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: 3 – 5 days, test set up Thursday and Friday

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 available from lab
Draw Volume:	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

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 April 2010