MAYO CLINIC LABORATORIES

Test Definition: LALBS

Lysosomal Acid Lipase, Blood Spot

Overview

Useful For

Evaluation of patients with a clinical presentation suggestive of lysosomal acid lipase deficiency using blood spot specimens

This test is **not useful** to determine carrier status for cholesteryl ester storage disease or Wolman disease.

Genetics Test Information

This test provides diagnostic testing for patients with clinical signs and symptoms suspicious for lysosomal acid lipase deficiency (LALD).

Lysosomal acid lipase deficiency is expressed phenotypically as infantile-onset Wolman disease or later-onset cholesterol ester storage disease.

Special Instructions

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Blood Spot Collection Card-Spanish Instructions
- Blood Spot Collection Card-Chinese Instructions
- Informed Consent for Genetic Testing (Spanish)
- Blood Spot Collection Instructions

Method Name

Fluorometric Enzyme Assay

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Necessary Information

Provide a reason for testing with each specimen.

Specimen Required

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)
Collection Container/Tube:
Preferred: Blood spot collection card
Acceptable: PerkinElmer 226 filter paper, Munktell TFN, or Whatman Protein Saver 903 Paper

Specimen Volume: 2 Blood spots

Collection Instructions:

1. Completely fill at least 2 circles on the filter paper card (approximately 100 microliters blood per circle).
2. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see How to Collect a Dried Blood Spot Sample.
3. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
4. Do not expose specimen to heat or direct sunlight.
5. Do not stack wet specimens.
6. Keep specimen dry.

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions.
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777).
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800).

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Biochemical Genetics Patient Information (T602)
3. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:
-Biochemical Genetics Test Request (T798)
-Gastroenterology and Hepatology Test Request (T728)

Specimen Minimum Volume

1 Blood spot

Reject Due To

Shows serum rings	Reject
Multiple layers	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	28 days	FILTER PAPER
	Ambient	7 days	FILTER PAPER
	Frozen	90 days	FILTER PAPER

Clinical & Interpretive

Clinical Information

Deficiency of lysosomal acid lipase (LAL) results in 2 clinically distinct phenotypes, Wolman disease (WD) and cholesteryl

ester storage disease (CESD). Both phenotypes follow an autosomal recessive inheritance pattern and are caused by variants in the *LIPA* gene.

Wolman disease, the early-onset phenotype of LAL deficiency, is a lipid storage disorder characterized by vomiting, diarrhea, failure to thrive, abdominal distension, hepatosplenomegaly, and liver failure. Enlarged adrenal glands with calcification, a classic finding in WD, can lead to adrenal cortical insufficiency. Unless successfully treated, survival is rare beyond infancy.

Cholesteryl ester storage disease, the late-onset phenotype of LAL deficiency, is clinically variable with patients presenting at any age with progressive hepatomegaly and often splenomegaly, serum lipid abnormalities, and elevated liver enzymes. In childhood, patients can also present with failure to thrive and delayed milestones. Common features include premature atherosclerosis leading to coronary artery disease and strokes, liver disease of varying severity, and organomegaly. Lipid deposition in the intestinal tract can lead to diarrhea and weight loss.

Cholesteryl ester storage disease is likely underdiagnosed and frequently diagnosed incidentally after liver pathology reveals findings similar to nonalcoholic fatty liver disease or nonalcoholic steatohepatitis. Birefringent cholesteryl ester crystals in hepatocytes or Kupffer cells in fresh-frozen tissues are visualized under polarized light and pathognomonic.

Enzyme replacement therapy is available for both WD and CESD making early diagnosis essential to improve long-term outcomes.

Reference Values

> or =21.0 nmol/h/mL

Interpretation

Enzyme activity below 1.5 nmol/h/mL in properly submitted samples is consistent with lysosomal acid lipase deficiency: Wolman disease or cholesteryl ester storage disease.

Normal results (> or =21.0 nmol/h/mL) are not consistent with lysosomal acid lipase deficiency.

Cautions

No significant cautionary statements

Clinical Reference

1. Balwani M, Vijay S. Lysosomal Acid Lipase Deficiency. 2015 . In: Adam MP, Bick S, Mirzaa GM, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 1993-2026. Updated January 15, 2026. Accessed May 5, 2026. Available at www.ncbi.nlm.nih.gov/books/NBK305870/

2. Pericleous M, Kelly C, Wang T, Livingstone C, Ala A. Wolman's disease and cholesteryl ester storage disorder: the phenotypic spectrum of lysosomal acid lipase deficiency. Lancet Gastroenterol Hepatol. 2017;2(9):670-679. doi:10.1016/S2468-1253(17)30052-3

3. Pastores GM, Hughes DA. Lysosomal acid lipase deficiency: Therapeutic options. Drug Des Devel Ther. 2020;14:591-601

Performance

Method Description

A 3-mm (one-eighth inch) disk is punched out of the dried blood spot into a microcentrifuge tube and water is added as a preincubation extraction that takes place on an orbital shaker. Extraction liquid is combined with either water (total activity well) or Lalistat (inhibited well) in a black 96-well plate. The plate is incubated. The substrate is then added to the same plate. After the incubation period, calibrators are prepared and analyzed on every plate to calculate enzyme activity results based on fluorescence units in patient wells vs calibrators. The calibration is derived from 4-methylumbelliferone (4-MU) that is serially diluted manually in the plate, with the highest calibrator being equivalent to an enzyme activity of 672.0 nmol/hour/mL blood. The plate is then ready to be read using the spectrofluorometer. Enzyme activity is calculated by subtracting the inhibited activity from total activity.(Hamilton J, Jones I, Srivastava R, Galloway P. A new method for the measurement of lysosomal acid lipase in dried blood spots using the inhibitor Lalistat 2. Clin Chim Acta. 2012;413[15-16]:1207-1210; Cowan T, Pasquali M: Laboratory investigations of inborn errors of metabolism. In: Sarafoglou K, Hoffman GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. 2nd ed. McGraw-Hill; 2017:1139-1158)

PDF Report

No

Day(s) Performed

Thursday

Report Available

2 to 8 days

Specimen Retention Time

1 year

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82657

LOINC® Information

MAYO CLINIC LABORATORIES

Test Definition: LALBS

Lysosomal Acid Lipase, Blood Spot

Test ID	Test Order Name	Order LOINC® Value
LALBS	Lysosomal Acid Lipase, BS	73958-1

Result ID	Test Result Name	Result LOINC® Value
62955	Lysosomal Acid Lipase, BS	73958-1
36341	Reviewed By	18771-6
36340	Interpretation (LALBS)	59462-2