Test Definition: LPCBS

Lysophosphatidylcholines, LC MS/MS, Blood Spot

Overview

Useful For

Second-tier newborn screen for X-linked adrenoleukodystrophy

This test is **not intended for** metabolic screening of symptomatic patients.

This test is supplemental and **not intended to** replace state mandated newborn screening.

Genetics Test Information

This test is used as a second-tier newborn screen for X-linked adrenoleukodystrophy (X-ALD).

Testing Algorithm

For more information see Newborn Screen Follow-up for X-linked Adrenoleukodystrophy.

If the patient has an abnormal newborn screening result for X-linked adrenoleukodystrophy, immediate action should be taken. Refer to the appropriate ACMG Newborn Screening ACT Sheet.(1)

Special Instructions

- Biochemical Genetics Patient Information
- Blood Spot Collection Card-Spanish Instructions
- Newborn Screen Follow-up for X-Linked Adrenoleukodystrophy
- Blood Spot Collection Card-Chinese Instructions
- Blood Spot Collection Instructions

Highlights

Testing for C24:0 lysophosphatidylcholines (LPC) and C26:0 LPC can aid in the diagnosis of X-linked adrenoleukodystrophy (XALD) as well as other types of peroxisomal disorders.

Analysis of LPC in a blood spot test is a useful second-tier newborn screen test for XALD.

Method Name

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Specimen Required

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)
Container/Tube:
Preferred: Blood Spot Collection Card
Acceptable: PerkinElmer 226 filter paper, Munktell filter paper, Whatman Protein Saver 903 Paper, local newborn screening card, or blood collected in tubes containing ACD or EDTA, and then spotted and dried on filter paper
Specimen Volume: 2 Blood spots
Collection Instructions:
1. 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.
2. Completely fill at least 2 circles on the filter paper card (approximately 100 microliters blood per circle).
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. Biochemical Genetics Patient Information (T602)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

1 Blood spot

Reject Due To

Blood spot specimen that shows serum rings or has multiple layers	Reject
Insufficient specimen	Reject
Specimens known to have been exposed to elevated temperature above ambient	Reject

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

This assay measures C20, C22, C24, and C26 lysophosphatidylcholine (LPC) species in dried blood spots by liquid chromatography tandem mass spectrometry.

Peroxisomes are organelles present in all human cells except mature erythrocytes. They carry out essential metabolic functions, including beta-oxidation of very long-chain fatty acids, alpha-oxidation of phytanic acid, and biosynthesis of plasmalogen and bile acids. Peroxisomal disorders include 2 major subgroups: disorders of peroxisomal biogenesis and single peroxisomal enzyme/transporter defects. Peroxisome biogenesis defects, such as Zellweger spectrum disorder (ZSD), are characterized by defective assembly of the entire organelle, whereas in single enzyme/transporter defects, such as X-linked adrenoleukodystrophy (XALD), the organelle is intact but a specific function is disrupted. These disorders are clinically diverse and range in severity from neonatal lethal to later onset milder variants.

X-linked adrenoleukodystrophy is an X-linked disorder affecting the nervous system, adrenal cortex, and testis. It is the most common of the peroxisomal disorders. XALD is caused by a disease-causing variant in the *ABCD1*. XALD shows a wide range of phenotypic expressions. The clinical phenotypes occurring in male patients can be subdivided in 4 main categories: cerebral inflammatory, adrenomyeloneuropathy (AMN), Addison only, and asymptomatic. The first 2 phenotypes account for almost 80% of the patients, while the frequency of the asymptomatic category diminishes with age and is very rare after age 40. It is estimated that approximately 65% to 80% of heterozygous individuals develop symptoms of an AMN-like phenotype. Treatment options include hormone replacement therapy, hematopoietic stem cell transplantation, gene therapy, or symptom management.

Elevations of C24 LPC and C26 LPC may be indicative of XALD. In 2016, XALD was added to the US Recommended Uniform Screening Panel, a list of conditions that are nationally recommended for newborn screening by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. Therefore, measurement of LPCs is a useful second-tier test for newborn screening for XALD.

Zellweger spectrum disorder are a continuum of severe disorders affecting the nervous system, vision, hearing, and liver function. Most affected individuals present in childhood, but adult patients have been identified. Most ZSD are inherited in an autosomal recessive pattern. At least 13 different genes have been implicated in ZSD, with approximately 60% to 70% of variants occurring in *PEX1*. The clinical phenotypes include Zellweger syndrome, neonatal adrenoleukodystrophy, and infantile Refsum disease. The phenotypic spectrum and disease severity is broad. There is no specific treatment for ZSD. Although ZSD are not a primary disease target for testing, this test can detect individuals with ZSD.

Reference Values

Analyte	Normal Range (nmol/mL)
C20 Lysophosphatidylcholine	Not applicable
C22 Lysophosphatidylcholine	Not applicable
C24 Lysophosphatidylcholine	< or =0.41
C26 Lysophosphatidylcholine	< or =0.31

Interpretation

In female patients: Elevations of C24 lysophosphatidylcholine (LPC) or C26 LPC may be indicative of heterozygosity for X-linked adrenoleukodystrophy (XALD) or other forms of peroxisomal disorders.

In male patients: Elevations of C24 LPC or C26 LPC may be indicative of XALD or other forms of peroxisomal disorders.

Abnormal results are not sufficient to conclusively establish a diagnosis of a particular disease. To verify a preliminary diagnosis based on the analysis, independent biochemical (eg, in vitro enzyme assay) or molecular genetic analyses are required.

Cautions

This test cannot reliably detect carrier status (heterozygosity) for these conditions.

A positive test result is strongly suggestive of a diagnosis but requires follow-up by stand-alone biochemical or molecular assay, which is best coordinated by local genetics providers.

Clinical Reference

1. ACMG Newborn Screening ACT Sheets. Accessed October 2, 2025. Available at www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms.aspx?hkey=9d6bce5a-182e-42a6-84a5-b2d88240c508
2. Huffnagel IC, van de Beek MC, Showers AL, et al. Comparison of C26:0-carnitine and C26:0-lysophosphatidylcholine as diagnostic markers in dried blood spots from newborns and patients with adrenoleukodystrophy. Mol Genet Metab. 2017;122(4):209-215. doi:10.1016/j.ymgme.2017.10.012
3. Klouwer FCC, Ferdinandusse S, van Lenthe H, et al. Evaluation of C26:0-lysophosphatidylcholine and C26:0-carnitine as diagnostic markers for Zellweger spectrum disorders. J Inherit Metab Dis. 2017;40(6):875-881. doi:10.1007/s10545-017-0064-0
4. Sandlers Y, Moser AB, Hubbard LE, Kratz LE, Jones RO, Raymond GV. Combined extraction of acyl carnitines and 26:0 lysophosphatidylcholine from dried blood spots: prospective newborn screening for X-linked adrenoleukodystrophy. Mol Genet Metab. 2012;105(3)416-420

Performance

Method Description

Internal standard in methanol is added to a dried blood spot. The extract is evaporated and reconstituted prior to injection onto a liquid chromatography tandem mass spectrometry (LC-MS/MS) system. The lysophosphatidylcholine

(LPC) concentrations are measured by MS/MS analysis in the multiple reaction monitoring positive mode to follow the precursor to product species transitions. The ratio of the extracted peak area to internal standard as determined by LC-MS/MS is used to calculate the concentration of LPC species in the sample.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Saturday

Report Available
2 days

Specimen Retention Time
6 months

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
82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
LPCBS	LysoPC by LC MS/MS, BS	105457-6

Result ID	Test Result Name	Result LOINC® Value
34860	C20 Lysophosphatidylcholine	90920-0
34861	C22 Lysophosphatidylcholine	90921-8
34862	C24 Lysophosphatidylcholine	90922-6
34863	C26 Lysophosphatidylcholine	90923-4

34864	Reviewed By	18771-6
34865	Interpretation (LPCBS)	59462-2