

TEST OBSOLETE

NOTIFICATION DATE: February 15, 2013 EFFECTIVE DATE: March 19, 2013

Lysosomal Disorders Screen, Blood Spot

Test ID: LDSBS Secondary ID: 89406

EXPLANATION: Effective March 19, 2013, Lysosomal Disorders Screen, Blood Spot will be obsolete. A recommended alternative test is detailed below.

RECOMMENDED ALTERNATIVE TEST:

Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot Test ID: PLSD

METHODOLOGY: Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS)

REFERENCE VALUES:

TELLECE VIEGES.					
Disease	Marker	Normal Range			
Gaucher	Acid Beta-Glucosidase	> or =3.0 nmol/mL/hr			
Niemann-Pick A/B	Sphingomyelinase	> or =5.5 nmol/mL/hr			
Pompe	Acid Alpha-Glucosidase	> or =4.0 nmol/mL/hr			
Krabbe	Galactocerebrosidase	> or =0.4 nmol/mL/hr			
Fabry	Alpha-Galactosidase	> or =4.0 nmol/mL/hr			
MPS I	Alpha-L-Iduronidase	> or =2.0 nmol/mL/hr			
NA	C20 Lysophosphatidylcholine	< or $=0.26$ mcg/mL			
NA	C22 Lysophosphatidylcholine	< or =0.11 mcg/mL			
ALD/PBD/ALDH	C24 Lysophosphatidylcholine	Female <0.10 mcg/mL			
		Female indeterminate = 0.10-0.20 mcg/mL			
		Male $<$ or $=0.20 \text{ mcg/mL}$			
ALD/PBD/ALDH	C26 Lysophosphatidylcholine	< or $=0.30 mcg/mL$			

Reflex Tests

Test ID	Reporting Name	Available Separately	Always Performed
LPCBS	Lysophosphatidylcholines	No	No

Testing Algorithm: If C26 lysophosphatidylcholine is >0.30, then lysophosphatidylcholines will be performed at an additional charge

SPECIMEN REQUIREMENTS:

Preferred: Whatman Protein Saver 903 Paper

Acceptable: Ahlstrom 226 filter paper, Munktell and Supplemental Newborn Screening

Card (Supply T493)

Specimen Volume: 2 blood spots

Collection Instructions:

- 1. An alternative blood collection option for a patient >1 year of age is fingerstick.
- 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
- 3. Do not expose specimen to heat or direct sunlight.
- 4. Do not stack wet specimens.
- 5. Keep specimen dry.

Forms: New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Whole Blood	Frozen (preferred)	56 days
	Ambient	7 days
	Refrigerated	56 days

DAY(S) SET UP: Tuesday

ANALYTIC TIME: 2 days