MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: February 15, 2013
EFFECTIVE DATE: March 19, 2013

LYSOSOMAL AND PEROXISOMAL STORAGE DISORDERS SCREEN, BLOOD SPOT

Test ID: PLSD

USEFUL FOR: Evaluation of patients with a clinical presentation suggestive of a lysosomal storage disorder, specifically Gaucher, Niemann-Pick type A or type B, Pompe, Krabbe, Fabry disease, or mucopolysaccharidosis I; or a peroxisomal disorder, either X-linked adrenoleukodystrophy or Zellweger spectrum syndrome

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

REFERENCE VALUES:

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 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

CAUTIONS: Specimens exposed to heat >25 degrees C will be cancelled.

A positive test result is strongly suggestive of a diagnosis but needs follow-up by stand-alone biochemical or molecular assay.

CPT CODE: 83789

DAY(S) SET UP: Tuesday **ANALYTIC TIME**: 2 days