Test Definition: PLSD

Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot

Reporting Title: Lysosomal/Peroxisomal D/O Scrn, BS
Performing Location: Rochester

Specimen Requirements:

Container/Tube:
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 Type	Temperature	Time
Whole blood	Frozen (preferred)	56 days
	Refrigerated	56 days
	Ambient	7 days

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
34811	Acid Beta-Glucosidase	Numeric	nmol/mL/h	In Process
34812	Sphingomyelinase	Numeric	nmol/mL/h	In Process
34813	Acid Alpha-Glucosidase	Numeric	nmol/mL/h	In Process
34814	Galactocerebrosidase	Numeric	nmol/mL/h	In Process
34815	Alpha-Galactosidase	Numeric	nmol/mL/h	In Process
34816	Alpha-L-Iduronidase	Numeric	nmol/mL/h	In Process
34817	C20 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
34818	C22 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
34819	C24 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
34820	C26 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
34821	Interpretation (PLSD)	Alphanumeric		In Process

Result ID	Reporting Name	Type	Unit	LOINC®
34822	Reviewed By	Numeric		In Process

CPT Code Information:

83789

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
LPCBS	LysoPC by LC MS/MS (Reflex), BS			No	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
LPCBS	34860	C20 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
LPCBS	34861	C22 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
LPCBS	34862	C24 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
LPCBS	34863	C26 Lysophosphatidylcholine	Numeric	mcg/mL	In Process
LPCBS	34864	Reviewed By	Numeric		In Process
LPCBS	34865	Interpretation (LPCBS)	Alphanumeric		In Process

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 Female indeterminate = 0.10-0.20 mcg/mL Male < or =0.20 mcg/mL
ALD/PBD/ALDH	C26 Lysophosphatidylcholine	< or =0.30 mcg/mL