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

To evaluate adult patients with a clinical presentation suggestive of adrenomyeloneuropathy, the recommended test is POX / Fatty Acid Profile, Peroxisomal (C22-C26), Serum. Lysophosphatidylcholine concentrations may not be consistently elevated in adult blood spots.

Specimen Requirements:

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 dried on acceptable 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 Dried Blood Spot Samples.
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)

Specimen Minimum Volume:

1 Blood spot

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 a Biochemical Genetics Test Request (T798) with the specimen.

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

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
34811	Acid Beta-Glucosidase	Numeric	nmol/mL/h	55917-9
34812	Sphingomyelinase	Numeric	nmol/mL/h	62316-5
34813	Acid Alpha-Glucosidase	Numeric	nmol/mL/h	55827-0
34814	Galactocerebrosidase	Numeric	nmol/mL/h	62310-8
34815	Alpha-Galactosidase	Numeric	nmol/mL/h	55908-8
34816	Alpha-L-Iduronidase	Numeric	nmol/mL/h	55909-6
620785	Iduronate 2-Sulfatase	Numeric	nmol/mL/h	79462-8
34817	C20 Lysophosphatidylcholine	Numeric	nmol/mL	90920-0
34818	C22 Lysophosphatidylcholine	Numeric	nmol/mL	90921-8
34819	C24 Lysophosphatidylcholine	Numeric	nmol/mL	90922-6
34820	C26 Lysophosphatidylcholine	Numeric	nmol/mL	90923-4
34821	Interpretation (PLSD)	Alphanumeric		62301-7
34822	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

83789

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
MPSBS	Mucopolysaccharidosis, BS			No	Yes
PSY	Psychosine, BS			No	Yes
GPSY	Glucopsychosine, BS			No	Yes
OXYBS	Oxysterols, BS			No	Yes
LPCBS	LysoPC by LC MS/MS, BS			No	Yes
PDBS	Pompe Disease, BS			No	Yes
LGBBS	Globotriaosylsphingosine, BS			No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
MPSBS	43695	Interpretation (MPSBS)	Alphanumeric		59462-2
MPSBS	43693	Dermatan Sulfate	Numeric	nmol/L	90233-8
MPSBS	43694	Heparan Sulfate	Numeric	nmol/L	90235-3
MPSBS	BA2869	Total Keratan Sulfate	Alphanumeric	nmol/L	90236-1
MPSBS	43696	Reviewed By	Alphanumeric		18771-6
PSY	62235	Psychosine	Numeric	nmol/L	93688-0
PSY	36342	Reviewed By	Alphanumeric		18771-6
PSY	36343	Interpretation (PSY)	Alphanumeric		59462-2
GPSY	62236	Glucopsychosine	Numeric	nmol/mL	92752-5
GPSY	36344	Reviewed By	Alphanumeric		18771-6
GPSY	36345	Interpretation (GPSY)	Alphanumeric		59462-2
OXYBS	36762	Interpretation (OXYBS)	Alphanumeric		59462-2
OXYBS	36760	Cholestane-3beta,5alpha,6beta-triol	Numeric	nmol/mL	92757-4
OXYBS	36761	Lyso-sphingomyelin	Numeric	nmol/mL	92749-1
OXYBS	36763	Reviewed By	Alphanumeric		18771-6
LPCBS	34865	Interpretation (LPCBS)	Alphanumeric		59462-2
LPCBS	34860	C20 Lysophosphatidylcholine	Numeric	nmol/mL	90920-0

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
LPCBS	34861	C22 Lysophosphatidylcholine	Numeric	nmol/mL	90921-8
LPCBS	34862	C24 Lysophosphatidylcholine	Numeric	nmol/mL	90922-6
LPCBS	34863	C26 Lysophosphatidylcholine	Numeric	nmol/mL	90923-4
LPCBS	34864	Reviewed By	Alphanumeric		18771-6
PDBS	602290	Interpretation	Alphanumeric		59462-2
PDBS	602300	Reviewed By	Alphanumeric		18771-6
LGBBS	BA4368	Interpretation (LGBBS)	Alphanumeric		59462-2
LGBBS	BA4367	Globotriaosylsphingosine	Numeric	nmol/mL	92754-1
LGBBS	BA4369	Reviewed By	Alphanumeric		18771-6

Reference Values:

Disease	Marker	Normal range
Gaucher	Acid beta-glucosidase	> or =1.75 nmol/mL/hr
Niemann-Pick A/B	Sphingomyelinase	> or =2.5 nmol/mL/hr
Pompe	Acid alpha-glucosidase	> or =3.0 nmol/mL/hr
Krabbe	Galactocerebrosidase	> or =0.4 nmol/mL/hr
Fabry	Alpha-galactosidase	> or =2.00nmol/mL/hr
MPS I	Alpha-L-iduronidase	> or =1.5 nmol/mL/hr
MPS II	Iduronate 2-sulfatase	> or =4.0 nmol/mL/hr
NA	C20 Lysophosphatidylcholine	< or =1.81 nmol/mL
NA	C22 Lysophosphatidylcholine	< or =0.43 nmol/mL
ALD/PBD/ALDH	C24 Lysophosphatidylcholine	< or =0.49 nmol/mL
ALD/PBD/ALDH	C26 Lysophosphatidylcholine	< or =0.47 nmol/mL