

Lysosomal/Peroxisomal Disorders Screen, Blood Spot

Test ID: PLSD

Explanation: On the effective date, the following test changes will be made.

Current Reference Value		
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 =2.75 nmol/mL/hr
MPS I	Alpha-L-iduronidase	> or =2.0 nmol/mL/hr
NA	C20 Lysophosphatidylcholine	< or =1.00 mcg/mL
NA	C22 Lysophosphatidylcholine	< or =0.25 mcg/mL
ALD/PBD/ALDH	C24 Lysophosphatidylcholine	< or =0.30 mcg/mL
ALD/PBD/ALDH	C26 Lysophosphatidylcholine	< or =0.30 mcg/mL

New Reference Value		
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 =2.00 nmol/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

Current Specimen Required
Supplies: Card-Blood Spot Collection (Filter Paper) (T493) 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, EDTA, or heparin and dried on acceptable filter paper

New Specimen Required
Supplies: Card-Blood Spot Collection (Filter Paper) (T493) 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

Current Specimen Stability			
Specimen	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	56 days	FILTER PAPER
	Ambient	7 days	FILTER PAPER
	Frozen	56 days	FILTER PAPER

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

Current Result Codes	
34811	Acid Beta-Glucosidase
34812	Sphingomyelinase
34813	Acid Alpha-Glucosidase
34814	Galactocerebrosidase
34815	Alpha-Galactosidase
34816	Alpha-L-Iduronidase
34817	C20 Lysophosphatidylcholine
34818	C22 Lysophosphatidylcholine
34819	C24 Lysophosphatidylcholine
34820	C26 Lysophosphatidylcholine
34821	Interpretation (PLSD)
34822	Reviewed By

New Result Codes	
34811	Acid Beta-Glucosidase
34812	Sphingomyelinase
34813	Acid Alpha-Glucosidase
34814	Galactocerebrosidase
34815	Alpha-Galactosidase
34816	Alpha-L-Iduronidase
620785	Iduronate 2-Sulfatase
34817	C20 Lysophosphatidylcholine
34818	C22 Lysophosphatidylcholine
34819	C24 Lysophosphatidylcholine
34820	C26 Lysophosphatidylcholine
34821	Interpretation (PLSD)
34822	Reviewed By

Current Reject Due To	
Blood spot specimen that shows serum rings or has multiple layers	Reject
Thawing	Cold OK; Warm OK
Insufficient specimen	Reject
Incubated/exposed to temperatures above 37 degrees C	Reject

New Reject Due To	
Blood spot specimen that shows serum rings or has multiple layers	Reject
Thawing	Cold OK; Warm OK
Insufficient specimen	Reject
Specimens known to be exposed to elevated temperatures above ambient	Reject

Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.