

Patient Name SAMPLEREP,PLSD	Patient ID SA00052376	Age 8M	Gender F	Order # SA00052376
Ordering Phys CLIENT,CLIENT				DOB 04/16/2012
Client Order # SA00052376	Account Information			Report Notes
Collected 12/27/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 02/15/2013 13:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Lysosomal/Peroxisomal D/O Scrn, BS			REPORTED	12/28/2012 10:49	
Acid Beta-Glucosidase		6.7	nmol/mL/h	>=3.0	MCR
Sphingomyelinase		12.4	nmol/mL/h	>=5.5	MCR
Acid Alpha-Glucosidase		9.9	nmol/mL/h	>=4.0	MCR
Galactocerebrosidase		1.6	nmol/mL/h	>=0.4	MCR
Alpha-Galactosidase		8.2	nmol/mL/h	>=4.0	MCR
Alpha-L-Iduronidase		3.6	nmol/mL/h	>=2.0	MCR
C20 Lysophosphatidylcholine		0.13	mcg/mL	<=0.26	MCR
C22 Lysophosphatidylcholine		0.06	mcg/mL	<=0.11	MCR
C24 Lysophosphatidylcholine		0.02	mcg/mL	<0.10	MCR
C26 Lysophosphatidylcholine		0.23	mcg/mL	<=0.30	MCR

Interpretation (PLSD)

In this sample, the activities of the following enzymes (acid beta-glucosidase, acid alpha-glucosidase, sphingomyelinase, galactocerebrosidase, alpha-galactosidase, and alpha-L-iduronidase) are normal indicating that this patient is not affected with the lysosomal storage disorders caused by deficiencies in these enzymes. In addition, the concentrations of C20 to C26 lysophosphatidylcholine species are normal. Please note that this assay does not rule out all known lysosomal storage and peroxisomal disorders and is not designed to determine carrier status. Enzyme levels may be normal in patients receiving enzyme replacement therapy or who have undergone bone marrow transplantation. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions or to determine if additional testing is warranted.

Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS) Laboratory developed test using analyte specific reagents. This test was developed, validated and its performance characteristics determined by Mayo Clinic. It has not been cleared or approved by the U.S. Food and Drug Administration.

Reviewed By

Jennifer Peterson

MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name SAMPLEREP,PLSD	Collection Date and Time 12/27/2012 13:00	Report Status Final
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* Report times for Mayo performed tests are CST/CDT