

Patient ID SA00166982	Patient Name TESTING, PLSD_NORMAL	Birth Date 2001-01-01	Sex F	Age 23
Order Number SA00166982	Client Order Number SA00166982	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 06 Mar 2024 00:00		

Lysosomal/Peroxisomal D/O Scrn, BS

Acid Beta-Glucosidase

1.75 nmol/mL/h

MCR

Reference Value
≥1.75

Sphingomyelinase

2.5 nmol/mL/h

MCR

Reference Value
≥2.5

Acid Alpha-Glucosidase

3.0 nmol/mL/h

MCR

Reference Value
≥3.0

Galactocerebrosidase

0.4 nmol/mL/h

MCR

Reference Value
≥0.4

Alpha-Galactosidase

2.00 nmol/mL/h

MCR

Reference Value
≥2.00

Alpha-L-Iduronidase

1.5 nmol/mL/h

MCR

Reference Value
≥1.5

Iduronate 2-Sulfatase

4.0 nmol/mL/h

MCR

Reference Value
≥4.0

C20 Lysophosphatidylcholine

1.81 nmol/mL

MCR

Reference Value
≤1.81

C22 Lysophosphatidylcholine

0.43 nmol/mL

MCR

Reference Value
≤0.43

C24 Lysophosphatidylcholine

0.49 nmol/mL

MCR

Reference Value
≤0.49

C26 Lysophosphatidylcholine

0.47 nmol/mL

MCR

Reference Value
≤0.47

Reviewed By

Amy Breitlow

MCR

Interpretation (PLSD)

1 MCR

NEGATIVE In this sample, the activities of the following enzymes (acid beta-glucosidase, acid alpha-glucosidase, sphingomyelinase, galactocerebrosidase, alpha-galactosidase, alpha-L-iduronidase, and iduronate-2-sulfatase) 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.

ADDITIONAL INFORMATION

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

Received: 06 Mar 2024 07:08

Reported: 06 Mar 2024 07:16

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292