MAYO CLINIC LABORATORIES

1-800-533-1710

PLSD

Lysosomal and Peroxisomal Disorders Screen, Blood Spot

Patient ID	Patient Name		Birth Date	Sex	Age
SA00166983	TESTING, PLSD_ABNORMAL		2002-02-02	F	22
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00166983	SA00166983	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		06 Mar 2024 07:04			

Lysosomal/Peroxisomal D/O Scrn, BS

Acid Beta-Glucosidase — MCR
1.75 nmol/mL/h
Reference Value ≥1.75

Sphingomyelinase — MCR
2.5 nmol/mL/h
Reference Value ≥2.5

Acid Alpha-Glucosidase — MCR
3.0 nmol/mL/h
Reference Value ≥3.0

Galactocerebrosidase — MCR
0.4 nmol/mL/h
Reference Value ≥0.4

Alpha-Galactosidase — MCR
2.00 nmol/mL/h
Reference Value ≥2.00

Alpha-L-Iduronidase — MCR
1.5 nmol/mL/h
Reference Value ≥1.5

Iduronate 2-Sulfatase — MCR
2.7 nmol/mL/h
Low
Reference Value ≥4.0

C20 Lysophosphatidylcholine — MCR
1.81 nmol/mL
Reference Value ≤1.81

C22 Lysophosphatidylcholine — MCR
0.43 nmol/mL
Reference Value ≤0.43

C24 Lysophosphatidylcholine — MCR
0.49 nmol/mL
Reference Value ≤0.49

C26 Lysophosphatidylcholine — MCR
0.47 nmol/mL
Reference Value ≤0.47

Reviewed By — MCR
Amy Breitlow

Interpretation (PLSD) — 1 MCR

POSITIVE In this sample, the activity of iduronate-2-sulfatase is reduced indicating that this individual is a carrier for Mucopolysaccharidosis type II (MPS II). The other enzymes tested by this method were normal (acid beta-glucosidase, acid alpha-glucosidase, sphingomyelinase, galactocerebrosidase, and alpha-L-iduronidase) as well as the concentrations of C20 to C26 lysophosphatidylcholine species. For confirmation, consider ordering molecular genetic analysis of the IDS gene (MCL test MPS2Z). If not already ordered, this could be performed on the existing sample by calling MCL (1–800–533–1710) within one month. Please note that this enzyme's activity can also be reduced in Multiple Sulfatase Deficiency (MSD). If clinically indicated, consider biochemical genetic testing of other sulfatases (MCL test MSDW) or molecular genetic testing of the SUMF1 gene (MCL test CGPH, gene list ID: IEMCP-PCUBX1) to exclude MSD. Please contact the Biochemical Genetics consultant or genetic counselor on call (1–800–533–1710) if you have any questions.

ADDITIONAL INFORMATION

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

Received: 06 Mar 2024 07:09 **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