



Patient ID SA00123643	Patient Name TESTINGRNV, ASMW SAMPLE A	Birth Date 2019-06-01	Gender F	Age 4 M
Order Number SA00123643	Client Order Number SA00123643	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 16 Oct 2019 15:30		

Acid Sphingomyelinase, Leukocytes

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Low

0.20 nmol/h/mg Prot

MCR

Reference Value
≥0.32

Reviewed By

Marie Quade

MCR

Received: 17 Oct 2019 11:30

Reported: 17 Oct 2019 13:19

Interpretation

1 MCR

In this sample, the activity of sphingomyelinase (ASM) was reduced indicating that this individual is affected with Niemann-Pick disease, Type A or B. However, because pseudodeficient ASM activity cannot be excluded, consider measurement of lysosphingomyelin in blood (MCL test OXNP) and molecular genetic analysis of the SMPD1 gene (MCL test NPABZ; could be performed on existing specimen by calling MCL within one month). 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)

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