

Patient ID SA00129560	Patient Name SAMPLEREPORT, PSY ABN	Birth Date 2020-06-06	Gender F	Age 1 D
Order Number SA00129560	Client Order Number SA00129560	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Jun 2020 12:00		

Psychosine, BS

Interpretation (PSY)

1 MCR

POSITIVE

In this sample, the concentration of psychosine was elevated at a level that has been observed in late-onset Krabbe disease, symptomatic patients with long-standing Krabbe disease, and in patients with atypical Krabbe disease due to saposin A cofactor deficiency caused by mutations in the prosaposin gene, PSAP. We recommend determination of galactocerebrosidase (GALC) activity in leukocytes (MCL test ID: GALCW) and for future patient monitoring, psychosine in red blood cells (MCL test ID: PSYR). If GALC activity is normal, consider molecular genetic analysis of PSAP. Please contact the Biochemical Genetics consultant on call (Tel. 1-800-533-1710) if you have any questions. Please contact the Biochemical Genetics consultant on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Psychosine



2.1 nmol/L

MCR

Reference Value
<2.0

Reviewed By

Colleen Landon

MCR

Received: 09 Jun 2020 08:17

Reported: 09 Jun 2020 08:25

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

Report Status: Final