

Patient ID SA00121299	Patient Name TESTINGRNV, SAMPLEREPOR	Birth Date 1982-06-29	Gender F	Age 36
Order Number SA00121299	Client Order Number SA00121299	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 25 Jun 2019 13:00		

Psychosine, RBC

Interpretation (PSYR)

ⓘ 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: CBGC/Galactocerebrosidase, WBC). If GALC activity is normal, consider molecular genetic analysis of the PSAP. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

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

Psychosine, RBC


15 pmol/g Hb


MCR

Reference Value
 <10

Reviewed By

Amy Breitlow

MCR

Received: 27 Jun 2019 06:48

Reported: 27 Jun 2019 07:02

Laboratory Notes

- ⓘ 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
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905