

Patient ID SA00154836	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 1982-06-29	Sex F	Age 40
Order Number SA00154836	Client Order Number SA00154836	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Oct 2022 13:00		

Psychosine, RBC

Interpretation (PSYR)

1 MCR

POSITIVE

In this sample, the concentration of psychosine was elevated. This result is consistent with Krabbe disease due to galactocerebrosidase (GALC) deficiency or atypical Krabbe disease due to saposin A cofactor deficiency caused by mutations in the prosaposin gene, PSAP. We recommend determination of GALC activity in leukocytes (MCL test ID: GALCW). 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.

ADDITIONAL INFORMATION

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

Psychosine, RBC

MCR



High

17 pmol/g Hb

Reference Value
<5

Reviewed By

MCR

Amy Breitlow

Received: 13 Oct 2022 14:07

Reported: 13 Oct 2022 14:21

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