MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GALCK	SA00047537	14	M	SA00047537
Ordering Phys				**DOB** 06/05/1998
Client Order # SA00047537	**Account Information**			**Report Notes**
Collected 07/10/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/24/2012 15:47				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Krabbe Disease, Known Mutation			REPORTED 07/10/2012 15:56		
Specimen		Blood			MCR
Specimen ID		1038348			MCR
Order Date		10 Jul 2012 15:54			MCR
Reason For Referral					MCR

Family history of Krabbe disease. Test for the presence of mutations in the GAlC gene.

Method MCR

A PCR-based assay was used to test for the presence of the 30-kb deletion spanning exon 11 through the end of the GALC gene. Testing was performed for this specific mutation because it was previously identified in a family member of this individual. Mutation nomenclature is based on GenBank accession number; NM_000153.2.

Result MCR

The G286D mutation and 30 kb deletion spanning exon 11 were NOT detected.

Interpretation MCR

The absence of the two mutations previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population for being affected with Krabbe disease.

Errors in the information or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of the test results.

This assay does not rule out the presence of other mutations within the GALC gene.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available upon request.

CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GALCK	SA00047537	14	M	SA00047537
Ordering Phys				**DOB** 06/05/1998
Client Order # SA00047537	**Account Information**			**Report Notes**
Collected 07/10/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/24/2012 15:47				

Test	Flag	Results	Unit	Reference Value	Perform Site*
clinical findings, additional testing should be considered.					
Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Reviewed By					MCR
Melody Elizabeth Kimball					
Release Date		10 Jul 2012 15:55			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

* Report times for Mayo performed tests are CST/CDT