MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GALCS	SA00047310	11	M	SA00047310
Ordering Phys				**DOB** 06/04/2001
Client Order # SA00047310	**Account Information**			**Report Notes**
Collected 06/27/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/25/2012 13:43				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Krabbe Disease, Full Gene Analysis			REPORTED 06/27/2012 15:14		
Specimen		Blood			MCR
Specimen ID		1038320			MCR
Order Date		27 Jun 2012 07:26			MCR
Reason For Referral					MCR

Patient reported to have features suggestive of Krabbe disease. Test for the presence of mutations in the GALC gene.

Method MCR

DNA sequence analysis was used to test for the presence of mutations in all 17 exons of the GALC gene. Additionally, a PCR-based assay is utilized to test for the presence of a 30-kb deletion spanning exon 11 through the end of the GALC gene. Mutation nomenclature is based on GenBank accession number; NM_000153.2.

Result MCR

The following homozygous sequence change was detected:
Exon: 8
DNA change: c.857G>A
Amino Acid change: p.G286D (Gly286Asp)
This sequence change is a pathogenic mutation.

Interpretation MCR

This result is consistent with a diagnosis of Krabbe disease.

While age of onset and severity of disease is variable, the G286D mutation, when observed in the context of a second mutation, is associated with the non-infantile, late-onset phenotype.

Since mutations have been identified in the GALC gene, genetic testing of other at risk family members is possible.

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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

TEST SITE

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Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the 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		D Brian Dawson PhD			MCR
Release Date		27 Jun 2012 15:12			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,GALCS	06/27/2012	Final
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TEST SITE