

Laboratory Service Report

1-800-533-1710

Patient Name TESTINGRNV,GALCS	Patient ID SA00047310	Age 11	Gender M	Order # SA00047310	
Ordering Phys			•	DOB 06/04/2001	
Client Order # SA00047310	Account Information			Report Notes	
Collected 06/27/2012	3050 SUPERIOR DRI	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 07/25/2012 13:43	ROCHESTER,MN 559	001			

Test	Flag	Results	Unit	Reference Value	Perform Site*
Worlds Discours Full Come Analysis			DEDODEED AC	/07/2012 15:14	
Krabbe Disease, Full Gene Analysis		D1 1	REPORTED 06	/27/2012 15:14	1400
Specimen		Blood			MCR
Specimen ID		1038320			MCR
Order Date		27 Jun 2012 07:26			MCR
Reason For Referral					MCR
Patient reported to have fea					
disease. Test for the presen	nce of mutat	cions in the GALC			
gene.					
Method					MCR
DNA sequence analysis was us		_			
mutations in all 17 exons of	_	_ :	a		
PCR-based assay is utilized		_	_		
30-kb deletion spanning exor	_				
gene. Mutation nomenclature	e is based c	on GenBank accession			
number; NM_000153.2.					
Result					MCR
The following homozygous sec	quence chang	ge was detected:			
Exon: 8					
DNA change: c.857G>A					
Amino Acid change: p.G286D					
This sequence change is a pa	athogenic mu	itation.			
Interpretation					MCR
This result is consistent w	ith a diagno	sis of Krabbe			
disease.					
While age of onset and sever	ritu of diag	ago is revieble th			

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

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TESTINGRNV,GALCS	06/27/2012	Final			
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Test Flag Results Unit Value Site*

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 Lab Director: Franklin R. Cockerill, III, M.D.

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TESTINGRNV,GALCS	06/27/2012	Final
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