

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GALCK	SA00047555	36	M	SA00047555
Ordering Phys				DOB
				05/23/1976
Client Order #	Account Information			Report Notes
SA00047555				
Collected	C7028846-DLMP ROC	HESTER		
07/11/2012		3050 SUPERIOR DRIVE		
Printed	ROCHESTER,MN 5590	01		
07/24/2012 15:48				

Test	Flag	Results	Unit	Reference Value	Perform Site*
bbe Disease, Known Mutation			REPORTED 07,	/11/2012 14:30	
Specimen		Blood			MCF
Specimen ID		1038353			MCF
Order Date		11 Jul 2012 14:24			MCF
Reason For Referral					MCF
Family history of Krabbe dis mutations in the GAlC gene.	ease. Test f	for the presence of			
Method					MCR
DNA sequence analysis and a test for the presence of the and the 30-kb deletion spann the GALC gene. Testing was mutations because they were family member of this indivibased on GenBank accession n	p.G286D (c. ing exon 11 performed fo previously i dual. Mutat	.857G>A) in exon 8 through the end of or these specific dentified in a cion nomenclature is			
Result					MCR
The following heterozygous s Exon: 8 DNA change: c.857G>A Amino Acid change: p.G286D (This sequence change is a pa	Gly286Asp)				
THIS sequence change is a pa	chogenic muc	Lacion.			
The following heterozygous 3	Okb deletion	ı:			
Exon: 11-17 (through the end					
Interpretation	01 0110 90110	-,			MCR
This result is consistent wi disease.	th a diagnos	sis of Krabbe			
While age of onset and sever G286D mutation, when observe mutation, is associated with phenotype.	d in the cor	ntext of a second			
Since mutations have been id genetic testing of other at					
A genetic consultation may b	e of benefit	=.			
A list of common polymorphis	ms identifie	ed for this patient			

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status	
TESTINGRNV,GALCK	07/11/2012	Final	
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^{*} Report times for Mayo performed tests are CST/CDT

Test results should be interpreted in context of clinical

is available upon request.

CAUTIONS:



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SA00047555				
Collected	C7028846-DLMP ROC	CHESTER		
07/11/2012	3050 SUPERIOR DRIV	3050 SUPERIOR DRIVE		
Printed	ROCHESTER,MN 559	01		
07/24/2012 15:48				

Test Flag Results Unit Value Site*

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 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
Melody Elizabeth Kimball
Release Date

MCR

11 Jul 2012 14:28 MCR

* Performing Site:

MCR Mayo Clinic Laboratories - Rochester Main Campus Lab Director: Franklin R. Cockerill, III, M.D.

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