

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,NPDKMREPORTS	SA00047725	37	M	SA00047725
Ordering Phys				DOB 06/30/1975
Client Order # SA00047725	Account Information			Report Notes
Collected 07/16/2012	C7028846-DLMP ROC 3050 SUPERIOR DRIV	Έ		
Printed 08/03/2012 15:37	ROCHESTER,MN 5590	01		

Test	Flag	Results	Unit	Reference Value	Perform Site*
emann-Pick A-B, Known Mutation			REPORTED 0	7/17/2012 08:38	
Specimen		Blood			MCF
Specimen ID		1038384			MCF
Order Date		16 Jul 2012 14:22			MCF
Reason For Referral					MCF
Family history of Niemann	-Pick disease (t	ype A or B). Test			
for the presence of famil:	ial alterations	in the SMPD1 gene.			
Method					MCR
DNA sequence analysis was	used to test for	r the presence of			
the p.Q21X (c.61C>T) alter	ration in exon 1	of the SMPD1 gene			
(GenBank accession number		-			
specific alteration was pe	erformed because	it was identified			
in a family member.					
Result					MCR
The p.Q21X alteration was	NOT detected.				
Interpretation					MCR
Absence of the mutation(s					
affected family member in					
no greater risk than some	_				
a carrier of Niemann-Pick	disease (type A	or B).			

causing mutations in this gene or other genes associated with metabolic disease. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of test results.

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 clinical findings, additional testing should be considered.

Bone marrow transplants from allogenic donors will interfere

Performing Site Legend on Last Page of Report

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TESTINGRNV,NPDKMREPORTS	07/16/2012	Final
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^{*} Report times for Mayo performed tests are CST/CDT



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				06/30/1975
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Collected	C7028846-DLMP ROC	CHESTER		
07/16/2012	3050 SUPERIOR DRIVE			
Printed	ROCHESTER,MN 559	01		
08/03/2012 15:37				

Reference Perform Test Flag Results Unit Site* Value

with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Laboratory developed test. Reviewed By Emily Christine Lauer Release Date

17 Jul 2012 08:36

MCR MCR

* Performing Site:

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

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Page 2 of 2		** End of Report **

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