

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,NPD	Patient ID SA00007373	Age 32	Gender F	Order # SA00007373
Ordering Phys				DOB 04/04/1980
Client Order # SA00007373	Account Information			Report Notes
Collected 07/31/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 08/15/2012 14:14	ROCHESTER,MN 559	901		

Test	Flag	Results	Unit	Reference Value	Perform Site*
Niemann-Pick Disease, Types A ar	nd B		REPORTED 0	8/14/2012 17:01	
Specimen		Blood			MCR
Specimen ID		976472			MCR
Order Date		01 Aug 2012 13:09			MCR
Reason For Referral					MCR
Carrier screen for Niema					
Test for the presence of	f a mutation in	the SMPD1 gene.			
Method					MCR
A PCR-based assay was us		-			
mutations in the SMPD1 of deltaR608.	gene: L302P, R4	196L, fsP330, and			
Result					MCR
One copy of the L302P m	utation was iden	ntified.			
Interpretation					MCR
This result indicates th	nat this individ	dual is a carrier of			
Niemann-Pick disease (Ty	ype A or B). Th	nis interpretation			
assumes that this indivi	idual is healthy	and is not			
clinically affected with	n Niemann-Pick d	lisease.			
Since a mutation has bee	en identified, t	esting of at risk			
family members is possib	ole. If appropr	riate, carrier testing	J		
should be offered to the	is individual's	reproductive partner.			

A genetic consultation may be of benefit.

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 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 14 Aug 2012 16:59 MCR

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status		
SAMPLEREPORT,NPD	07/31/2012 13:00	Final		
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^{*} Report times for Mayo performed tests are CST/CDT



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* 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
SAMPLEREPORT.NPD	07/31/2012 13:00	Final
Page 2 of 2	07/01/2012 10:00	** End of Report **

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