MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,NPDMSREPORTS	SA00047726	26	M	SA00047726
Ordering Phys				**DOB** 12/24/1985
Client Order # SA00047726	**Account Information**			**Report Notes**
Collected 07/16/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/03/2012 15:46				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Niemann-Pick A-B Full Gene Analysis			REPORTED 07/17/2012 08:36		
Specimen		Blood			MCR
Specimen ID		1038387			MCR
Order Date		16 Jul 2012 14:23			MCR
Reason For Referral					MCR
Patient reported to have features suggestive of Niemann-Pick disease (type A or B). Test for the presence of mutations in the SMPD1 gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of a mutation in all 6 exons of the SMPD1 gene. Mutation nomenclature is based on GenBank accession number; NM_000543.3.					
Result					MCR
The following heterozygous sequence change was detected: Exon: 1 DNA change: c.61C>T Amino acid change: p.Q21X (Gln21X) This sequence change is a known pathogenic mutation.					
Interpretation					MCR

This result indicates that this individual is at minimum a carrier of Niemann-Pick disease (type A or B). This result also increases the likelihood of a diagnosis of Niemann-Pick disease (type A or B) but neither proves nor excludes it. We predict that there may be disease causing mutations in the SMPD1 gene not identifiable by the method described (e.g., large deletions/duplications, promoter mutations, or deep intronic mutations).

This assay does not rule out the presence of disease causing mutations in other genes associated with metabolic disease. This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. sphingomyelinase activity in leukocytes, dried blood spots, or skin fibroblasts).

Since a mutation has been identified in the SMPD1 gene, genetic testing of 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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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 Emily Christine Lauer		MCR
Release Date	17 Jul 2012 08:34	MCR

* Performing Site:

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

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