

Patient Name SAMPLEREP, NPD	Patient ID SA00007372	Age 32	Gender F	Order # SA00007372
Ordering Phys				DOB 04/04/1980
Client Order # SA00007372	Account Information			Report Notes
Collected 07/31/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER, MN 55901			
Printed 08/15/2012 14:13				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Niemann-Pick Disease, Types A and B			REPORTED 08/14/2012 17:01		
Specimen		Blood			MCR
Specimen ID		976471			MCR
Order Date		01 Aug 2012 13:09			MCR
Reason For Referral		Carrier screen for Niemann-Pick disease (types A and B). Test for the presence of a mutation in the SMPD1 gene.			MCR
Method		A PCR-based assay was used to test for the following mutations in the SMPD1 gene: L302P, R496L, fsP330, and deltaR608.			MCR
Result		None of the listed mutations were detected.			MCR
Interpretation		Having excluded the presence of the listed mutations, the risk for this individual to be a carrier of another Niemann-Pick disease type A mutation is approximately 1/2968. This risk assumes that there is no family history of Niemann-Pick disease. Additionally, this risk is based on a mutation detection rate of 97% (for Ashkenazi Jewish ancestry) and a population carrier frequency of 1/90. The risk for this individual to be a carrier of another Niemann-Pick disease type B mutation is reduced but not eliminated. Because there is little information about the carrier frequency and mutation detection rate for individuals of non-Ashkenazi Jewish ancestry, we are unable to provide a specific revised risk assessment for individuals of other ethnicities at this time. 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			MCR

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
marrow transplant.					
Laboratory developed test.					
Reviewed By:		D Brian Dawson PhD			MCR
Release Date		14 Aug 2012 16:59			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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