MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,TSD	SA00007375	32	F	SA00007375
Ordering Phys				**DOB** 04/04/1980
Client Order # SA00007375	**Account Information**			**Report Notes**
Collected 07/31/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/15/2012 14:15				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Tay-Sachs, Mutation Analysis			REPORTED 08/14/2012 17:14		
Specimen		Blood			MCR
Specimen ID		976474			MCR
Order Date		01 Aug 2012 13:09			MCR
Reason For Referral					MCR
Carrier screen for Tay-Sachs disease. Test for the presence of a mutation within the HEXA gene.					
Method					MCR
A PCR-based assay was used to test for the following mutations within the HEXA gene: 1278insTATC, G269S, IVS12(+1)G>C, IVS9(+1)G>A, delta7.6kb, and two pseudo-deficiency alleles (R247W and R249W).					
Result					MCR
One copy of the G269S mutation was identified.					
Interpretation					MCR
This finding indicates that this individual is a carrier of Tay-Sachs disease. This interpretation assumes that this individual is healthy and is not clinically affected with Tay-Sachs disease.					
Since a mutation has been identified, carrier testing of other at risk family members is possible. Additionally, carrier testing should be offered to the reproductive partner of this individual.					
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

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

TEST SITE

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Release Date		14 Aug 2012 17:12			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.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,TSD	07/31/2012	Final
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TEST SITE