MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,GAUW	SA00007368	32	F	SA00007368
Ordering Phys				**DOB** 04/04/1980
Client Order # SA00007368	**Account Information**			**Report Notes**
Collected 07/31/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/15/2012 14:10				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Gaucher Disease, Mutation Analysis			REPORTED 08/14/2012 16:57		
Specimen		Blood			MCR
Specimen ID		976467			MCR
Order Date		01 Aug 2012 13:07			MCR
Reason For Referral					MCR

Carrier screen for Gaucher disease. Test for the presence of a mutation in the GBA gene.

Method MCR

A PCR-based assay was used to test for the following mutations in the GBA gene: N370S, IVS2+1G>A, 84G>GG, R496H, L444P, delta55bp V394L, and D409H.

Result MCR

None of the listed mutations were detected.

Interpretation MCR

Having excluded the presence of the listed mutations, the risk that this individual is a carrier of another Gaucher disease mutation is approximately 1/341.

This risk assumes that there is no family history of Gaucher disease. Additionally, this calculation is based on a mutation detection rate of 95% (for Ashkenazi Jewish ancestry) and a population carrier frequency of 1/18.

There is considerable variability both in the carrier frequency and detection rate among different ethnic groups. The risk estimate provided is our best approximation given the information available to us for this patient and population.

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

Performing Site Legend on Last Page of Report

* 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 16:56			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,GAUW	07/31/2012 13:00	Final
Page 2 of 2		** End of Report **

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TEST SITE