MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GBAKM	SA00005805	8	F	SA00005805
Ordering Phys				**DOB** 05/30/2003
Client Order # SA00005805	**Account Information**			**Report Notes**
Collected 05/30/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 05/31/2012 09:21				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Gaucher Disease, Known Mutation			REPORTED 05/30/2012 09:33		
Specimen		Blood			MCR
Specimen ID		963444			MCR
Order Date		30 May 2012 08:24			MCR
Reason For Referral					MCR

Family history of Gaucher disease. Test for the presence of familial alterations in the GBA gene.

Method MCR

DNA sequence analysis was used to test for the presence of the p.N370S (c.1226A>G) alteration in exon 9 of the GBA gene (GenBank accession number; NM_ 000157.2). Analysis for this specific alteration was performed because it was identified in a family member.

Result MCR

The following heterozygous sequence change was detected:
Exon: 9
DNA change: c.1226A>G (AAC>AGC)
Amino acid change: p.N370S (Asn370Ser)
This sequence change is a known pathogenic mutation.

Interpretation MCR

This result indicates that this individual is a carrier of Gaucher disease. This interpretation assumes that this individual is clinically healthy and is not affected with Gaucher disease.

While the clinical presentation of Gaucher disease is variable, the N370S mutation is often associated with a lack of neurological manifestations for individuals with at least one copy of this mutation.

Since a mutation has been identified in the GBA gene, genetic testing of at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner to clarify their risk of having a child with Gaucher disease.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available upon request.

CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GBAKM	SA00005805	8	F	SA00005805
Ordering Phys				**DOB** 05/30/2003
Client Order # SA00005805	**Account Information**			**Report Notes**
Collected 05/30/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 05/31/2012 09:21				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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 Melody Elizabeth Trimble					MCR
Release Date		30 May 2012 09:32			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

* Report times for Mayo performed tests are CST/CDT