MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,GAAMS	SA00043812	45	F	SA00043812
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00043812	**Account Information**			**Report Notes**
Collected 02/28/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 02/29/2012 10:23				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Pompe Disease, Full Gene Sequencing			REPORTED 02/29/2012 10:06		
Specimen		Blood			MCR
Specimen ID		1037906			MCR
Order Date		29 Feb 2012 09:41			MCR
Reason For Referral					MCR

Carrier screening for Pompe disease. Test for the presence of a mutation within the GAA gene.

Method MCR

Fluorescent DNA sequence analysis was performed to test for the presence of mutations in all 19 coding exons of the GAA gene. GenBank accession number; NM_000152.3.

Result MCR

The following heterozygous sequence change was detected:
Exon: 14
DNA change: c.1941C>G
Amino Acid change: p.C647W (Cys647Trp)
This sequence change is a pathogenic mutation.

Interpretation MCR

This result indicates that this individual is a carrier of Pompe disease and may be at risk to have an affected child. This interpretation assumes that this individual is not clinically affected with Pompe disease.

Carrier screening for Pompe disease should be offered to this individual's reproductive partner if appropriate.

Since a mutation has been identified, testing of other 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.

CAUTIONS:
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.

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.

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*
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.					
Extraction Performed?		Yes			MCR
Reviewed By Emily Christine Lauer					MCR
Release Date		29 Feb 2012 10:04			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.

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