MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FABKM	SA00046666	45	F	SA00046666
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00046666	**Account Information**			**Report Notes**
Collected 05/21/2012 12:10	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/15/2012 11:40				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Fabry Disease Known Mutation			REPORTED 07/13/2012 10:03		
Specimen		Blood			MCR
Specimen ID		1038188			MCR
Order Date		21 May 2012 14:57			MCR
Reason For Referral					MCR

Family history of Fabry disease. Test for the presence of the familial alteration in the GLA gene.

Method MCR

DNA sequence analysis was used to test for the presence of the p.R227X (c.679C>T) alteration in exon 5 of the GLA gene. Analysis for this specific alteration was performed because it was previously identified in a family member. Mutation nomenclature is based on GenBank accession number NM_000169.2.

Result MCR

The following heterozygous sequence change was detected:
Exon: 5
DNA change: c.679C>T
Amino Acid change: p.R227X (Arg227X)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

The presence of a mutation previously identified in a family member indicates that this individual is a carrier of Fabry disease. Studies suggest that female carriers of GLA mutations may be at risk for developing symptoms associated with Fabry or variant Fabry disease.
Since a mutation has been identified in the GLA gene, genetic testing of at risk family members is possible.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are 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.

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*
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.					
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					MCR
Melody Elizabeth Kimball					
Release Date		13 Jul 2012 09:59			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.

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