MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FABMS	SA00046667	45	F	SA00046667
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00046667	**Account Information**			**Report Notes**
Collected 05/20/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/15/2012 11:55				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Fabry Disease Full Gene Analysis		REPORTED 07/13/2012 10:03			
Specimen		Blood			MCR
Specimen ID		1038189			MCR
Order Date		21 May 2012 14:57			MCR
Reason For Referral					MCR
Patient reported to have features of Fabry or variant Fabry disease. Test for the presence of a mutation in the GLA gene.					
Method					MCR
Bi-directional sequence analysis was used to test for the presence of a mutation in all coding regions and intron/exon boundaries of the GLA gene. Mutation nomenclature is based on GenBank accession number NM_000169.2.					
Result					MCR
A mutation was NOT detected.					
Interpretation					MCR
This result decreases the likelihood, but does not rule out the diagnosis of Fabry disease or variant Fabry disease. We predict that there may be disease causing mutations in the GLA gene that are not identified by the method described (e.g., large deletions/duplications, promoter mutations, or deep intronic mutations). This assay does not rule out the presence of disease causing mutations in other genes associated with metabolic disease.					
This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g., alpha-galactosidase activity in leukocytes and ceramide trihexoside/sulfatide accumulation in urine sediment).					
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: 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

TEST SITE TEST SITE

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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					MCR
Melody Elizabeth Kimball					
Release Date		13 Jul 2012 10:00			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

TEST SITE