

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,FABMS	Patient ID SA00046668	Age 45	Gender F	Order # SA00046668
Ordering Phys		,		DOB 06/10/1966
Client Order # SA00046668	Account Information			Report Notes
Collected 05/20/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 09/15/2012 11:57	ROCHESTER,MN 55901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
-555	5		J		2200
Fabry Disease Full Gene Analysis			REPORTED 07/	13/2012 10:03	
Specimen		Blood			MCR
Specimen ID		1038190			MCR
Order Date		21 May 2012 14:57			MCR
Reason For Referral					MCR
Patient reported to have fea	atures of Fa	abry or variant Fabry			
disease. Test for the prese	ence of a mu	tation in the GLA			
gene.					
Method					MCR
Bi-directional sequence ana					
presence of a mutation in a					
boundaries of the GLA gene.					
on GenBank accession number	NM_000169.2	2.			
Result					MCR
The following heterozygous :	sequence cha	inge was detected:			
Exon: 5					
DNA change: c.679C>T					
Amino Acid change: p.R227X	(Arg227X)				
Classification: DELETERIOUS					
Interpretation					MCR
This alteration is a known o	deleterious	mutation.			

This result indicates that this individual is a carrier of Fabry disease, and is consistent with symptoms reported for this individual. Studies suggest that female carriers of GLA mutations may be at risk for developing symptoms associated with Fabry or variant Fabry disease. There is a 50% chance that a male child of this individual will be

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for Fabry disease is available at Mayo Medical Laboratories by ordering FABKM/88266 Fabry Disease Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

A genetic consultation may be of benefit.

affected with Fabry disease.

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.

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, FABMS	05/20/2012	Final
Page 1 of 2		>> Continued on Next Page >>

^{*} Report times for Mayo performed tests are CST/CDT



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Printed 09/15/2012 11:57	ROCHESTER,MN 559	901		

Test Flag Results Unit Value Site*

CAUTIONS:

Release Date

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 marrow transplant.

Laboratory developed test. Reviewed By Melody Elizabeth Kimball

MCR

 ${\tt MCR}$

* Performing Site:

MCR Mayo Clinic Laboratories - Rochester Main Campus Lab Director: Franklin R. Cockerill, III, M.D.

13 Jul 2012 10:00

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, FABMS	05/20/2012	Final
Page 2 of 2		** End of Report **

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