

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,GAAKM	Patient ID SA00043814	Age 45	Gender F	Order # SA00043814
Ordering Phys		1.5		DOB 06/10/1966
Client Order # SA00043814	Account Information	l		Report Notes
Collected 02/28/2012	C7028846-DLMP RO 3050 SUPERIOR DRI	VE		
Printed 02/29/2012 10:22	ROCHESTER,MN 559	901		

		·	·	·	
				Reference	Perform
Test	Flag	Results	Unit	Value	Site*
Pompe Disease, Known Mutation			REPORTED 0	2/29/2012 10:08	
Specimen		Blood			MCR
Specimen ID		1037908			MCR
Order Date		29 Feb 2012 09:56			MCR
Reason for Referral					MCR
Family history of Pompe dis	ease. Test	for the presence of			
the familial alteration(s)	in the GAA g	gene.			
Method					MCR
DNA sequence analysis was u	sed to test	for the presence of			
the p.C647W (c.1941C>G) alt	eration in e	exon 14 of the GAA			
gene (GenBank accession num	ber; NM_0001	.52.3). Analysis for			
this specific alteration wa	-	because it is known			
to be present in a family m	ember.				
Result					MCR
The following heterozygous	sequence cha	inge was detected:			
Exon: 14					
DNA change: c.1941C>G					
Amino Acid change: p.C647W					
This sequence change is a p	athogenic mu	ıtation.			
Interpretation					MCR
This result indicates that					
Pompe disease and may be at					
This interpretation assumes					
clinically affected with Po	_				
assumes that the alteration	(s) listed i	n the methods was			

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

Since a GAA mutation has been identified, testing of other at risk family members is possible.

A genetic consultation may be of benefit.

previously identified in a family member.

This assay does not rule out the presence of other alterations within this gene or within other genes that are associated with metabolic disease. Errors in the diagnosis or pedigree provided to us, including non-paternity, may lead to an erroneous interpretation of test results.

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

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,GAAKM	02/28/2012	Final
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MCR

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Reference Perform Flag Results Unit Value Site* Test

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. ${\tt Misinterpretation}\ {\tt of}\ {\tt results}\ {\tt may}\ {\tt occur}\ {\tt if}\ {\tt the}\ {\tt information}$ 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.

Extraction Performed? Reviewed By

Release Date 29 Feb 2012 10:05

Emily Christine Lauer MCR

Yes

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