

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT, HMBSS	SA00048842	11	М	SA00048842
Ordering Phys		•	•	DOB
-				06/15/2001
Client Order #	Account Information		Report Notes	
SA00048842				
Collected	C7028846-DLMP RO	CHESTER		
09/05/2012	3050 SUPERIOR DRI	3050 SUPERIOR DRIVE		
Printed	ROCHESTER,MN 559	01		
10/05/2012 16:38				

Test	Flag	Results	Unit	Reference Value	Perform Site*
HMBS Gene, Full Gene Analysis			REPORTED 0	9/07/2012 11:59	
Specimen		Blood			MCR
Specimen ID		1038746			MCR
Order Date		05 Sep 2012 13:24			MCR
Reason For Referral					MCR
Possible diagnosis of acute into Test for the presence of a muta					
Method		are mad gene.			MCR
Bi-directional sequence analysi	is was per	formed to test for			11010
the presence of a mutation in a	_				
intron/exon boundaries of the I	_	_			
nomenclature is based on GenBar					
NM_000190.3.					
Result					MCR
The following heterozygous sequ	uence char	nge was detected:			
Exon: 3					
DNA change: c.77G>A					
Amino acid change: p.R26H (Arg	26His)				
Classification: DELETERIOUS					
Interpretation					MCR
This alteration is a known dele	eterious m	nutation.			

This result is consistent with a diagnosis of AIP due to $\ensuremath{\mathsf{HMBS}}$ deficiency.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for AIP due to HMBS deficiency is available at Mayo Medical Laboratories by ordering HMBSK/61217 HMBS Gene, 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.

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

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status	
SAMPLEREPORT, HMBSS	09/05/2012	Final	
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^{*} Report times for Mayo performed tests are CST/CDT



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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.

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
Release Date

Devin Oglesbee PhD 07 Sep 2012 11:57

MCR 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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