

### **Laboratory Service Report**

### 1-800-533-1710

Patient Name SAMPLEREPORT, MAHMS N	Patient ID SA00057882	<b>Age</b> 46	Gender F	<b>Order #</b> SA00057882
Ordering Phys CLIENT,CLIENT		>	•	<b>DOB</b> 06/10/1966
Client Order # SA00057882	Account Information			Report Notes
<b>Collected</b> 05/29/2013 00:00	3050 Superior Drive			
<b>Printed</b> 06/03/2013 10:51	Rochester, MN 55901			

Test	Flag Results	Unit	Reference Value	Perform Site*
MMACHC Gene, Full Gene Analysis				
<b>RECEIVED:</b> 05/30/2013 16:47 <b>REPO</b>	RTED: 06/03/2013 09:52			
Specimen	Blood			MCR
Specimen ID	1061992			MCR
Order Date	03 Jun 2013 08:1	L6		MCR
Reason For Referral				MCR
Possible diagnosis of me homocystinuria, cobalami presence of mutations in	n C type (MMACHC). Test for the	e		
Method				MCR
Bi-directional sequence the presence of mutation intron/exon boundaries o	analysis was performed to test of some solutions and for the MMACHC gene. Mutation GenBank accession number	for		
Result				MCR
A mutation was NOT detec	ted.			11011
Interpretation				MCR
This result decreases th the diagnosis of MMACHC that some individuals wh	e likelihood, but does not rule for this individual. We predict o have a diagnosis of MMACHC may ations in the MMACHC gene not od described (e.g. large	t		
2 2 4 4 7 2 4 4 4				

This result should be interpreted in light of clinical findings, family history, and other laboratory data (e.g. analysis of total plasma homocysteine, urine organic acids, or complementation studies of skin fibroblasts).

deletions/duplications, promoter mutations, or deep intronic

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:

mutations).

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\*\*\*

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Patient Name	Collection Date and Time	Report Status	
SAMPLEREPORT, MAHMS N	05/29/2013 00:00	Final	
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<sup>\*</sup> Report times for Mayo performed tests are CST/CDT



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Test Flag Results Unit Value Site\*

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.

Extraction Performed?
Reviewed By
Release Date

Yes. Jessica Rose Chavey 03 Jun 2013 09:51 MCR 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.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, MAHMS N	05/29/2013 00:00	Final
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