

<b>Patient Name</b> SAMPLEREP,MAHMS A	<b>Patient ID</b> SA00058670	<b>Age</b> 46	<b>Gender</b> F	<b>Order #</b> SA00058670
<b>Ordering Phys</b> CLIENT,CLIENT			<b>DOB</b> 06/10/1966	
<b>Client Order #</b> SA00058670	<b>Account Information</b> C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			<b>Report Notes</b>
<b>Collected</b> 06/02/2013 00:00				
<b>Printed</b> 06/03/2013 14:07				

Test	Flag	Results	Unit	Reference Value	Perform Site*
<b>MMACHC Gene, Full Gene Analysis</b>					
<b>RECEIVED:</b> 06/03/2013 12:04 <b>REPORTED:</b> 06/03/2013 13:56					
Specimen		Blood			MCR
Specimen ID		1062008			MCR
Order Date		03 Jun 2013 13:15			MCR
Reason For Referral		Patient reported to have features suggestive of methylmalonic aciduria and homocystinuria, cobalamin C type (MMACHC). Test for the presence of mutations in the MMACHC gene.			MCR
Method		Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the MMACHC gene. Mutation nomenclature is based on GenBank accession number NM_015506.2.			MCR
Result		The following homozygous duplication was detected: Exon: 2 DNA change: c.271dupA Amino acid change: p.R91KfsX14 (Arg91LysfsX14) Classification: DELETERIOUS			MCR
Interpretation		This alteration is a known deleterious mutation.  This finding is consistent with a diagnosis of MMACHC.  Since mutations have been identified, testing of at risk family members is possible. Mutation-specific testing for MMACHC is available at Mayo Medical Laboratories by ordering MAHKM/89135 MMACHC 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			MCR

\*\*\*Performing Site Legend on Last Page of Report\*\*\*

<b>Patient Name</b> SAMPLEREP,MAHMS A	<b>Collection Date and Time</b> 06/02/2013 00:00	<b>Report Status</b> Final
Page 1 of 2	>> Continued on Next Page >>	

\* Report times for Mayo performed tests are CST/CDT

<b>Patient Name</b> SAMPLEREP,MAHMS A	<b>Patient ID</b> SA00058670	<b>Age</b> 46	<b>Gender</b> F	<b>Order #</b> SA00058670
<b>Ordering Phys</b> CLIENT,CLIENT				<b>DOB</b> 06/10/1966
<b>Client Order #</b> SA00058670	<b>Account Information</b>			<b>Report Notes</b>
<b>Collected</b> 06/02/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
<b>Printed</b> 06/03/2013 14:07				

Test	Flag	Results	Unit	Reference Value	Perform Site*
<p>or positive results. If results obtained do not match the clinical findings, additional testing should be considered.</p> <p>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.</p> <p>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.</p> <p>Laboratory developed test.</p>					
Extraction Performed?		Yes.			MCR
Reviewed By		Jessica Rose Chavey			MCR
Release Date		03 Jun 2013 13:54			MCR

\* Performing Site:

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

<b>Patient Name</b> SAMPLEREP,MAHMS A	<b>Collection Date and Time</b> 06/02/2013 00:00	<b>Report Status</b> Final
Page 2 of 2		** End of Report **

\* Report times for Mayo performed tests are CST/CDT