MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MAHKM A	SA00057881	46	F	SA00057881
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00057881	**Account Information**			**Report Notes**
Collected 05/29/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/03/2013 10:51				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MMACHC Gene, Known Mutation					
RECEIVED: 05/30/2013 16:21 **REPORTED:** 06/03/2013 09:52					
Specimen		Blood			MCR
Specimen ID		1061991			MCR
Order Date		03 Jun 2013 08:15			MCR
Reason For Referral					MCR
Family history of methylmalonic aciduria and homocystinuria, cobalamin C type (MMACHC). Test for the presence of familial alteration(s) in the MMACHC gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of the p.R91KfsX17 (c.271dupA) alteration in exon 2 of the MMACHC gene. Analysis for this specific alteration was performed because it has been identified in a family member. Mutation nomenclature is based on GenBank accession number NM_015506.2.					
Result					MCR
The following heterozygous duplication was detected: Exon: 2 DNA change: c.271dupA Amino acid change: p.R91KfsX17 (Arg91LysfsX14) Classification: DELETERIOUS					
Interpretation					MCR
This alteration is a known deleterious mutation. This result indicates that this individual is a carrier of MMACHC. This interpretation assumes that this individual is clinically healthy and is not affected with MMACHC. This assay does not rule out the presence of other disease causing mutations in this gene or 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. Since an MMACHC mutation has been identified, testing of at risk family members is possible. 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					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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available upon request.

CAUTIONS:
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.

Extraction Performed?	Yes.		MCR
Reviewed By	Jessica Rose Chavey		MCR
Report Date	03 Jun 2013 09:49		MCR

* Performing Site:

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