MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,MHDMS	SA00048201	8	M	SA00048201
Ordering Phys				**DOB** 06/15/2004
Client Order # SA00048201	**Account Information**			**Report Notes**
Collected 08/02/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/29/2012 10:07				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MMADHC Gene, Full Gene Analysis			REPORTED 08/06/2012 17:01		
Specimen		Blood			MCR
Specimen ID		1038532			MCR
Order Date		02 Aug 2012 10:34			MCR
Reason For Referral					MCR

Patient reported to have features suggestive of methylmalonic aciduria and homocystinuria, cblD type, (MMADHC). Test for the presence of mutations within the MMADHC gene.

Method MCR

Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the MMADHC gene (GenBank accession number; NM 015702.2).

Result MCR

The following homozygous sequence change was detected:
Exon: 8
DNA change: c.748C>T
Amino acid change: p.R250X (Arg250X)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of methylmalonic aciduria and homocystinuria, cblD type, (MMADHC).

Since mutations have been identified, genetic testing of other at risk family members is possible. Mutation-specific testing for disease is available at Mayo Medical Laboratories by ordering MHDKM/61098 MMADHC 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:
Test results should be interpreted in context of clinical

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* Report times for Mayo performed tests are CST/CDT

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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.

Reviewed By	Devin Oglesbee PhD	MCR
Release Date	06 Aug 2012 17:00	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.