

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,MHDMS	SA00048200	12	M	SA00048200
Ordering Phys		•		DOB
				06/15/2000
Client Order #	Account Information			Report Notes
SA00048200				
Collected	C7028846-DLMP ROCHESTER			
08/02/2012	3050 SUPERIOR DRIVE			
Printed	ROCHESTER,MN 559	01		
08/29/2012 10:06				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MADHC Gene, Full Gene Analysis			REPORTED (08/06/2012 17:01	
Specimen		Blood			MCF
Specimen ID		1038531			MCF
Order Date		02 Aug 2012 10:34			MCF
Reason For Referral					MCF
Patient reported to have fe methylmalonic aciduria and (MMADHC). Test for the pre MMADHC gene.	homocystinuri	a, cblD type,			
Method					MCF
Bi-directional sequence ana the presence of mutations i intron/exon boundaries of t number; NM 015702.2).	n all coding	regions and			
Result					MCF
A mutation was NOT detected					
Interpretation					MCF
These results decrease the the diagnosis of methylmalo cblD type, (MMADHC) for thi some individuals who have a disease causing mutations widentified by the method de	nic aciduria s individual. diagnosis of ithin the MMA	and homocystinuria, We predict that MMADHC may have DHC gene not			

These results should be interpreted in the context 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).

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

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,MHDMS	08/02/2012	Final
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^{*} Report times for Mayo performed tests are CST/CDT



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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 Release Date Devin Oglesbee PhD 06 Aug 2012 17:00

MCR MCR

* Performing Site:

MCR Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester MN 55905 Lab	ab Director: Franklin R. Cockerill, III, M.D.

Patient Name		Report Status
TESTINGRNV,MHDMS	08/02/2012	Final
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