

Patient Name TESTINGRNV,MHDKM	Patient ID SA00048202	Age 11	Gender M	Order # SA00048202
Ordering Phys			DOB 06/15/2001	
Client Order # SA00048202	Account Information C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			Report Notes
Collected 08/02/2012				
Printed 08/29/2012 10:03				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MMADHC Gene, Known Mutation			REPORTED	08/06/2012 17:03	
Specimen		Blood			MCR
Specimen ID		1038534			MCR
Order Date		02 Aug 2012 10:37			MCR
Reason For Referral		Family history of methylmalonic aciduria and homocystinuria, cobalamin D type (MMADHC). Test for the presence of familial alteration(s) in the MMADHC gene.			MCR
Method		DNA sequence analysis was used to test for the presence of the p.R250X (c.748C>T) alteration in exon 8 of the MMADHC gene (GenBank accession number; NM 015702.2). Analysis for this specific alteration was performed because it has been identified in a family member.			MCR
Result		The p.R250X alteration was NOT detected.			MCR
Interpretation		Absence of the mutation(s) previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population to be a carrier of MMADHC.			MCR
		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.			
		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 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			

Performing Site Legend on Last Page of Report

Patient Name TESTINGRNV,MHDKM	Collection Date and Time 08/02/2012	Report Status Final
Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

Patient Name TESTINGRNV,MHDKM	Patient ID SA00048202	Age 11	Gender M	Order # SA00048202
Ordering Phys				DOB 06/15/2001
Client Order # SA00048202	Account Information			Report Notes
Collected 08/02/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/29/2012 10:03				

Test	Flag	Results	Unit	Reference Value	Perform Site*
------	------	---------	------	-----------------	---------------

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 17:01	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 TESTINGRNV,MHDKM	Collection Date and Time 08/02/2012	Report Status Final
Page 2 of 2		** End of Report **

* Report times for Mayo performed tests are CST/CDT