

Patient Name TESTING,89436	Patient ID	Age	Gender	Order # W2906035
Ordering Phys		DOB		
Client Order # W2906035	Account Information C7999998-STUSTEST 200 FIRST STREET SW ROCHESTER, MN 55901	Report Notes		
Collected 10/12/2009 06:00				
Printed 10/19/2009 11:17	(507)266-5730			

Test	Flag	Results	Unit	Reference Value	Perform Site*
MMACHC Gene, Full Gene Analysis				REPORTED 10/13/2009 12:07	
Specimen		Blood			MCR
Specimen ID		740558			MCR
Order Date		05 Aug 2009 11:49			MCR
Reason For Referral		Not provided. Test for the presence of mutations within the MMACHC gene.			MCR
Method		DNA sequence analysis was used to test for the presence of a mutation in all 4 exons of the MMACHC gene (GenBank accession number; NM_015506.2).			MCR
Result		A mutation was NOT identified			MCR
Interpretation		<p>Although this analysis did not identify a mutation within the MMACHC gene, this result does not exclude the possibility that this individual is a carrier of or affected with methylmalonic aciduria and homocystinuria, cobalamin C type (MMACHC). We predict that there may be disease-causing mutations not identified by the method described above (e.g. large deletions and duplications, or mutations in the promoter and intronic regions).</p> <p>If the patient has a family history, referral for genetic counseling is recommended. If MMACHC is a suspected diagnosis, correlation between other laboratory tests (e.g. analysis of total plasma homocysteine, urine organic acids, or complementation studies of skin fibroblasts) and clinical history is recommended.</p> <p>A genetic consultation may be of benefit.</p> <p>A list of common polymorphisms identified for this patient is available upon request.</p> <p>CAUTIONS: 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.</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>			MCR

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Extraction Performed?		YES			MCR
Reviewed By Dimitar K Gavrilov MD, PhD					MCR
Release Date		20 Aug 2009 11:55			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director:
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