

Patient Name TESTING,83934	Patient ID	Age	Gender	Order # W2834628
Ordering Phys		DOB		
Client Order # 83934	Account Information C7999998-STUSTEST	Report Notes		
Collected 10/06/2009 09:40	200 FIRST STREET SW ROCHESTER, MN 55901			
Printed 10/06/2009 12:03	(507)266-5730			

Test	Flag	Results	Unit	Reference Value	Perform Site*
MCAD, Known Mutation				REPORTED 10/06/2009 10:03	
Specimen		Blood			MCR
Specimen ID		749971			MCR
Order Date		15 Sep 2009 08:45			MCR
Reason for Referral		Family history of Medium-Chain Acyl-CoA Dehydrogenase (MCAD) deficiency. Test for the presence of familial alteration(s) within the ACADM gene.			MCR
Method		DNA sequence analysis was used to test for the presence of the p.G253R (c.757G>A) and p.985A>G (c.K329E) alterations in exons 9 and 11, respectively, of the MCAD gene (ACADM) (GenBank accession number; NM_000016). Analysis for these specific alterations was performed because they are known to be present in a family member.			MCR
Result		The p.G253R and p.K329E alterations were NOT detected.			MCR
Interpretation		Absence of the K329E mutation and the G253R variant of uncertain significance previously identified for an affected family member indicates that this individual is at no greater risk than someone in the general population for developing symptoms related to MCAD.			MCR
		This assay does not rule out the presence of other mutations within this gene or within 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.			
		A list of common polymorphisms identified for this patient is available upon request.			
		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.			
		Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of			

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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<p>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>					
Reviewed By:		Devin Oglesbee PhD			MCR
Release Date		23 Sep 2009 11:08			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
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