

Patient Name SAMPLEREP,MCADK N	Patient ID SA00057904	Age 46	Gender F	Order # SA00057904
Ordering Phys CLIENT,CLIENT			DOB 06/10/1966	
Client Order # SA00057904	Account Information C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			Report Notes
Collected 05/30/2013 00:00				
Printed 06/03/2013 10:43				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MCAD, Known Mutation					
RECEIVED: 05/31/2013 10:44 REPORTED: 06/03/2013 09:58					
Specimen		Blood			MCR
Specimen ID		1061998			MCR
Order Date		03 Jun 2013 08:22			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.K329E (c.985A>G) alteration in exon 11 of the MCAD gene (ACADM). Analysis for this specific alteration was performed because it is known to be present in a family member. Mutation nomenclature is based on GenBank accession number; NM 000016.			MCR
Result		The p.K329E 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 MCAD deficiency.			MCR
		This assay does not rule out the presence of other mutations in this gene or in 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.			

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREP,MCADK N	Collection Date and Time 05/30/2013 00:00	Report Status Final
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* Report times for Mayo performed tests are CST/CDT

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<p>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>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> <p>Laboratory developed test.</p>					
Reviewed By:		Jessica Rose Chavey			MCR
Release Date		03 Jun 2013 09:56			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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