MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MCADK A	SA00057905	46	F	SA00057905
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00057905	**Account Information**			**Report Notes**
Collected 05/30/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/03/2013 10:42				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MCAD, Known Mutation					
RECEIVED: 05/31/2013 10:46 **REPORTED:** 06/03/2013 09:58					
Specimen		Blood			MCR
Specimen ID		1061999			MCR
Order Date		03 Jun 2013 08:24			MCR
Reason For Referral					MCR
Family history of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. Test for the presence of familial alteration(s) within the ACADM gene.					
Method					MCR
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.					
Result					MCR
The following heterozygous sequence change was detected: Exon: 11 DNA change: c.985A>G Amino acid change: p.K329E (Lys329Glu) Classification: DELETERIOUS					
Interpretation					MCR
This alteration is a known deleterious mutation. This result indicates that this individual is a carrier of MCAD deficiency. This interpretation assumes that this individual is clinically normal and is not affected with MCAD deficiency. 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. Since a mutation has been identified, testing of other at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner to clarify their risk of having a child with MCAD deficiency. A genetic consultation may be of benefit.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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:
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 results may occur if the information provided is inaccurate or incomplete.

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:	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: