Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MCADS	SA00048726	10	M	SA00048726
Ordering Phys				**DOB** 06/15/2002
Client Order # SA00048726	**Account Information**			**Report Notes**
Collected 08/27/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/19/2012 15:21				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MCAD Mutation Screen			REPORTED 09/04/2012 11:31		
Specimen		Blood			MCR
Specimen ID		1038716			MCR
Order Date		27 Aug 2012 15:15			MCR
Reason For Referral					MCR
Patient reported to have features suggestive of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. Test for the presence of mutations in the ACADM gene.					
Method					MCR
Bi-directional sequence analysis is used to test for the presence of a mutation in all coding regions and intron/exon boundaries of the ACADM gene. Mutation nomenclature is based on GenBank accession number; NM 000016.					
Result					MCR
A mutation was NOT identified.					
Interpretation					MCR

This result decreases the likelihood but does not completely exclude the diagnosis of MCAD deficiency.

We predict that some individuals who have a diagnosis of MCAD deficiency may have disease causing mutations in the ACADM gene that are not identifiable by the method described (e.g. large deletions/duplications, promoter mutations, or deep intronic mutations).

This result should be interpreted in light of clinical findings, family history, and other laboratory data, such as a plasma acylcarnitine profile and urinary acylglycines.

Additionally, this assay does not exclude the presence of disease causing mutations in other genes that are associated with metabolic disease.

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: 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

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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					MCR
W Edward Highsmith Jr., PhD					
Release Date		04 Sep 2012 11:30			MCR

* Performing Site:

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