

Laboratory Service Report

1-800-533-1710

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

We are	77	P16	***** d #	Reference Value	Perform Site*
Test	Flag	Results	Unit	value	Site*
MCAD, Known Mutation					
RECEIVED: 05/31/2013 10:44 REPO	RTED: 06/03/201	.3 09:58			
Specimen		Blood			MCR
Specimen ID		1061998			MCR
Order Date		03 Jun 2013 08:22			MCR
Reason For Referral					MCR
Family history of medium	-chain acyl-CoA	dehydrogenase (MCAD))		
deficiency. Test for th	e presence of f	amilial alteration(s	;)		
within the ACADM gene.					
Method					MCR
DNA sequence analysis wa	s used to test	for the presence of			
the p.K329E (c.985A>G) a					
gene (ACADM). Analysis	_				
performed because it is		_			
member. Mutation nomeno	lature is based	l on GenBank accessio	n		
number; NM 000016.					
Result					MCR
The p.K329E alteration w	as NOT detected	1.			
Interpretation					MCR
Absence of the mutation(
affected family member i					
no greater risk than som	_	eral population to b	e		
a carrier of MCAD defici	ency.				
This assay does not rule	out the presen	ace of other mutation	ıs		
in this gene or in other	_				
	J-1100 01100 0110				

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.

metabolic disease. Errors in the diagnosis or pedigree provided to us, including non-paternity, may lead to an

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

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



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MCR

MCR

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Ordering Phys CLIENT,CLIENT		,		DOB 06/10/1966	
Client Order # SA00057904	Account Information			Report Notes	
Collected	C7028846-DLMP Roches	ter			
05/30/2013 00:00		3050 Superior Drive			
Printed	Rochester, MN 55901				
06/03/2013 10:43					

Reference Perform
Test Flag Results Unit Value Site*

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.

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 Release Date 03 Jun 2013 09:56

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,MCADK N	05/30/2013 00:00	Final
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