Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MCDKM A	SA00058677	46	F	SA00058677
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058677	**Account Information**			**Report Notes**
Collected 06/02/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/05/2013 08:51				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MLYCD Gene, Known Mutation					
RECEIVED: 06/03/2013 13:52 **REPORTED:** 06/04/2013 11:49					
Specimen		Blood			MCR
Specimen ID		1062017			MCR
Order Date		04 Jun 2013 11:28			MCR
Reason For Referral					MCR

Family history of malonyl-coenzyme A decarboxylase (MCD) deficiency. Test for the presence of familial alterations in the MLYCD gene.

Method MCR

DNA sequence analysis was used to test for the presence of the p.G3D (c.8G>A) alteration in exon 1 of the MLYCD gene. Analysis for this specific alteration was performed because it was identified in a family member of this individual. Mutation nomenclature is based on GenBank accession number NM_012213.2.

Result MCR

The following heterozygous sequence change was detected:
Exon: 1
DNA change: c.8G>A
Amino acid change: p.G3D (Gly3Asp)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result indicates that this individual is a carrier of MCD deficiency. This interpretation assumes that this individual is healthy and not clinically affected with MCD deficiency.

Since a mutation has been identified in the MLYCD gene, genetic testing of 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 MCD deficiency.

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.

Performing Site Legend on Last Page of Report

TEST SITE

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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.					
Extraction Performed?		Yes.			MCR
Reviewed By Emily Christine Lauer					MCR
Release Date		04 Jun 2013 11:48			MCR

* Performing Site:

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

TEST SITE