Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,BTDKM	SA00045674	45	F	SA00045674
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045674	**Account Information**			**Report Notes**
Collected 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/17/2012 17:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*
BTD Gene, Known Mutation			REPORTED 07/13/2012 09:54		
Specimen		Blood			MCR
Specimen ID		1038134			MCR
Order Date		08 May 2012 08:09			MCR
Reason For Referral					MCR

Family history of biotinidase deficiency. Test for the presence of familial alterations in the BTD gene.

Method MCR

DNA sequence analysis was used to test for the presence of the p.D444H (c.1330G>C)and p.Q456H (c.1368A>C) alterations in exon 4 of the BTD gene. Testing was performed for these specific alterations because they were identified in a family member. Mutation nomenclature is based on GenBank accession number; NM_000060.2.

Result MCR

The p.D444H and p.Q456H alterations were NOT detected.

Interpretation MCR

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 biotinidase deficiency.

This assay does not rule out the presence of other disease causing mutations in this gene or other genes 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.

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the

Performing Site Legend on Last Page of Report

TEST SITE

Patient Name	Patient ID	Age	Gender	Order #
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Ordering Phys				**DOB** 06/10/1966
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Printed 07/17/2012 17:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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 Melody Elizabeth Kimball					MCR
Release Date		13 Jul 2012 09:53			MCR

* Performing Site:

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

TEST SITE