MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREOPRT,BTDMS	SA00045679	45	F	SA00045679
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045679	**Account Information**			**Report Notes**
Collected 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/17/2012 17:47				

Test	Flag	Results	Unit	Reference Value	Perform Site*
BTD Gene, Full Gene Analysis			REPORTED 07/13/2012 09:56		
Specimen		Blood			MCR
Specimen ID		1038136			MCR
Order Date		08 May 2012 08:16			MCR
Reason For Referral					MCR

Biochemical analysis is consistent with profound biotinidase deficiency (enzyme activity <1.4U/L). Test for the presence of mutations in the BTD gene.

Method MCR

Bi-directional sequence analysis was used to test for the presence of a mutation in all coding regions and intron/exon boundaries of the biotinidase (BTD) gene. Mutation nomenclature is based on GenBank accession number; NM_000060.2.

Result MCR

A mutation was NOT detected.

Interpretation MCR

The observed biotinidase activity level (1.0 U/L) is most consistent with profound biotinidase deficiency. However, no mutations were identified by molecular analysis. We predict that there may be disease causing mutations in the BTD gene not identifiable by the method described (e.g. large deletions/duplications, promoter mutations, or deep intronic mutations). Given the reduced biotinidase activity level, this patient is at risk to develop symptoms related to this disorder. Treatment with oral biotin supplementation is recommended.

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

* Report times for Mayo performed tests are CST/CDT

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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