MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,BTDMS	SA00045681	45	F	SA00045681
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045681	**Account Information**			**Report Notes**
Collected 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/17/2012 17:51				

Test	Flag	Results	Unit	Reference Value	Perform Site*
BTD Gene, Full Gene Analysis		REPORTED 07/13/2012 09:56			
Specimen		Blood			MCR
Specimen ID		1038137			MCR
Order Date		08 May 2012 08:16			MCR
Reason For Referral					MCR
Biochemical analysis is consistent with partial biotinidase deficiency (enzyme activity between 1.4-3.5U/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
The following heterozygous sequence changes were detected: Exon: 4 DNA change: c.1330G>C Amino Acid change: p.D444H (Asp444His) This sequence change is a pathogenic mutation associated with partial biotinidase deficiency. Exon: 4 DNA change: c.1368A>C Amino Acid change: p.Q456H (Gln456His) Classification: DELETERIOUS					
Interpretation					MCR
The p.Q456H alteration is a known deleterious mutation. The p.D444H alteration is a known deleterious mutation associated with partial biotinidase deficiency. The observed biotinidase activity level (2.4 U/L) and this molecular analysis are consistent with a diagnosis of partial biotinidase deficiency. This patient may be at risk to develop symptoms related to this disorder. Treatment with oral biotin supplementation is recommended. Since mutations have been identified, genetic testing of at risk family members is possible. Mutation specific testing is available at Mayo Medical Laboratories by ordering BTDKM/89013 BTD Gene, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Testing of samples from the parents of this patient would be necessary to confirm that the two mutations are on different chromosomes.

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 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 MCR
Melody Elizabeth Kimball
Release Date 13 Jul 2012 09:54 MCR

* Performing Site:

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