MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORTS,BTDKM	SA00045675	45	F	SA00045675
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045675	**Account Information**			**Report Notes**
Collected 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/17/2012 17:46				

Test	Flag	Results	Unit	Reference Value	Perform Site*
BTD Gene, Known Mutation		REPORTED 07/13/2012 09:54			
Specimen		Blood			MCR
Specimen ID		1038135			MCR
Order Date		08 May 2012 08:15			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 following heterozygous sequence change was 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.					
Interpretation					MCR
The p.D444H alteration is a known deleterious mutation associated with partial biotinidase deficiency. The presence of a mutation previously identified in an affected family member indicates that this individual is a carrier of partial biotinidase deficiency. This interpretation assumes that this individual is healthy and not clinically affected with biotinidase deficiency. Since a mutation has been identified in the BTD gene, genetic testing of at risk family members is possible. If appropriate, enzymatic and molecular studies should be offered to this individual's reproductive partner to clarify their risk of having a child with 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.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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 09:53 MCR

* Performing Site:

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