

Patient Name TESTING,89740	Patient ID	Age	Gender	Order # W2934985
Ordering Phys		DOB		
Client Order # W2934985	Account Information C7999998-STUSTEST 200 FIRST STREET SW ROCHESTER, MN 55901	Report Notes		
Collected 10/15/2009 06:00				
Printed 10/16/2009 15:11	(507)266-5730			

Test	Flag	Results	Unit	Reference Value	Perform Site*
Rapid DNA Extraction				REPORTED 10/15/2009 13:07	
Comment		See Comment			MCR
Genomic DNA was extracted.					

BTK Known Mutation Panel, B	REPORTED 10/15/2009 13:07
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BTK Gene, Known Mutation
 BTK Known Mut Result MCR

This individual was shown to have one copy of the following familial mutation in the BTK gene:
 g.46199G>T, c.135G>T, p.M1I

BTK Known Mut Interpretation MCR

The above mutation within the BTK gene was previously identified in a family member affected with X-linked Agammaglobulinemia (XLA). The presence of this mutation, therefore, is consistent with a diagnosis of XLA for this individual. Clinical correlation is recommended.

Reviewed By MCR

Roshini S. Abraham, Ph.D.
 See Comment
 Fluorescent DNA sequencing was used to test for the presence of a specific mutation in the BTK gene which was previously identified in a family member.

We predict that a small percentage of individuals who have a diagnosis of XLA may have a mutation that is not identified by the methods described above.

The presence of a BTK mutation does not necessarily confirm a diagnosis of XLA. Clinical correlation recommended. Please see: Graziani S, Di Matteo G, Benini L, Di Cesare S, Chiriaco M, Chini L, Chianca M, De Iorio F, La Rocca M, Iannini R, Corrente S, Rossi P, Moschese V. Identification of a Btk mutation in a dysgammaglobulinemic patient with reduced B cells: XLA diagnosis or not? Clinical Immunology. 2008; 128: 322-8. And also: Fleisher T and Notarangelo L. What does it take to call it a pathogenic mutation? Clinical Immunology. 2008; 128, 285-6.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available from the lab upon request.

CAUTIONS:

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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<p>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.</p> <p>Test results should be interpreted in the context of clinical findings, family history, and other laboratory data.</p>					
BTK Gene, Known Mutation Sequencing		Performed			MCR
Btk Protein Flow, B		Normal Expression			MCR
<p>Normal expression of BTK in monocytes and B cells, does not appear to be consistent with XLA. If clinical evidence for XLA is present, suggest Btk genotyping (89307) to confirm that no Btk mutations are present since approximately 30% of Btk mutations can affect protein function but maintain normal protein expression.</p> <p>Analyte Specific Reagent This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.</p>					

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director:
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