MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,BTKK A	SA00059532	47	F	SA00059532
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00059532	**Account Information**			**Report Notes**
Collected 06/27/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 09/16/2013 08:20				

Test	Flag	Results	Unit	Reference Value	Perform Site*
BTK Gene, Known Mutation					
RECEIVED: 07/03/2013 13:32 **REPORTED:** 09/04/2013 11:21					
BTK Gene, Known Mutation					
BTK Known Mut Result					MCR
This individual was shown to have one copy (heterozygous) of the following familial mutation in the BTK gene: missense substitution - g.100500969, c.862C>T, p.Arg288Trp (p.R288W). See interpretation for details.					
BTK Known Mut Interpretation					MCR

This female individual has a heterozygous missense variation in Exon 10 of the BTK gene, which encodes the SH2 domain of the Btk protein. This variation results in a change from a basic amino acid (Arg) to an aromatic, hydrophobic amino acid (Trp) at position 288 in the Btk protein. This variant has been previously reported in BTKBase, a database of BTK mutations in XLA patients. In male patients, this variant is reported to be possibly pathogenic by the Uniprot database and deleterious by SIFT.

The presence of one copy of this variation would confer carrier status for a possible pathogenic variation in the BTK gene.

Since the familial mutation has been identified in the BTK gene in this female, genetic testing for this specific mutation in symptomatic male family members and/or female relatives of childbearing age is recommended. Please contact the laboratory at 1-800-533-1710 or the online test catalog at mayomedicallaboratories.com for information about how to order the "BTK Gene, Known Mutation" (89306).

The Btk gene has 19 exons, 18 of which are coding and produce an approximately 77kD protein. There are over 600 mutations reported in the BTK gene, including missense, nonsense, frameshift, deletions, insertions and splice-site mutations. The full-gene sequencing method can identify 92% of mutations within the BTK gene. However, 8% of mutations, which include large deletions, duplications or rearrangements cannot be detected by this method, but could potentially be identified by Btk flow cytometry due to absent Btk protein.
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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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A genetic consultation may be of benefit.

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

CAUTIONS: 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.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data.
Laboratory developed test.

Reviewed By	Jamie Bruflat	MCR
BTK Gene, Known Mutation Sequencing	Performed	MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.