MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, 89306	L3MRNW4075493	58	F	W4075493

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
04/21/11 02:02 P	04/21/11 02:02 P	04/25/11 10:19 A	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

TEST REQUESTED	HI LO		REF RANGE	PERFORM SITE *
Rapid DNA Extraction				**REPORTED: 04/21/11 02:44 P**
Comment		**Genomic DNA was extracted.**		**MCR**
BTK Gene, Known Mutation				**REPORTED: 04/21/11 02:56 P**
BTK Known Mut Result				**MCR**

A heterozygous nonsense mutation was identified in exon 2 (old)/exon 1 (new nomenclature) of the BTK gene (PH domain) - c.37C>T, p.R13X. BTKBASE nomenclature: g.46233 (old), g.100516892 (new); a familial mutation previously identified in a male offspring of this patient.

BTK Known Mut Interpretation **MCR**

This result is consistent with carrier status for X-linked Agammaglobulinemia (XLA) for this female.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)

Reviewed By **Roshini S. Abraham, Ph.D.** **MCR**

This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. It has not been cleared or approved by the U.S. Food and Drug Administration.
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.

** Perform Site Legend on last page of report*

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, 89306	Final	04/21/11 02:02 P

Specimen receipt and report times are in CST/CDT

REPRINT

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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 genetic consultation may be of benefit.
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.

Sequencing	**Performed**	**MCR**

*** PERFORMING SITE**

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.