MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

BTKK

Bruton Tyrosine Kinase (BTK) Genotype, Known Mutation

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059531	SAMPLEREPORT, BTKK N		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059531	SA00059531	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		27 Jun 2013 00:00			

BTK Gene, Known Mutation

BTK Gene, Known Mutation

BTK Known Mut Result

MCR

The following familial mutation was NOT detected in the BTK gene: c.1426_1430dup, p.Met477Ilefs*9.

BTK Known Mut Interpretation

MCR

The familial mutation previously identified in the male offspring of this patient was not detected. This result was verified by full-gene sequencing to reduce the risk of a false negative result for this patient. This result suggests that this patient is not a carrier for XLA, and suggests that her male offspring likely has a de novo mutation in the BTK gene, accounting for the XLA phenotype.

There is a small (<5%) risk that this patient carries the familial BTK mutation in a population of cells in the germline (germline mosaicism). This status would not be identified with the described testing methodology. Individuals with germline mosaicism are at increased risk for passing the mutation to future offspring.

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.

ADDITIONAL INFORMATION

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.

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 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 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.

If the patient has had an allogeneic blood or marrow transplant or a recent (i.e. less than 6 weeks from time of sample collection) heterologous blood transfusion these results may be inaccurate due to the presence of donor DNA. Laboratory developed test.

Reviewed By

MCR

Jamie Bruflat

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

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BTK Gene, Known Mutation Sequencing MCR

Performed

Received: 03 Jul 2013 13:31 **Reported:** 04 Sep 2013 11:18

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905