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

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
Roshini S. Abraham, Ph.D.

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.


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.

BTK Gene, Known Mutation Sequencing

Sequencing
Performed
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.

Received: 15 Oct 2009 13:07