BTK Gene

Subjects: Genetics
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Definition

Bruton tyrosine kinase

1. Normal Function

The *BTK* gene provides instructions for making a protein called Bruton tyrosine kinase (BTK), which is essential for the development and maturation of B cells. B cells are specialized white blood cells that help protect the body against infection. These cells can mature into cells that produce special proteins called antibodies or immunoglobulins. Antibodies attach to specific foreign particles and germs, marking them for destruction. The BTK protein transmits important chemical signals that instruct B cells to mature and produce antibodies.

2. Health Conditions Related to Genetic Changes

2.1. Isolated Growth Hormone Deficiency

A few mutations in the *BTK* gene have been found to cause isolated growth hormone deficiency type III, a condition characterized by slow growth, short stature, and a weakened immune system. Mutations that cause this condition lead to production of a nonfunctional version of the BTK protein. People with isolated growth hormone deficiency are prone to infections because they produce very few B cells and have a shortage of antibodies (agammaglobulinemia). A lack of the BTK protein is likely responsible for the immune system symptoms, but how a shortage of BTK protein causes short stature in affected individuals is unclear.

2.2. X-Linked Agammaglobulinemia

More than 600 different mutations in the *BTK* gene have been found to cause X-linked agammaglobulinemia (XLA). Most of these mutations result in the absence of the BTK protein. Other mutations change a single protein building block (amino acid), which probably leads to the production of an abnormal BTK protein that is quickly broken down in the cell. The absence of functional BTK protein blocks B cell development and leads to a lack of antibodies, causing an increased susceptibility to infections in people with XLA.

Some people with XLA have large DNA deletions that remove one end of the *BTK* gene and all of a neighboring gene known as *TIMM8A*. Mutations in *TIMM8A* cause deafness-dystonia-optic neuronopathy (DDON) syndrome, which is characterized by hearing loss, vision problems, a decline in intellectual function (dementia), and involuntary muscle tensing (dystonia) or difficulty coordinating movements (ataxia). Individuals with large DNA deletions that include the *BTK* gene and the *TIMM8A* gene have the signs and symptoms of both XLA and DDON syndrome.

3. Other Names for This Gene

- AGMX1
- AT
- ATK
- BPK
- Bruton agammaglobulinemia tyrosine kinase
- Bruton's tyrosine kinase
- BTK_HUMAN
- dominant-negative kinase-deficient Bruton's tyrosine kinase
- IMD1
- MGC126261

- MGC126262
- PSCTK1
- tyrosine-protein kinase BTK
- XLA

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Keywords

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