# Bruton's Agammaglobulinemia

Bruton's agammaglobulinemia is a rare X-linked recessive immunodeficiency caused by an inability to generate mature B-cells. Mature B-cells are an essential component of the immune response as they normally manufacture immunoglobulins, which defend the body from infections. Bruton's agammaglobulinemia is caused by a mutation in the BTK tyrosine kinase gene, which controls the differentiation of B-cells. Decreased immunoglobulins of all classes are observed. Because the disease is X-linked recessive, it is commonly seen in boys and presents as recurrent bacterial infections after 6 months of age. Infants typically do not get infections before 6 months because they have maternal IgG antibodies that protect them. The diagnosis can be suspected when patients have a normal amount of early B-cells (or pre-B cells) but decreased number of mature B-cells.


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## Pathophysiology

### X-linked Recessive

X-suit with Recessive-chocolate

This disease is inherited in an X-linked recessive fashion, meaning this disease is more common in boys.

### Defective Bruton Tyrosine Kinase

Broken Tire Kite-ace

Bruton tyrosine kinase, or BTK, normally controls the differentiation of immature B-cells to mature B-cells. This tyrosine kinase is defective in Bruton's agammaglobulinemia.

### Blocks B Cell Maturation

Mature Basketballs Blocked

B-cell maturation is blocked due to dysfunction of Bruton tyrosine kinase, or BTK. This results in normal amounts of pre-B-cells (or immature B-cells), but decreased number of B-cells and decreased immunoglobulin production of all classes.

## Signs & Symptoms

### Decreased Immunoglobulins of All Classes

Down-arrow In-moon-goblins of all Classes

Defective B-cell maturation affects all classes of immunoglobulins, including IgG, IgM, IgE and IgA.

### Bacterial Infections After Six Months

Bacteria-guy and (6) Sax

Infants with Bruton's agammaglobulinemia typically present with recurrent bacterial infections after 6 months of age due to poor immunoglobulin production. Infants typically do not get infections before six months because they have maternal IgG antibodies that protect them.

### Maternal IgG Protects Before Six Months

(IgG) Gold-goblin wearing I-heart-mom-shirt Protecting Baby

Infants typically do not get infections before six months because they have maternal IgG antibodies that protect them.