

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.