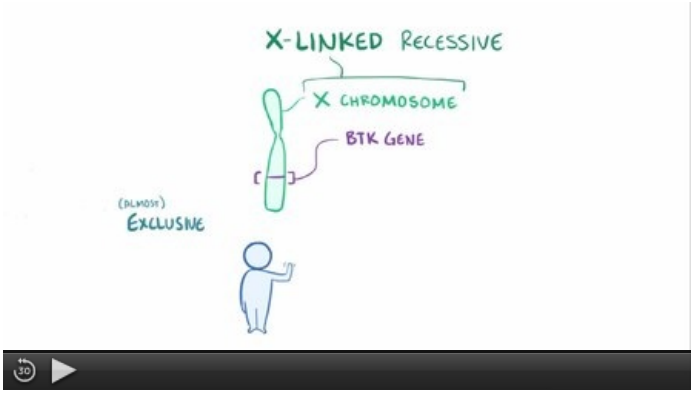
Burton's agammaglobulinemia

Bruton's agammaglobulinemia (X-linked agammaglobulinemia, X-linked hypogammaglobulinemia, XLA, OMIM: & nbsp; 300755 (https://www.omim.org/entry/300755)) was the first described genetic immunodeficiency (described by Bruton in 1952). It belongs to antibody immunodeficiency.



Video in English. Definition, pathogenesis, symptoms, complications, and treatment.

Etiology and pathogenesis

BTK protein kinase was identified as an abnormal product (**B**ruton **t**yrosine **k**inase), which is highly expressed in precursors of B-lymphocytes. Under normal circumstances, the development of B-lymphocytes first rearranges the immunoglobulin heavy chain genes, followed by the rearrangement of the light chain genes. It turns out that when BTK is mutated, development ends by rearranging the immunoglobulin heavy chain gene. Light chains are not synthesized and immunoglobulin molecules cannot be assembled. BTK kinase is responsible for biosignal transfer from B-cell receptors (and their precursors) to effector mechanisms.

The critical segment in this disease is the part of the long arm X chromosome - Xq21.3-q22. Thus, as GR, the hereditary disease is much more common in boys.

Clinical picture

The disease does not manifest fully until **6 months of age,** until then the newborn is at least partially **protected by maternal immunoglobulins**. Manifestations mainly include recurrent bacterial infections of the respiratory system (bronchitis, otitis, pharyngitis, sinusitis); almost always involved pathogens are *Haemophilus influenzae*, *Streptococcus pneumoniae* or *Staphylococcus aureus*. There is also a high susceptibility to viral infections caused, for example, by enteroviruses (polioviruses, echoviruses, coxsackieviruses).

The classic finding is a marked reduction (or complete absence) of B-lymphocytes, plasma cells and all classes of peripheral blood immunoglobulins. T cell levels are normal or elevated.

There are different alleles variants; in some, an association with other manifestations is described, such as deafness, growth hormone deficiency, or various neurological disorders.

Therapy

Treatment includes immunoglobulin replacement therapy or prophylactic administration of antibiotics or antivirals.

Links

Related Articles

- Primary immunodeficiency
- Autosomal inherited agammaglobulinemia
- Antibodies

External links

- Bruton's agammaglobulinemia - Youtube video (https://www.youtube.com/watch?v=GRra7J3ahUc&list=PLY33uf2n4e6Neq5yeTHn2zlW7zEzPYjYZ&index=5)
- Bruton's agammaglobulinemia - English Wikipedia (https://en.wikipedia.org/wiki/X-linked_agammaglobulinemia)

Sources

- ŠÍPEK, Antonín. *Genetic disorders of the immune system* [online]. [cit. December 5, 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

Used literature

- BARTŮŇKOVÁ, Jiřina. *Immunodeficiency.* 1. edition. Prague : Grada, 2002. 228 pp. ISBN 80-247-0244-4.