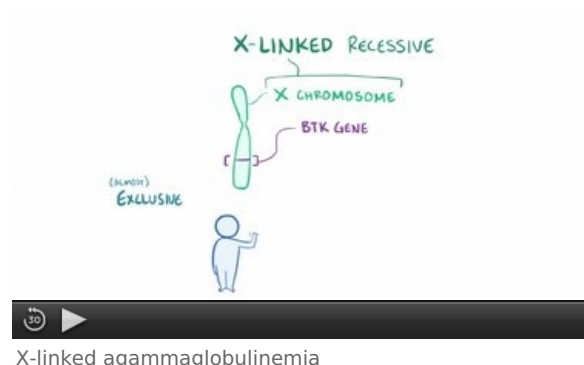


Bruton's agammaglobulinemia

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

Etiology and pathogenesis

The protein kinase BTK (Bruton tyrosine kinase), which is expressed in large quantities in B-lymphocytes precursors. Under normal conditions, B-lymphocyte development first involves the rearrangement of genes for the immunoglobulin heavy chain, followed by the rearrangement of genes for the light chain. It turns out that if BTK is mutated, development ends with rearrangement of the immunoglobulin heavy chain gene. Light chains are not synthesized and immunoglobulin molecules cannot be assembled. BTK kinase is responsible for biosignal transmission from B-lymphocyte receptors (and their precursors) to effector mechanisms.



The critical section in this disease is part of the long arm of the X chromosome – Xq21.3-q22. Thus, as a GR hereditary disease, it occurs much more often in boys.

Clinical Picture

The disease begins to fully manifest itself only after the 6th month of age because until then the newborn is at least partially protected by maternal immunoglobulins. Manifestations mainly include repeated bacterial infections of the respiratory system (bronchitis, otitis, pharyngitis, sinusitis...); the pathogens involved are almost always *Haemophilus influenzae*, *Streptococcus pneumoniae*, or *Staphylococcus aureus*. There is also a high susceptibility to viral infections, caused by, for example, enteroviruses (polioviruses, echoviruses, coxsackieviruses). A classic finding is a clear reduction (or complete absence) of B-lymphocytes, plasma cells and all immunoglobulin classes in peripheral blood. T-lymphocyte levels are normal or elevated.

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

Therapy

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

Links

Related articles

- Primary immunodeficiency
- Autosomally inherited agammaglobulinemia
- Antibody

External links

- Brutonova agammaglobulinemie – Youtube video (<https://www.youtube.com/watch?v=GRra7J3ahUc&list=PLY33uf2n4e6Neq5yeTHn2zIW7zEzPYjYZ&index=5>)
- Brutonova agammaglobulinemie – anglická Wikipedie (https://en.wikipedia.org/wiki/X-linked_agammaglobulinemia)

Source

- ŠIPEK, Antonín. *Genetically determined disorders of the immune system* [online]. Last revision 9/6/2006, [cit. 5/12/2009]. < <http://www.genetika-biologie.cz/primarni-immunodeficiency> >.

References

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