

Wiskott-Aldrich syndrome

This article has been translated from WikiSkripta; ready for the **editor's review**.

Wiskott-Aldrich syndrome (WAS) is a recessive X-linked disease characterized by **thrombocytopenia**, **eczema** and an **increased susceptibility to infectious diseases**. The responsible WAS gene was located on the X chromosome (region Xp11.22-p11.23). The protein encoded by this gene (**WASP** - **W**iskott- **A**ldrich **S**ndrome **P**rotein) is probably involved in the association of membrane receptors with cytoskeletal components, the exact importance of the protein for platelet function has not yet been clarified. In ZAP-70 T cells, it activates WASp, which induces the formation of an immunological synapse by the actin cytoskeleton.

Clinical picture

In patients (since it is an X-linked disease, most patients are male), the syndrome is manifested by thrombocytopenia (characterized by a **small platelet size**), which is manifested by increased bleeding. Another manifestation is **eczema**, which appears shortly after birth. T-cell counts are initially normal, but eventually decline due to their impaired proliferative capacity. We show **decreased levels of IgM** in the serum, but normal levels of IgG. IgA and IgE levels are paradoxically elevated. Autoimmune reactions are also described.

Prognosis and therapy

The prognosis of the disease is not good, death most often occurs due to massive **bleeding** or due to **chronic infections**. Malignant tumors of the lymphatic system also develop. The only treatment is a bone marrow transplant. Splenectomy can help treat thrombocytopenia.

Links

Related articles

- Primary immunodeficiency

Source

- ŠÍPEK, Antonín. Genetic disorders of the immune system [online]. Last revision June 9, 2006, [cited. December 9, 2009]. < <http://www.genetika-biologie.cz/primarni-imunodeficiency> >.
- ws:Wiskottův-Aldrichův syndrom

References

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

