Hyperimmunoglobulin M syndrome

Hyperimmunoglobulin M syndrome is a heterogeneous group of several types of antibody primary immunodeficiencies, the most common type of them is type 1.

Hyperimmunoglobulin M syndrome, type 1

This type (Hyper-IgM immunodeficiency X-linked, HIGM1, XHIM, OMIM: 308230) represents X-linked immunodeficiency caused by a mutation in the antigenic ligand gene **CD40** (CD40LG, localization Xq26). This ligand is essential for the interaction of T and B cells before the onset of isotype switching from IgM production to other classes of immunoglobulins. Studies have shown that T-lymphocytes are defective, as carrier B-lymphocytes (heterozygous women) are properly stimulated even with a small number of non-defective T-lymphocytes.

The clinical picture

Manifestation involves **high levels of IgM** and the absence of other immunoglobulin classes (IgG, IgA). In addition to antibody deficiency, there is also a **cellular deficit** - due to defective T-lymphocytes and autoimmune **neutropenia** (tendency to develop autoimmune diseases). Manifestations include susceptibility to various bacterial and viral infections, including opportunistic infections (for example, diarrhea caused by Cryptosporidium parvum is typical).

Other types

Other types of IgM hyperimmunoglobulinemia syndrome are mentioned on a separate page.

References

- ws:Syndrom hyperimunoglobulinemie IgM/další typy

Related articles

- Protilátky
- Primární imunodeficience
- Brutonova agamaglobulinemie

References

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. The last revision 9. 6. 2006, [cit. 5. 12. 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

Used literature

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

Kategorie:Imunologie Kategorie:Genetika Kategorie:Pediatrie