

Autosomally inherited agammaglobulinemia

Autosomally inherited agammaglobulinemia	
Clinical picture	repeated bacterial infections of the respiratory system, susceptibility to viral infections, congenital immunodeficiency
Cause	x-chromosome mutation
Diagnostics	examination of differential blood count, DNA diagnosis
Classification and references	

In addition to the most common (X-linked) **Bruton's agammaglobulinemia**, there are also rare, **autosomally inherited variants of agammaglobulinemia** (non-Bruton's agammaglobulinemia[1] (<https://www.omim.org/entry/601495>)), which can also affect **girls** to a greater extent. However, they manifest phenotypically as classic X-linked agammaglobulinemia.

Autosomal recessive inherited forms can be caused by a mutation in the gene for the μ IgM heavy chain (IGHM, location 14q32.33), in the gene for the κ 5 light chain (IGLL1, location 22q11.21) or in the gene for the adapted protein BLNK (location 10q23.2), whose activity is associated with BTK kinase.

An autosomal dominant inherited form was also described, caused by disruption of the LRRC8 gene (Leucine-rich repeat-containing protein 8; localization 9q34.13) by balanced translocation of the 9th and 20th chromosomes.

Links

related articles

- Bruton's agammaglobulinemia
- Primary immunodeficiency
- Antibodies

Source

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. [cit. 24. 1. 2009]. <<http://www.genetika-biologie.cz/primarni-imunodeficiencie>>.

References

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