Autosomal inherited agammaglobulinemia

In addition to the most common (X-linked) **Bruton's agammaglobulinemia,** there are rare, **autosomal inherited variants of agammaglobulinemia** (non-Bruton's agammaglobulinemia) ; OMIM: 601495 (https://www.omim.org/entry/601495)), which can affect **girls** to a greater extent. However, the phenotype manifests itself as classic X-linked agammaglobulinemia.

Autosomal recessive inherited forms can be caused by a mutation in the gene for heavy chain μ IgM (IGHM, localization 14q32.33), for κ5 light chain (IGLL1, localization 22q11.21) or in the gene for the adapted BLNK protein (localization 10q23.2), whose activity is associated with BTK kinase.

The autosomal dominantly inherited form, caused by disruption of the LRRC8 gene (Leucine-rich repeat-containing protein 8; localization 9q34.13) balanced translocations of 9 and 20 chromosomes, has also been described.

Links

Related Articles

- Bruton's agammaglobulinemia
- Primary immunodeficiency
- Antibodies

Sources

- ŠÍPEK, Antonín. *Genetic disorders of the immune system* [online]. [cit. January 24, 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.