Autoimmune lymphoproliferative syndrome

(Canale-Smith Syndrome, OMIM: 601859 (https://www.omim.org/entry/601859))

It is caused by a mutation in the gene for **FAS** or **FAS ligand.**

- The FAS antigen or **CD95** is encoded by the **TNFRSF6** gene, located in the 10q24.1 region.
- The FAS ligand (**CD95L**) is encoded by the **TNFSF6** gene located at 1q23.

The syndrome is inherited autosomal recessively, although even an autosomal dominant type cannot be completely ruled out.

Clinical manifestations

They can be different. In principle, **apoptosis disorders** occur with various consequences on the overall condition of the organism. The onset of the disease may be as early as the prenatal period as well as after several years of life. The most common manifestation is a **lymphoproliferative syndrome**, most often accompanied by **splenomegaly**. **Hepatomegaly** and lymph node enlargement are not always present. The **increase** in circulating lymphocytes is not constant, mostly B-lymphocytes multiply. T-lymphocytes infiltrate some tissues (these are so-called double-negative CD4-CD8- T-lymphocytes). Another typical manifestation is **autoimmune reactions**. The formation of **autoantibodies** against cells of the hematopoietic lineage has been described, which results in possible thrombocytopenia, neutropenia, or anemia. However, **immunodeficiency** as a direct manifestation of autoimmune lymphoproliferative syndrome is relatively rare.

Links

Related articles

- Primary immunodeficiency

Resource

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. The last revision 9. 6. 2006, [cit. 28. 12. 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

Literature

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