X-linked lymphoproliferative syndrome

X-linked lymphoproliferative syndrome (Purtil's syndrome, Duncan's disease, OMIM: 308240) is an X-linked inherited disease caused by a mutation in the SH2D1A gene (also called SAP, localization Xq25). This mutation leads to uncontrolled cytotoxic T-cell responses to Ebstein-Barr virus (EBV). The function of the SLAM molecule (Signaling lymphocyte activation molecule), which affects the cooperation of T- and B-lymphocytes, is impaired.

Affected individuals are healthy until the first contact with EBV, which can result in three different reactions:

1. Infectious mononucleosis – with severe, often fatal consequences, 50% of cases.
2. Lymphoproliferative syndrome – mainly affecting the B-line, 25% of cases.
3. Hypogammaglobulinemia – 25% of cases.

All three conditions can occur in one patient over time. The overall prognosis is not favorable, 70% of boys die within 10 years of age.

Links

Related

- Primary immunodeficiency

Source

- ŠÍPEK, Antonín. *Genetic disorders of the immune system* [online]. The last revision 9. 6. 2006, [cit. 17. 12. 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

References

- BARTŮŇKOVÁ, Jiřina. *Imunodeficiency.* 1. edition. Prague : Grada, 2002. 228 pp. ISBN 8024702444.