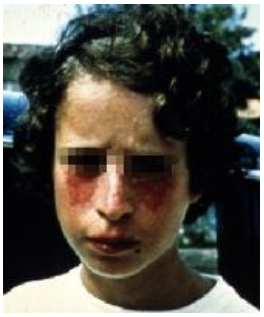
Bloom's syndrome

Bloom syndrome (BLM, BLS, OMIM: 210900 (https://omim.org/entry/210900)) is an autosomal recessive inherited chromosomal instability syndrome. The responsible gene (BLM, Bloom syndrome, RecQ helicase-like) is located in region 15q26.1. The normal gene product is one type of DNA helicase (RECQL3).


Bloom Syndrome

Already in the first year of life, those affected develop **telangiectatic erythema** that is sensitive to sunlight. Immunological manifestations include B-cell **disorders** with **low levels of** IgG, IgA, and IgM immunoglobulins with consequent susceptibility to infections.

We also observe tendencies in cells to develop cytogenetic abnormalities, which often result in **malignant transformation**.

Links

Related articles

- Ataxia telangiectasia
- Chromosomal instability syndromes
- Hereditary tumor syndromes
- Primary immunodeficiency

External links

- Bloom syndrome, Genetics Home Reference (https://ghr.nlm.nih.gov/condition/bloom-syndrome)

Sources

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. Poslední revize 9. 6. 2006, [cit. 7. 12. 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.
- BARTŮŇKOVÁ, Jiřina. *Imunodeficience.* 1. vydání. Praha : Grada, 2002. 228 s. ISBN 80-247-0244-4.