

Ataxia teleangiectasia

Ataxia-telangiectasia (Louis-Bar syndrome) is a complex syndrome with neurological, immunological, hepatic, cutaneous and endocrinological abnormalities. The inheritance of the syndrome is **autosomal recessive**, the participating gene (**ATM**) has been located in the 11q22-q23 region. The normal gene product is a **DNA-dependent protein kinase** (ATM), which is involved in cell cycle regulation and, in interaction with the **p53 protein**, the cell's response to genotoxic stress. ATM protein kinase is activated immediately after the break of both strands of DNA and initiates signalling towards repair mechanisms and cell cycle checkpoints to minimize the consequences of damage. When this protein is mutated, the DNA repair mechanisms are weakened, which is reflected in the **increased sensitivity of cells to ionizing radiation** and the susceptibility to **malignant transformation**.

ATM is also involved in the physiological process of **genetic recombination** (V (D) J recombination) in the development of **T and B lymphocytes**, which may be disrupted by the inability to repair double DNA breaks. Significantly reduced levels of IgE and especially IgA form a typical picture from the point of view of immunology. The reduction may also affect immunoglobulins IgM and IgG2 (or total IgG). From a morphological point of view, we observe **hypoplasia of the thymus and lymph nodes**.

Other manifestations include **cerebellar ataxia**, **telangiectasia** of small vessels and an **increased risk of various malignancies**.



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Source

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. Poslední revize 9. 6. 2006, [cit. 7. 12. 2009]. <<http://www.genetika-biologie.cz/primarni-imunodeficiency>>.

Literature

- BARTŮŇKOVÁ, Jiřina. *Imunodeficiency*. 1. vydání. Praha : Grada, 2002. 228 s. ISBN 80-247-0244-4.