Khediak-Higashi syndrome

Khediak-Higashi syndrome is an inherited (**AR** inheritance) disease caused by mutations in the **LYST gene** (lysosomal trafficking regulator, localization 1q42.1-q42.2). As the name implies, the product of this gene is involved in the formation of lysosomes, primarily affecting the composition of their contents, which is defective in the case of the gene mutation. Both lysosomes and melanosomes are enlarged (sometimes up to giant size) and dysmorphic.

The defective composition of **neutrophilic granulocytes** causes **inefficiency of the phagocytic mechanism**, resulting in **increased susceptibility to certain infections**, mainly **bacterial (mainly Staphyloccocus aureus) and mycotic**. Abnormal granules are also responsible for defects in T-lymphocytes and NK cells. In advanced syndrome, infiltration of tissues may occur as a result of lymphoproliferation.

Affected individuals have **reduced pigmentation** - skin is pale and hair has a light to silver tinge. **Photophobia** and increased sensitivity to sunlight are present. The cause is defective melanocyte granules.

Links

Related links

- Primary immunodeficiency
- Neutropenia in children
- Phagocytosis
- LRO (lysosome related organelles)

Zdroj

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. Poslední revize 9. 6. 2006, [cit. 19. 12. 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

Literature

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