Chediak-Higashi syndrome

Chédiak-Higashi syndrome is an inherited (AR inheritance) disease caused by a mutation in the LYST gene (lysosomal rafficking regulator, localization 1q42.1 – q42.2). As the name suggests, the product of that gene is involved in the formation of lysosomes, mainly affecting the composition of their contents, which is defective in the case of a gene mutation. Both lysosomes and melanosomes are **enlarged** (sometimes to gigantic proportions) and **dysmorphic**.

Defective granule composition of neutrophilic granulocytes causes **inefficiency of the phagocytic mechanism**, which causes **increased susceptibility to certain infections**, especially bacterial (mainly Staphyloccocus aureus) and fungal infections. Abnormal granules are also responsible for defects in T-lymphocytes and NK cells. In advanced syndrome, tissue infiltration may occur due to lymphoproliferative disorders.

Affected individuals have **reduced pigmentation** - the skin is light and the hair has a light to silver tinge. They also suffer from **photophobia** and increased sensitivity to sunlight due to defective melanocyte granules.

Links

Source

Chediak Higashi Syndrome - NORD (National Organization for Rare Disorders). *Home - NORD (National Organization for Rare Disorders)* [online]. Copyright ©2021 NORD [cit. 26.03.2022]. Available from <https://rarediseases.org/rare-diseases/chediak-higashi-syndrome/>

References

Chediak-Higashi syndrome: MedlinePlus Genetics. *MedlinePlus - Health Information from the National Library of Medicine* [online]. Available from <https://medlineplus.gov/genetics/condition/chediak-higashi-syndrome/>

Chediak-Higashi Syndrome: Practice Essentials, Background, Pathophysiology. *Diseases & Conditions - Medscape Reference* [online]. Copyright © 1994 [cit. 26.03.2022]. Available from <https://emedicine.medscape.com/article/1114607-overview>