Chédiak–Higashi syndrome

Chédiak–Higashi syndrome(CHS, OMIM: 214500) is a congenital (AR heredity) disease which is caused by LYST (lysosomal trafficking regulator, localization 1q42.1–q42.2) gene mutation. The product of this gene what its name suggests is involved in lysosomal forming, first of all, it influences the composition of lysosomal content, which is defected in case of mutation. Lysosomes and melanosomes are enlarged (sometimes enormously) and dysmorphic. The defect in the composition of the neutrophilic granules causes the inefficiency of the phagocytic mechanism, which causes higher susceptibility to certain types of infections, particularly to bacterial (mainly Staphyloccocus aureus) and mycotic ones. Abnormal granules are also a reason for T cell and NK cell defects. In advanced syndrome, lymphoproliferation may cause tissue infiltration. Affected individuals show decreased pigmentation - they have white skin and light even silver shaded hair. Photophobia and solar radiation sensitivity are also present. The cause is a melanocyte granules defect.