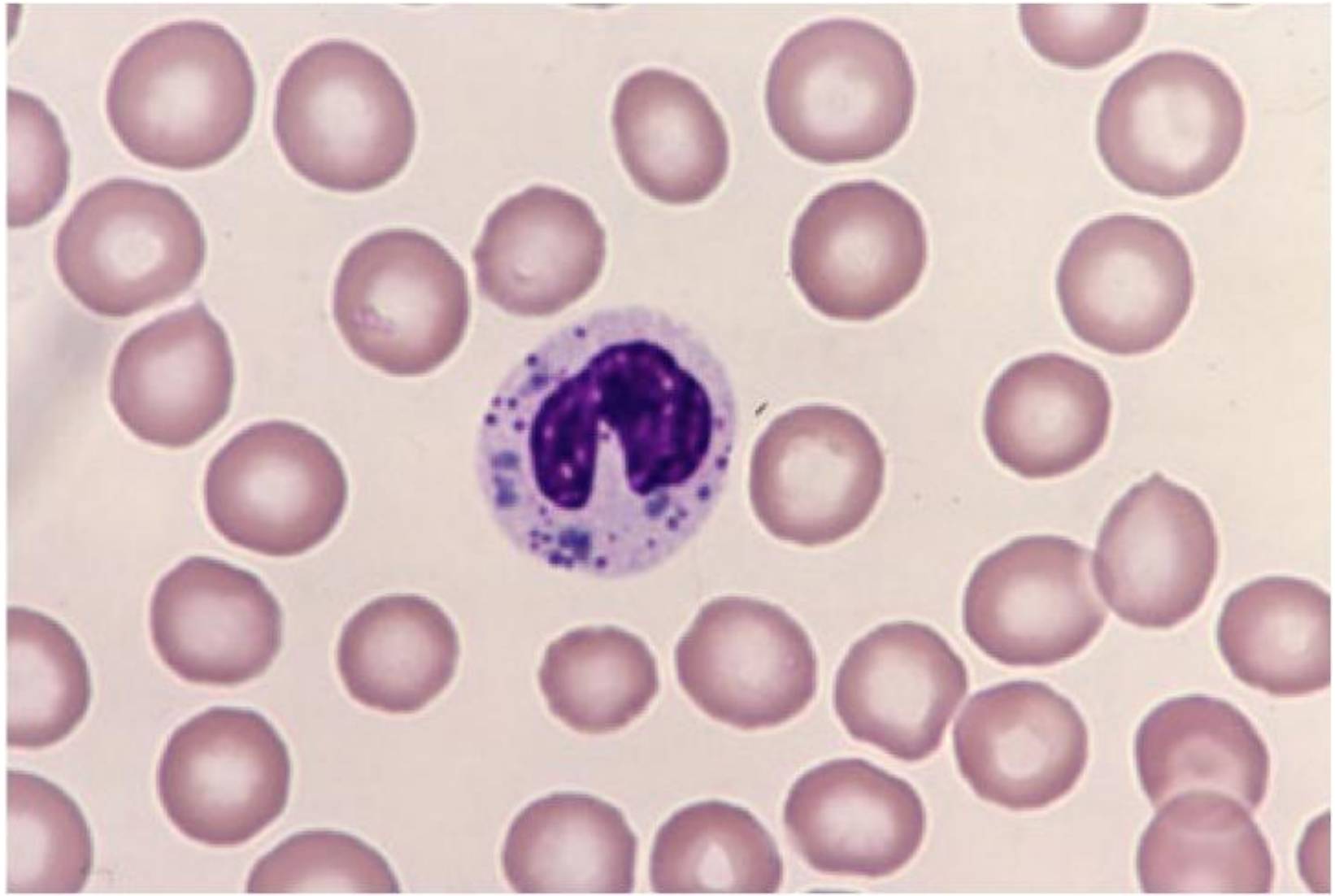
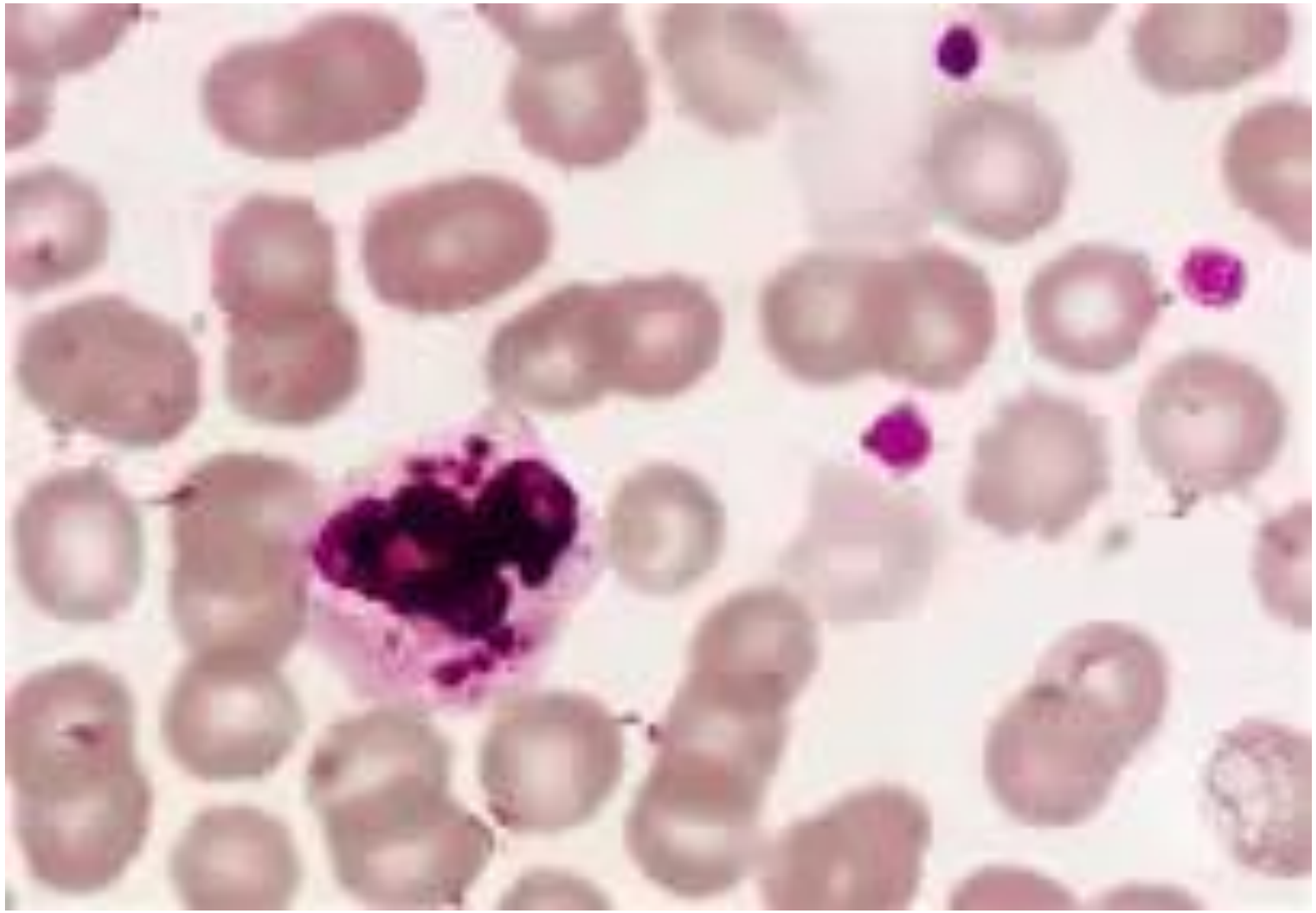
Chédiak-Higashi syndrome

Chédiak-Higashi syndrome is an autosomal recessive disorder featured by recurrent bacterial (pyogenic) infections, oculocutaneous albinism, progressive neurologic abnormalities, mild coagulation defects, and a high risk of developing hemophagocytic lymphohistiocytosis (known as the "accelerated phase" of the disease). The disease is caused by mutations in CHS1/lysosomal trafficking regulator (LYST) gene. The CHS1/LYST protein plays roles in regulating microtubule functions and trafficking intracellular granules. Decreased neutrophil function and neutropenia are characteristic, and coarse granules with activities of myeloperoxidase and acid phosphatase appear in the neutrophil cytoplasm. Dysfunction of NK cells and cytotoxic T cells is also noted. Albinism is caused by disturbance in melanocyte granule transportation.

Ref.: Talbert ML, et al. Chediak-Higashi syndrome. Curr Opin Hematol 2023; 30(4): 144-151. doi: 10.1097/MOH.0000000000000766



Chédiak-Higashi syndrome/ The peripheral blood smear reveals coarse granules in the cytoplasm of neutrophil (May-Giemsa-1) (borrowed from an Internet site).



Chédiak-Higashi syndrome/ The peripheral blood smear reveals coarse granules in the cytoplasm of neutrophil (May-Giemsa-2) (borrowed from an Internet site).