Images from the Haematologica atlas of hematologic cytology: Chediak-Higashi syndrome

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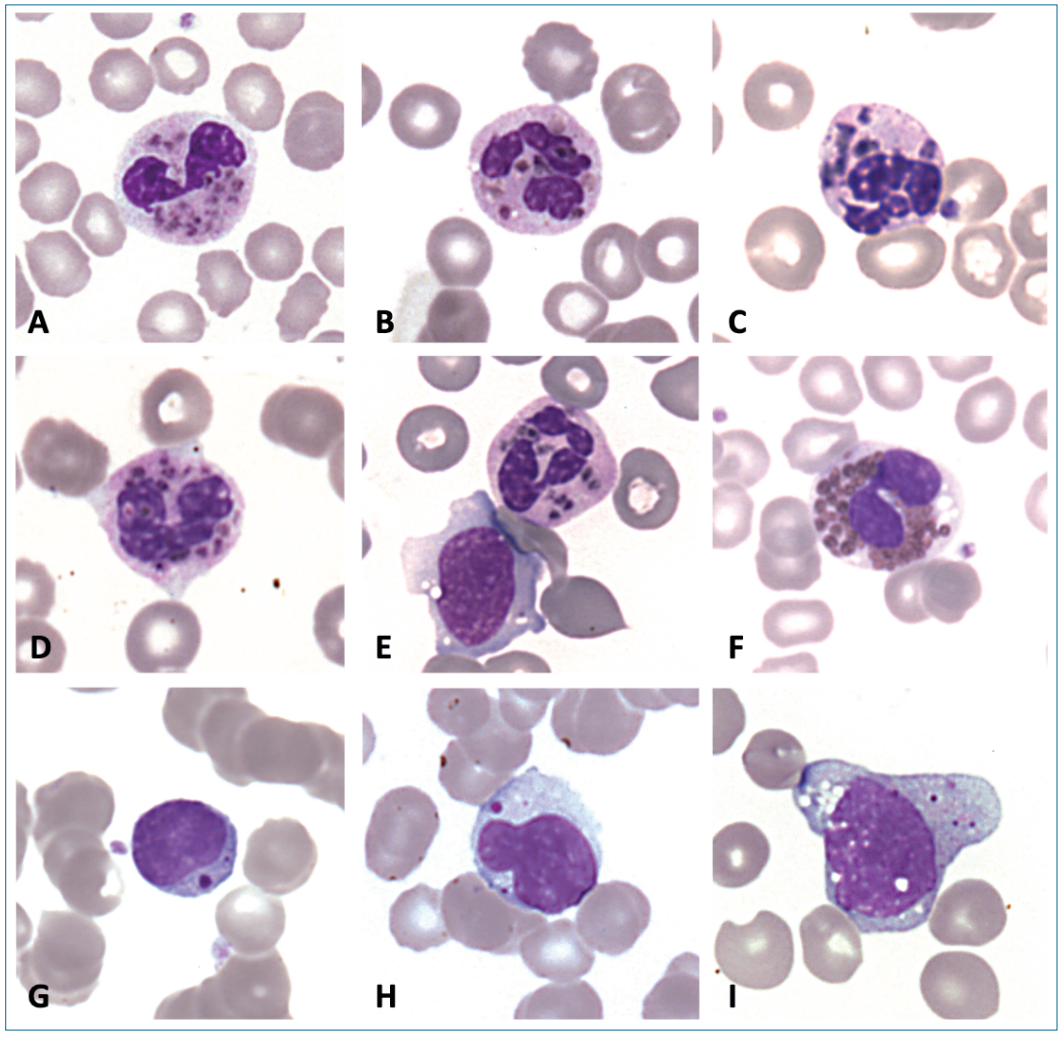

Chediak-Higashi syndrome (CHS) is a complex inherited disorder caused by homozygous or compound heterozygous mutation in the lysosomal trafficking regulator gene *LYST* on chromosome 1q42. A peculiar feature of this condition is the presence of typical, giant granules in leukocyte cytoplasm. Pseudo Chédiak-Higashi granules can occasionally be observed in myeloid elements in a few acquired hematologic disorders such as acute leukemia, myeloproliferative and myelodysplastic syndromes. However, the abnormal granulation in acquired conditions affects a few cells as opposed to generalized involvement of white blood cells in CHS. Giant granules are particularly evident and numerous in neutrophils (A-E). In these cells, they derive from the fusion of azurophilic granules in early precursors, with subsequent incorporation of some specific granules in the myelocyte stage. The mature neutrophils therefore contain both the abnormal azurophilc granules and a variable number of normal specific granules. Concentration of digestive enzymes in giant granules is reduced and this results in defective bactericidal function. Giant granules are observed also in eosinophils (F), basophils, lymphocytes (E, G, H) and monocytes (I). In mononuclear cells the number of granules is usually small. Note the different colorability of the granules in the polymorphonuclear cells and in the mononuclear cells, which reflects their different content. CHS may also show abnormalities of nuclear lobation in granulocytes as seen in A, D and F.[1]

References

1. de Azambuja AP, Balduini CL. Haematologica. 2020; 105(Suppl 1):S225-226.