HEMATOLOGY, TRANSFUSION AND CELL THERAPY

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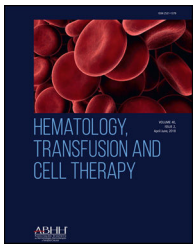

Images in Clinical Hematology

Gaucher-like cells in myelodysplastic syndrome with ring sideroblasts

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An 81-year-old male was hospitalized for fatigue, pallor and transfusion-dependent anemia. His physical exam was negative for lymphadenopathy or splenomegaly. Tests showed hemoglobin concentration of 8 g/dl, platelet count of 160×10^9/l, WBC of 6.2×10^9/l with the following differential: neutrophils 60%, lymphocytes 20%, monocytes 7%, eosinophils 3%, basophils 1% and a low reticulocyte count of 0.5%RBC. Bone marrow aspirate showed increased cellularity mainly due to the proliferating erythroid lineage that displayed dyserthropoietic features (Figure 1A) with no evidence of blasts excess. Few atypical macrophages showing "Gaucher-like" morphological features were also noted (Figure 1B).

Iron stain revealed significantly increased ring sideroblasts which represented 54% of the counted erythroblasts (Figure 1C).

Secondary causes of sideroblastic anemia were ruled out. Additional investigations revealed a normal level of leukocyte β-glucosdiase activity and a positive SF3B1 mutation.

Findings were consistent with a diagnosis of Myelodysplastic syndrome with ring sideroblasts (MDS-RS).

MDS-RS is an MDS characterized by cytopenias, morphological dysplasias and ≥15% marrow ring sideroblasts. In most cases, there is an associated SF3B1 mutation.

Gaucher-like cells have been described in the context of hematologic disorders with high cellular turnover related to malignant proliferation such as chronic myeloid leukemia or multiple myeloma [1–5]. They have also been reported in association with some subtypes of Congenital dyserythropoietic anemia [6]. Proliferation of the erythroid lineage reflecting an underlying inefficient erythropoiesis represents the common finding with this reported MDS-RS case.

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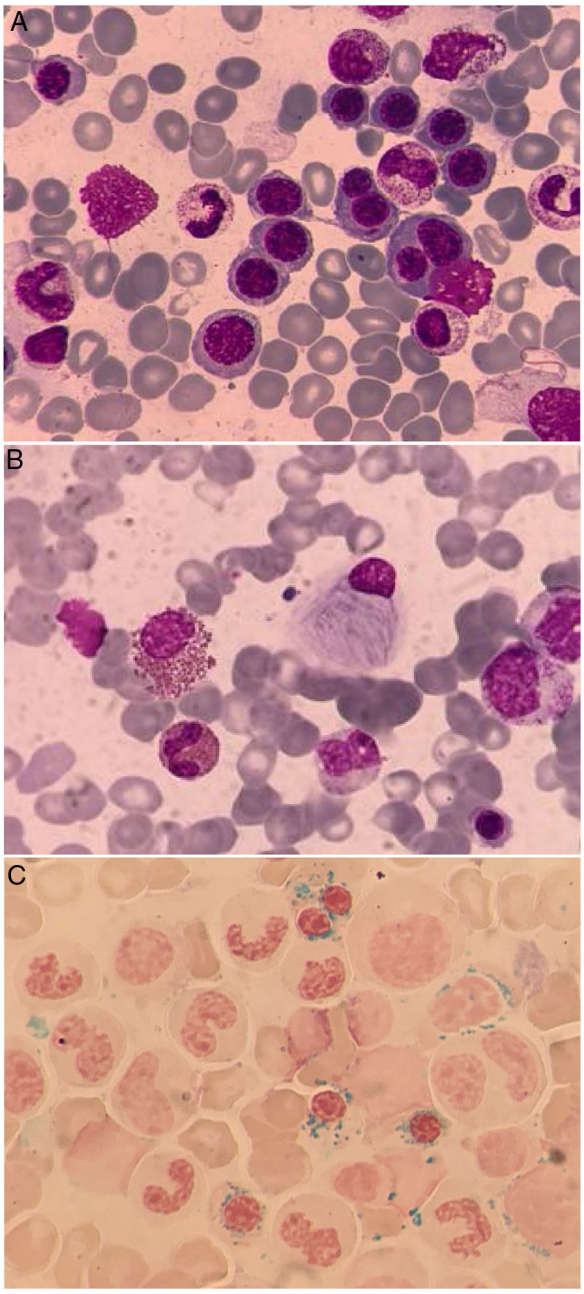

Figure 1 – (A) Bone marrow aspirate showing several erythroblasts displaying dyserythropoietic features (May Grunwald Giemsa stain, 1000×). (B) Atypical macrophage showing Gaucher-like cell morphology (May Grunwald Giemsa stain, 1000×). (C) Iron stain of bone marrow aspirate showing numerous ring sideroblasts (Iron stain, 1000×).

Conflicts of interest

The authors declare no conflicts of interest.

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