Hematology clinicopathological exercise

Case Presentation

A 15-year-old male admitted to the Haematology Department of Baghdad Teaching Hospital with a history of severe anemia and splenomegaly since early childhood that required multiple packed cell transfusions. He was previously diagnosed as hereditary spherocytosis because of splenomegaly and the positive family history of the disease in his family. He visited the Thalassemia Centre of Ibn Al-Baladi Hospital in Baghdad, and hemoglobin electrophoresis and hemoglobin H preparations were repeatedly goes normal. Complete blood picture showed anemia with normal reticulocyte count and normal mean cell volume, mean cell hemoglobin, and mean corpuscular hemoglobin concentration with many nucleated red blood cells but no spherocytosis. Bone marrow aspiration [Figure 1a and b] and its iron stain [Figure 2] were illustrated.

Questions

1. Describe the bone marrow findings
2. What is the most likely diagnosis?

Answers

1. Figure 1a shows bone marrow aspirate with two erythroblasts having binuclearity, one with trinuclearity and one with multinuclearity. Figure 1b shows erythroblasts with intercytoplasmic bridging. Figure 2 shows iron stain of the bone marrow aspirate fragment with increased iron in store (iron overload)
2. These bone marrow findings together with the patient's history of transfusion-dependent anemia with positive family history and the presence of splenomegaly and bone marrow findings of dyserythropoiesis and iron overload pointed to the diagnosis of congenital dyserythropoietic anemia (CDA). The presence in the bone marrow of bi-, tri-, and multi-nuclearity and intercytoplasmic bridging [Figure 1] referred to the diagnosis of type II CDA.

The CDAs are autosomal recessive bone marrow failure syndromes marked by morphological abnormalities in erythroblasts (dyserythropoiesis).

Many patients who have CDA have spent years with an incorrect diagnosis of hemolytic anemia, myelodysplasia, iron deficiency anemia, thalassemia, erythrocyte membrane abnormality, or hemochromatosis. This has exposed the patients to potentially harmful iron supplements, aggressive transfusion or steroids, and cocktails of vitamins. CDAs also present varied management challenges because the ineffective erythropoiesis can be associated with severe iron overload (with secondary organ dysfunction), cholelithiasis, and hepatosplenomegaly.[1]

The key to correct diagnosis is congenital anemia associated with suboptimal reticulocyte response and abnormal bone marrow red cell precursors.

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Conflicts of interest
There are no conflicts of interest.

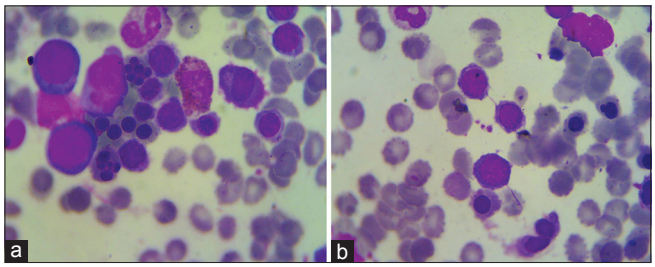

Figure 1: (a) Bone marrow aspirate with two erythroblasts having binuclearity, one with trinuclearity and one with multinuclearity. (b) Erythroblasts with intercytoplasmic bridging

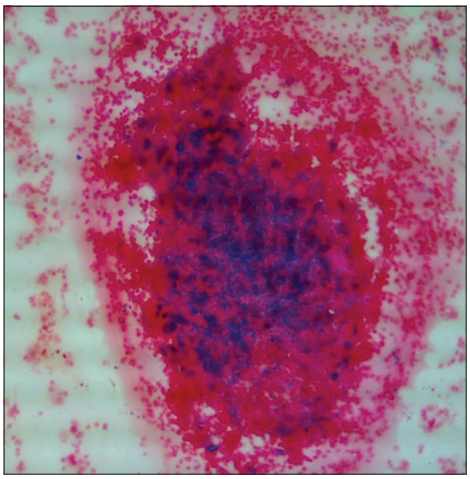

Figure 2: Iron stain of bone marrow aspirate fragment with increased iron in store (iron overload)

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