Images in Haematology

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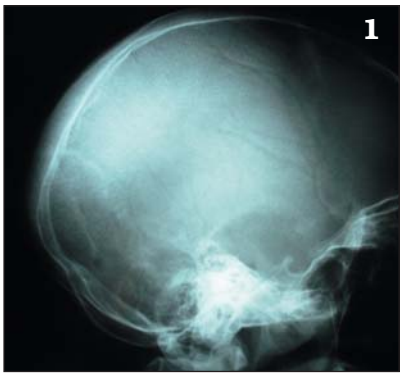

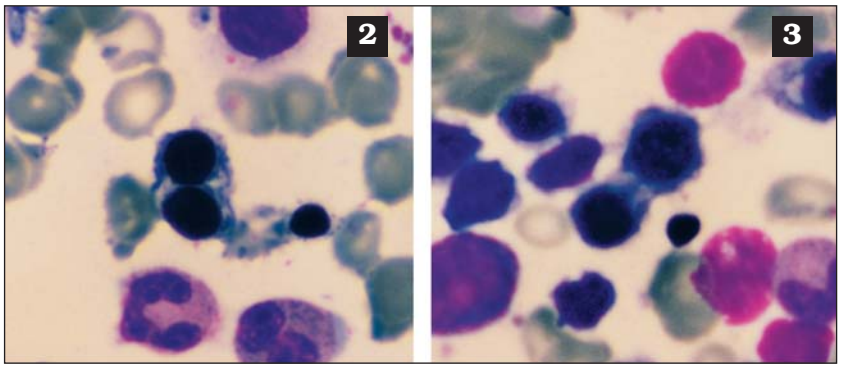

A 13-year-old female patient was admitted to hematology unit because of paleness. On her physical examination; she was pale, there were neither petechiae, purpurae nor ecchymosis. Her hair was loose. There was sulcus anemicus and thalassemic face. Her respiratory and cardiovascular examinations were normal. No organomegaly was detected on abdominal examination but abdominal ultrasonography revealed splenomegaly.

She has no history of blood transfusion, drug usage, fever or jaundice. Her parents are first degree relatives. She has one older sister and a brother who are totally healthy.

Laboratory examination revealed, Hb: 8.7 g/dL, RBC: 3.01 x 10^9/L, MCV: 89 fl, Hct: 27%, RDW: 33.1, WBC: 4700/μL, Plt: 339.000/μL, reticulocyte: 5.4%. Peripheral blood smear revealed the presence of macrocytosis, anisocytosis, poikilocytosis and basophilic stippling. Direct bilirubin: 0.31 mg/dL, total bilirubin: 1.2 mg/dL, direct coombs (-), indirect coombs (-). On Hb electrophoresis HbF: 1.2%, HbA2: 3.4%. P5'N and G6PD were normal. Vitamin B12 and folic acid levels were in normal range. Lateral skull radiogram revealed "hair on end" appereance (Figure 1). Bone marrow aspirate presented cellular marrow with erythroid hyperplasia (57% normoblast). Normoblasts were dyserythropoietic and bi-nucleated (Figure 2,3).

The patient was diagnosed as having Congenital Dyserythropoietic Anemia Type I by the presence of macrocytosis with bone marrow megaloblastoid changes and internuclear chromatin bridges without extreme multinuclearity (differentiates from CDA type III). Red cells were not lysed by acidified sera (HEMPAS) obtained from normal individuals (differentiates from CDA type II).

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