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Vacuolization in Myeloid and Erythroid Precursors in a child with Menkes Disease
Menkes Hastalıklı Bir Çocukta Myeloid ve Eritroid Öncüllerde Vakuolizasyon

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Vacuolization in Myeloid and Erythroid Precursors in a child with Menkes Disease

A five year-old boy, who was under follow-up with a clinical and biochemical diagnosis of Menkes Disease (MD) since ten months of age, was admitted with diarrhea. On examination he had a characteristic cherubic face hypopigmented, sparse hair, hepatosplenomegaly, hypotonia with brisk deep tendon reflexes. Hemogram revealed: Hb:5.5g/dL, Hct:16.2%, RBC:1.69x10¹²/L, MCV:95.8fL, MCH:32.3pg, RDW:19.2%, WBC:2.2x10⁹/L and platelet count;157x10⁹/L. Serum vitamin B12 level was 575pg/mL. Serum copper level was 81µg/dL and serum zinc level was 152µg/dL. Peripheral blood smear revealed 34% PMNL, 62% lymphocyte, 4% monocytes. Bone marrow examination revealed normocellular with megaloblastic changes and widespread cytoplasmic vacuolization in myeloid and erythroid progenitors (Figure 1).

Menkes disease is a neurodegenerative disorder due to mutations in the *ATP7A* gene, which ends-up with deficiency of copper dependent enzymes.

Cytoplasmic vacuoles at myeloid and erythroid lineages have been described in patients with copper deficiency (2), Pearson Syndrome (3) and acute alcoholic intoxication (4). There have also been reports of megaloblastic changes in copper deficiency (2). Herein, we exhibited both erythroid and myeloid vacuolizations and severe megaloblastic changes together in a patient with MD. All of these morphological findings in our patient were attributed to copper deficiency.

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Keywords: Menkes Disease, copper deficiency, vacuolization, bone marrow

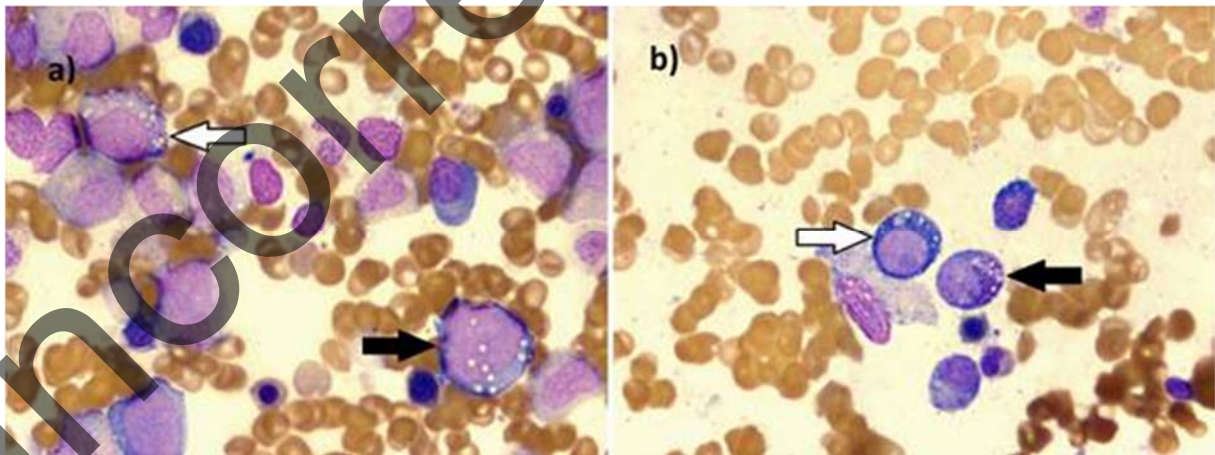


Figure 1. Bone marrow aspiration smears. (a) Cytoplasmic vacuolization in myeloid precursors (white arrow) and erythroid precursors (black arrow) (b) Cytoplasmic vacuolization in myeloid precursors (black arrow) and erythroid precursors (white arrow) (MGG-Giemsa stain, original magnification 100x)