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Gaucher disease in an adult: A rare cause of hepatosplenomegaly in adults

Volkan Karakus,[1] Yelda Dere,[2] Ozcan Dere,[3] Fahri Sahin,[4] Nazan Ozsan[5]

[1]Department of Hematology, Mugla Sıtkı Kocman University Training and Research Hospital, Mugla, Turkey
[2]Department of Pathology, Mugla Sıtkı Kocman University Faculty of Medicine, Mugla, Turkey
[3]Department of Surgery, Mugla Sıtkı Kocman University Faculty of Medicine, Mugla, Turkey
[4]Department of Hematology, Ege University Faculty of Medicine, Izmir, Turkey
[5]Department of Pathology, Ege University Faculty of Medicine, Izmir, Turkey

A 19-year-old woman with no significant medical history presented with weakness. Physical examination revealed hepatosplenomegaly with no peripheral lymphadenopathies. Laboratory tests revealed anemia (Hb: 11.9 mg/dL) and thrombocytopenia (platelet: 108000/mm^3). Other results were normal. Peripheral blood smear revealed 60% PMNL (polymorphonuclear leukocytes) and 30% lymphocytes, with slightly polychromatic erythrocytes, and 8–9 thrombocytes in every field. Color Doppler ultrasonography (USG) revealed the liver to be 195 mm in size and the spleen to be 175 mm in size, with no paranchymal abnormalities in either. Hematological tests revealed negative gene mutations for JAK2 and t(9;22). Therefore, bone marrow aspiration and biopsy was performed. In bone marrow aspirates, macrophages with abundant sea-blue "crumpled tissue paper" cytoplasm were observed (Fig. 1, arrows). In the bone marrow biopsy, cellularity was 50%, consisting of mostly macrophages (Fig. 2, circle). These cells were positive for CD68, lyzozyme, and TRAP, but negative for S100 and CD1a. Histomorphologic features were reported to be concordant with those of Gaucher disease. After the diagnosis, enzymatic activity and level were investigated and the results were as follows: β-glycosidase, 0.08 nmol/mL/h (normal range for adults: 0.94–5.29) and cytotriosidase, 193 nmol/mL/h (normal range for adults: 0–1074). In addition, genetic studies showed heterozygous mutations (p. N370S) and (p. L444P) and confirmed the diagnosis. Cerezym® treatment was started. The patient recovered and remained healthy on her first-year follow-up with the following findings: Hb 12.5 mg/dL, platelet 163000/mm^3, liver size 178 mm, and spleen size 137 mm (on her last USG).

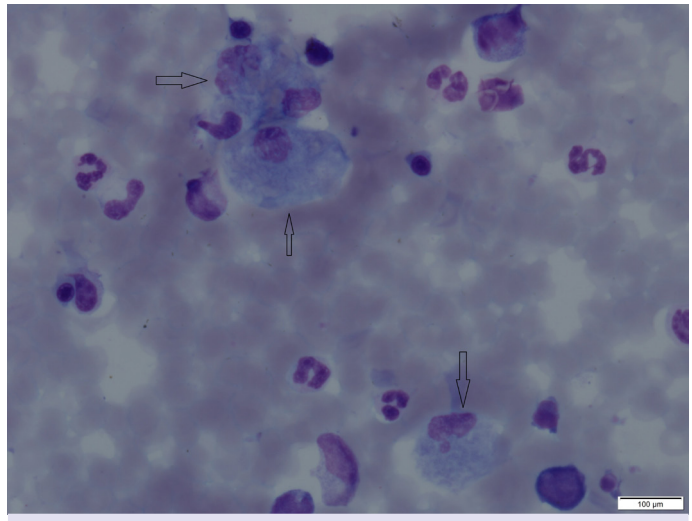

Figure 1. Large histiocytes with crumpled tissue paper cytoplasm. Giemsa stain (×400).

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Correspondence: Dr. Yelda DERE. Mugla Sitki Kocman Universitesi Tip Fakultesi, Patoloji Anabilim Dali, Mugla, Turkey.
Tel: +90 505 465 31 98 e-mail: yeldamorgul@gmail.com

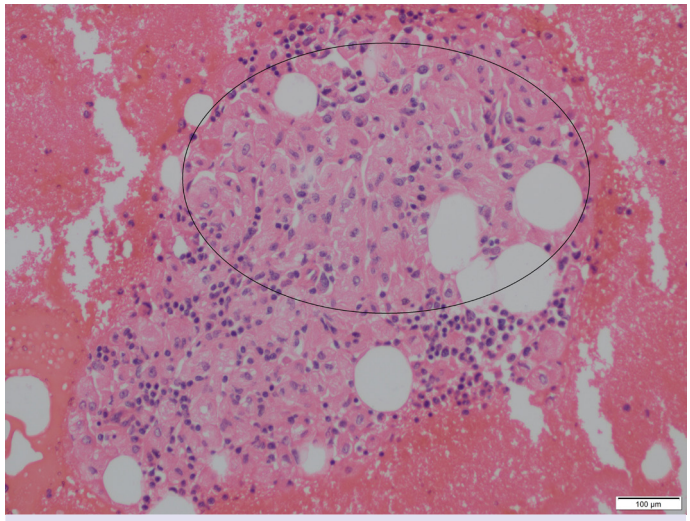

FIGURE 2. Histiocytes with pink pale cytoplasm in the bone marrow on cell block prepared from the aspirate. H&E stain, ×400.

Gaucher disease is the most common autosomal recessive lysosomal storage disease worldwide and is generally diagnosed in children with splenomegaly and cytopenias. The diagnosis depends on the demonstration of low enzymatic activity of glucocerebrosidase. Hepatosplenomegaly is the most common finding; Gaucher disease should be kept in mind in patients with unexplained hepatosplenomegaly. Cytopenia secondary to bone marrow infiltration and hypersplenism may be seen. The gold standard for the diagnosis is to measure the enzymatic activity of glucocerebrosidase. Enzymatic replacement is the main treatment modality for these cases. Gaucher disease is a progressive disease and generally diagnosed in childhood. However, in asymptomatic adults, progression may be slow or the disease may regress spontaneously.