



Visceral Leishmaniasis and Gaucher Disease: Case Report of an Usual Association

Ghizlane Zoulati*, Ramatou Yèya Maïga, Majdouline Elkhayat, Moncef Hassani Amrani

Hematology Department, Hassan II University Teaching Hospital of Fez, Morocco

***Corresponding author:** Ghizlane Zoulati, Hematology Department, Central Laboratory of Medical Analysis, Hassan II University Teaching Hospital of Fez, Morocco, BP 1835 Atlas Fez, Morocco

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Abstract

Introduction: Visceral Leishmaniasis (VL) is a public health problem of a significant burden in Morocco. It was described in association with many diseases, particularly as an opportunistic infection. Nevertheless, the association with Gaucher's Disease (GD) remains exceptional.

Case Report: An eight years old Moroccan girl presented with pancytopenia and splenomegaly, in which bone marrow contained Leishman-Donovan bodies and foamy vacuolated macrophages evoking Gaucher cells. GD was confirmed by deficient betaglucoocerebrosidase activity in the peripheral leukocytes leading to the diagnosis of an original association, VL and GD.

Conclusion: We are reporting this case to make aware of this very rare co-existence to avoid misdiagnosis, and also to highlight that the presence of Gaucher cells or pseudo-Gaucher cells in bone marrow should not be obscuring a possible other pathology with hematopoietic involvement such as VL, especially in leishmania endemic areas.

Keywords: Bone marrow; Cytologist; Gaucher's disease; Visceral leishmaniasis

Introduction

Visceral Leishmaniasis (VL) has been among the most important health problems in Morocco. This parasitic disease is caused by the proliferation of a protozoan parasite *Leishmania* in the reticulo-histiocyte system [1]. VL is frequently reported in patients with immunocompromised states due to HIV infection, post organ transplantation, solid tumors, hematological malignancies and chemotherapy [2]. However, the association with Gaucher's Disease (GD) remains exceptional in literature, it was described only once in an Indian child [3]. By reporting this case, we aim to spread awareness about possible association of VL and GD and to discuss some aspects and implications of their coexistence.

Case Report

An eight-year old child presented with a four years history of pallor, weakness, bone pain, decreased appetite, poor growth and abdominal distension. Clinical examination showed a severe pallor, a temperature of 38.2 °C while other vital signs were stable.

A massive splenomegaly along with hepatomegaly were also observed. Anthropometric measurements found a stature-ponderal delay (below two standard deviations). Laboratory reports revealed pancytopenia with haemoglobin of 5.5 g/dl, platelet count of 10500/mm³, leucocyte count of 1550/μl; and peripheral blood smear shows microcytic hypochromic anaemia with pancytopenia (Figure 1). Other laboratory findings, including liver function tests, were normal. Due to the patient's pancytopenia and splenomegaly, bone marrow aspirate was performed, which showed a slightly hyperplastic marrow with trilineage hematopoiesis and extracellular Leishman-Donovan bodies (amastigote form) (Figure 2).

Moreover, numerous macrophages with eccentric nuclei, abundant blue-gray cytoplasm containing fibrillar inclusions, are seen in medullar smear in addition to vacuolated cells (Figures 3-5). These morphologic findings are compatible with storage diseases features; particularly, the atypical macrophages are evoking Gaucher cells. Therefore, enzyme testing was performed showing reduced activity of β-glucocerebrosidase enzyme in peripheral blood lymphocytes to 0,32 mkat/kg de proteins (normal range: 2,1- 3,8). Thus, the diagnosis of an original association was made, GD and VL.

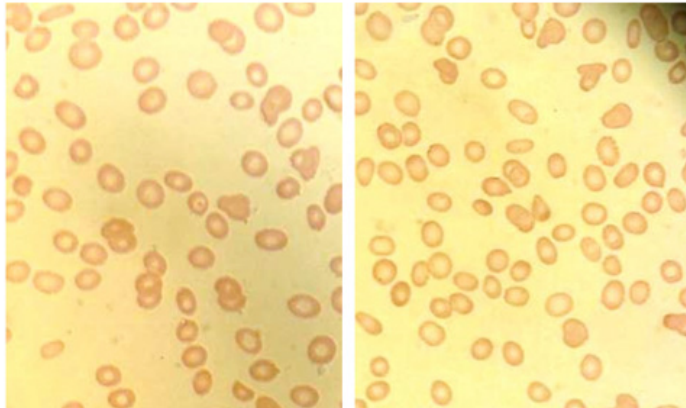


Figure 1: Peripheral blood smear showing microcytic hypochromic anemia and pancytopenia (MGGiemsa stain x1000).

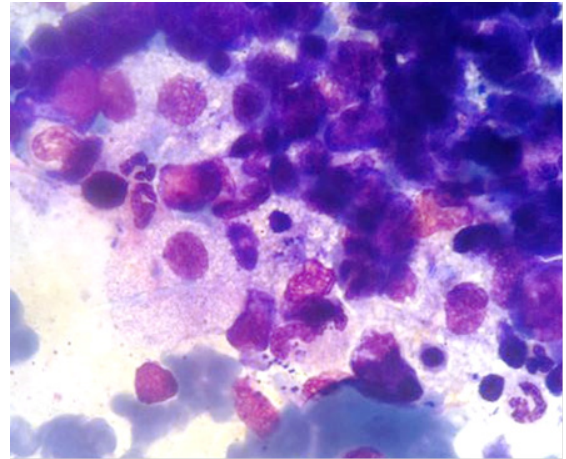


Figure 4: Bone marrow smear showing atypical macrophages with eccentric nuclei, abundant cytoplasm containing fibrillar inclusions. (MGG stain stain x1000).

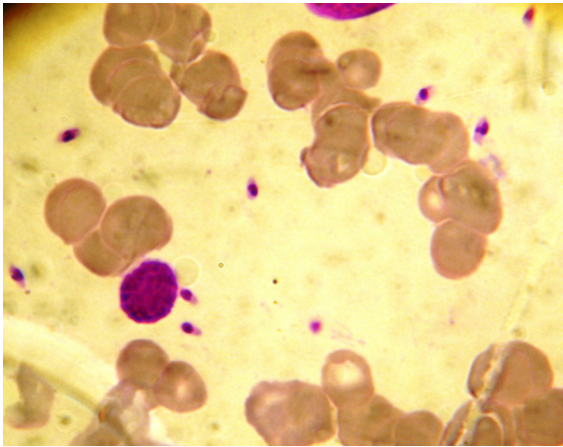


Figure 2: Bone marrow smear showing extracellular LD body of amastigote form. (MGGiemsa stain x1000).

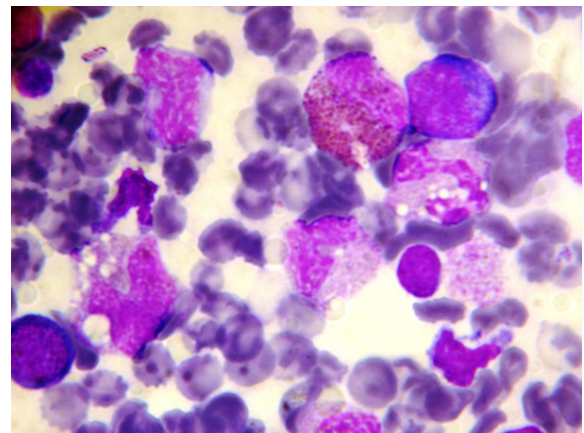


Figure 5: Vacuolated cells on bone marrow smear. (MGG stain stain x1000).

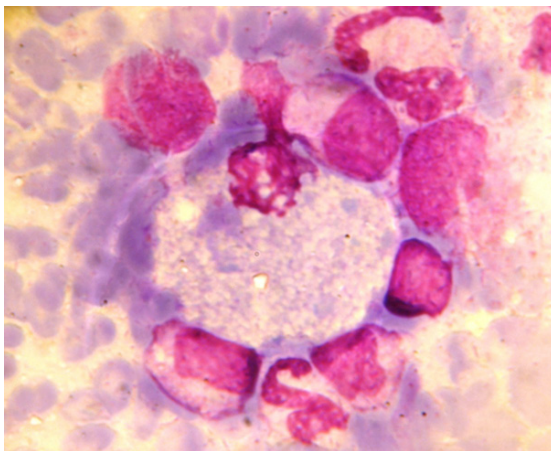


Figure 3: Gaucher cell on bone marrow smear. (MGG stain stain x1000).

Discussion

Gaucher's Disease (GD) is an autosomal recessive lysosomal storage disease resulting in reduced activity of the lysosomal enzyme glucocerebrosidase which is involved in the breakdown of glycosphingolipids. Its deficiency results in accumulation of glucosylceramide, particularly in cells of the macrophage lineage [4]. The deposit of glucocerebrosides in the cytoplasm of histiocytes leads to the formation of atypical vacuolated foamy macrophages known as "Gaucher cells". Typically, Gaucher cells are large cells measuring 50 to 60 μm in diameter with a small eccentric nucleus and fibrillary cytoplasm [5,6]. Because of constant presence of hematological symptoms in GD, hematologists have always been at the forefront of specialists, who performed initial diagnostics of GD. Gaucher cells, the lipid-laden storage macrophages, are the

pathologic hallmark of GD [7]. However, Cells morphologically very similar to Gaucher cells can found in infectious diseases and hematologic abnormalities such as chronic myeloid leukemia, malignant lymphoma, multiple myeloma and myelodysplastic syndrome [5,6].

Therefore, measurement of glucocerebrosidase activity in leukocytes, or in cultured fibroblasts obtained by skin biopsy, is the gold standard for the diagnosis of GD [4,8]. To the best of our knowledge, there is only one report case of VL associated to GD in an indian child [3]. The case presented herein is an extremely rare case of association of VL and GD, right from the beginning, which arises the question about the possible physiopathological linkings between the two disorders. GD has occasionally been described with cancer, gammopathies and hematologic malignancies [9,10]. Furthermore, it is linked to an increased risk of Parkinson disease [11]. However, GD is not recognized to induce a breeding ground for opportunistic infections like leishmaniasis. Therefore, a causal relationship between these two pathologic entities remains speculative. We are reporting this unusual case to make aware of this very rare co-existence to avoid misdiagnosis, and also to highlight that the presence of Gaucher cells or Pseudo-Gaucher cells in bone marrow should not be obscuring a possible other pathology with hematopoietic involvement such as VL, especially in leishmania endemic areas.

Conclusion

Finally, we would like to emphasize the importance of early diagnosis, to highlight the clinical manifestations and biological findings so that early treatments by enzyme replacement therapy can be started delaying the complications. Moreover, awareness of possible association of VL and GD, meticulous and methodical study of bone marrow smear along with relevant additional investigations based on clinical findings are necessary for early diagnosis, fast institution of the treatment, and consequently, the improvement of the forecast and the life quality of patients.

Authors' Contributions

G Zoulati, the principal author, made major contributions in writing the manuscript.

M Elkhiyat took the figures photography.

RYM and MHF analyzed and interpreted the patient data and the reviews of the literature.

All authors read and approved the final manuscript.

Conflict of Interest

The authors declare that they have no potential conflict of interest relevant to this article.

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