Splenomegaly and Pancytopenia in an Elderly Male

A 62-year-old male presented with generalized weakness and abdominal discomfort since the past 4 months; there were no other significant complaints. On examination, the patient was pale and his abdominal palpitation revealed an enlarged, firm, nontender splenomegaly 10-cm below the costal margin. His complete blood count results were as follows: white blood cell (WBC) count, 3.0×10^9/L; red blood cell count, 2.37×10^{12}/L; hemoglobin, 6.7 g/dl; and platelet count, 30×10^9/L. A peripheral blood smear demonstrated normocytic normochromic red cells with a WBC differential count of 22% polymorphs, 70% lymphocytes, 2% monocytes, 1% eosinophils and 5% atypical cells [Figure 1a and b]. Bone marrow aspiration was not possible ("dry tap"). The hematoxylin and eosin staining of the trephine bone marrow biopsy showed an infiltration by cells with a characteristic "fried egg" appearance of the cytoplasm [Figure 2].

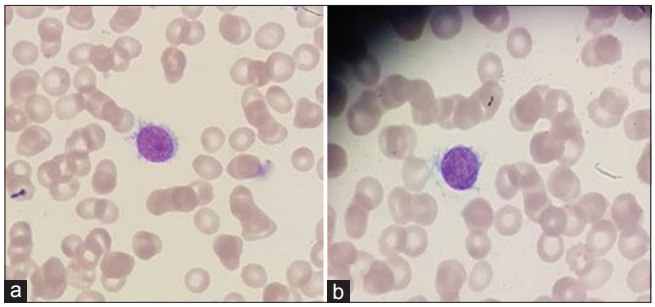

Figure 1: (a and b) Peripheral blood smear showing an atypical cell with multiple cytoplasmic projections

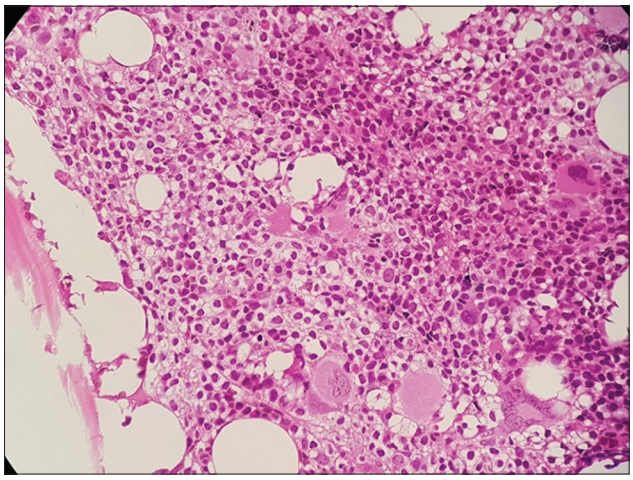

Figure 2: Hematoxylin and eosin staining of the bone marrow biopsy

QUESTIONS

1. What are the atypical cells seen in Figure 1?
2. What are the confirmatory tests and what is the final diagnosis?

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ANSWERS

1. Hairy cells
2. The final diagnosis is hairy cell leukemia (HCL). Immunophenotyping and immunohistochemistry analysis are critical in establishing this diagnosis.

DISCUSSION

HCL is an uncommon chronic lymphoid leukemia that constitutes approximately 2% of all adult leukemias.[1] HCL affects males more commonly than females (male: female ratio = 4:1) and the median age at diagnosis is 50–59 years.[2] In general, HCL patients present with splenomegaly (>3 cm below the costal margin) and pancytopenia without lymphadenopathy and with the associated fatigue, abdominal pain in the left upper quadrant, fever and/or infections.[1,2]

Identification of the hairy cells can be made by careful examination of the peripheral blood smears and assessment of the complete blood count, as monocytopenia is characteristically observed.[1,2] Hairy cells are lymphoid cells having an oval or bean-shaped nucleus with loose chromatin and abundant pale blue cytoplasm with circumferential hair-like projections.[2,3] The characteristic immunophenotypic profile includes positive expression of CD19, CD20, CD22 and CD200 antibodies as well as positivity for at least three of the following antibodies: CD11c, CD25, CD103 and CD123. In contrast, for CD5, CD10, CD23, CD27 and CD79b antibodies, hairy cells are negative or dim.[1,3]

A bone marrow examination is necessary to understand the extent of infiltration as well as to assess response to treatment; however, bone marrow aspiration is extremely difficult to obtain, as it is usually a "dry tap."[3] Nonetheless, a bone marrow trephine biopsy would help determine the degree of infiltration and the presence of BRAF V600E somatic mutation, which is present in about 90% of HCL cases. Immunohistochemical stains using CD20, CD76, annexin A1 and tartrate-resistant acid phosphatase stain would support HCL diagnosis and highlight the extent of lymphoid infiltrates.[1,3] The differential diagnosis of HCL includes the HCL variant, splenic marginal zone lymphoma, chronic lymphocytic leukemia, prolymphocytic leukemia and mantle cell lymphoma.[1] However, with the support of immunophenotyping and immunohistochemistry, HCL diagnosis can be established.

Over the decades, HCL treatment modality has changed from splenectomy to recombinant interferon-alpha to adenosine deaminase inhibitor deoxycoformycin (pentostatin). However, purine analogs are the mainstay of HCL therapy, of which 2-chlorodeoxyadenosine (cladribine) is the most active with very high response rates.[3,4]

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Conflicts of interest
There are no conflicts of interest.

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