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CASE REPORT

Atypical Presentation of Hairy Cell Leukemia Masquerading as Isolated Thrombocytopenia: A Diagnostic Odyssey Emphasizing Vigilance in Healthcare

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Abstract

Hairy Cell Leukemia (HCL) is a rare chronic B-cell lymphoproliferative neoplasm typically presenting with splenomegaly and pancytopenia. This case report discusses the atypical presentation of HCL in a 29-year-old male who exhibited isolated thrombocytopenia without classical symptoms leading to initial misdiagnosis as dengue delayed recognition until immunophenotyping revealed monotypic B-cell markers characteristic of HCL. This case underscores the challenges in diagnosing atypical HCL emphasizing the importance of vigilance, detailed morphological assessment and advanced diagnostic tools for early detection and contributes to awareness of non-classical HCL presentations.

Key Words

Leukemia, Hairy cell, Lymphoproliferative

Introduction

Hairy cell leukemiaaka Leukemic reticuloendotheliosis is a rareform of chronic B-cell lymphoproliferative neoplasm (LPN) of elderly that accounts fornearly 2% of all adult leukemias. Hairy cell leukemia (HCL) is a rare chronic B-cell lymphoproliferative disorder with an annual incidence of about 0.3 cases per 100,000 persons in most populations.[1] It is characterized by the proliferation of small sized lymphoid cells with cytoplasmic circumferential fine "hair-like" projections seen in the peripheral blood, bone marrow andsplenic red pulp. The hallmark presentation of Hairy cell leukemia (HCL) are splenomegaly, pancytopenia(s) ormonocytopenia and circulating CD19+, bright CD20+/CD22+/CD200+, CD25+, CD103+ and CD11c+ hairy cells demonstrated classically by flowcytometry.[2]

We report the diagnostic journey of a 29-year-old male patient with isolated thrombocytopenia illustrating the complexity of medical diagnoses and the need for comprehensive evaluation when faced with unusual or atypical clinical presentations.This case was finally diagnosed as Hairy Cell Leukemia (HCL) and emphasizes the importance of vigilance in healthcare.

Case Report

A 29-year young male patient initially presented in our hospital with isolated thrombocytopenia, characterized by a platelet count of 1.2 lacs/ iL. The patient was completely asymptomatic and had noted thrombocytopenia a general health checkup done almost 10 days prior to presentation. Other Complete Blood Count parameters including white blood cell count (WBC) of 4800/iL, a red blood cell count (RBC) of 5.12 million /iL, hemoglobin (Hgb) levels of 14.8 g/dL, and a hematocrit (Hct) of 43.7%. Other

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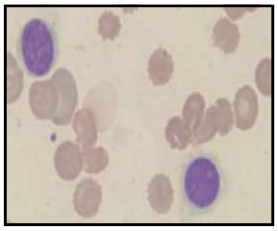


Figure 1

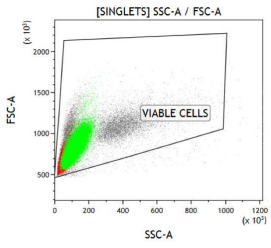
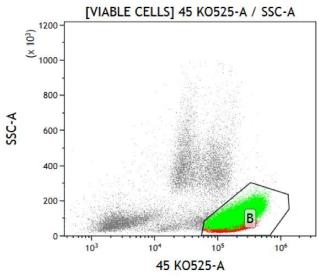


Figure 2(a)



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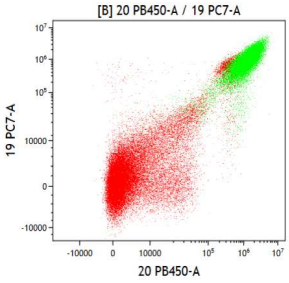

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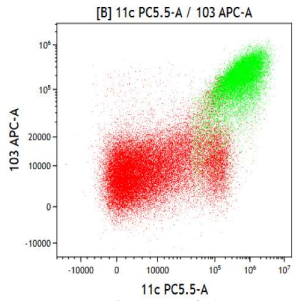

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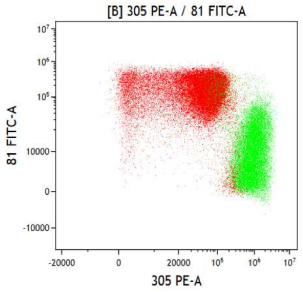

Figure 2(e)

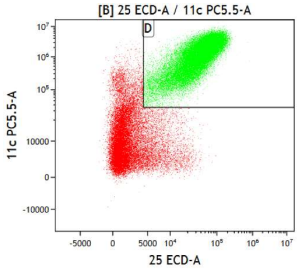

Figure 2(f)

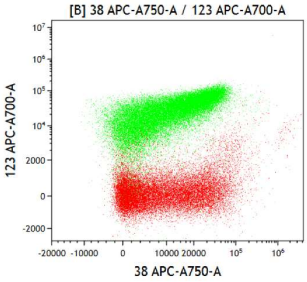

Figure 2(g)

parameters like mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), mean corpuscular hemoglobin concentration (MCHC), and red cell distribution width (RDW) fell within the biological reference interval, it a case of isolated thrombocytopenia. Initially, due to the prevalent dengue season in the Delhi NCR region, the patient's thrombocytopenia was attributed to a potential dengue infection. The platelet count failed to improve even after the patient's clinical condition stabilized, further investigation was necessary.

The peripheral blood smear examination revealed the presence of rare circulating hairy cells, although these cells were not immediately recognized as a diagnostic clue. **(Figure 1)** It was only after the persistence of thrombocytopenia.

The immunophenotyping was suggested and revealed a unique cellular profile:revealed a monotypic B-cell population (viable singlet) showing expression of CD45, CD19, CD 20, CD11c, CD103, CD 25, CD 81, CD305, CD 12, CD 38 with clear monoclonality demonstrated through light chain restriction. **(Figure 2a-g)** This diagnosis of HCL, a rare form of leukemia, was confirmed. Notably, this patient did not exhibit splenomegaly, lymphadenopathy, or other typical symptoms, making the case even more exceptional.

Discussion

HCL is an indolent, small mature B lymphoid malignancy accounting for 2% of all leukemias. The three most diagnostic findings are presence of splenomegaly, cytopenia(s) and dry tap bone marrow resulting from marrow fibrosis.[3] HCL patients may be asymptomatic or develop symptoms of cytopenia, particularly infections.The exact prevalence of atypical presentation of HCL is difficult to be determined because most cases were published as incidental reports.In unsuspected cases with atypical presentation, the ideal approach for diagnosis is careful morphological identification of typical hairy cells on peripheral smears and bone marrow aspiration/biopsy which should further be confirmed upon characteristic immunophenotypic profiles, as in our case. Hairy cells typically range 10-15 µm in diameter having central/ eccentric round to oval and often indented nuclei with reticular/netlike chromatin, indistinct nucleoli, paleblue cytoplasm with fine, "hair-like" ruffled borders. HCL show positive staining for tartrate-resistant acid phosphatase. A combination of immunophenotypic markers typically expressed byhairy cells i.e. CD19, CD22, CD79bwith brighter expression of CD20, along with co-expression of CD103/CD123/CD25/CD11c confirms the diagnosis.(4) As hairy cell leukemia may present with isolated presentation which could result in a diagnostic dilemma.Differential diagnoses of HCL include chronic lymphocytic leukemia, prolymphocytic leukemia, splenic marginal zone lymphoma, HCL-v and mantle cell lymphoma, which can be excluded based on characteristic morphological and immunophenotypic features.[5]

This case highlights the need for thorough diagnosis

when symptoms are atypical, the value of advanced diagnostic tools like immunophenotyping, and the importance of collaboration in medicine. It showcases the unpredictability of diagnoses and the dedication of medical professionals in achieving accurate diagnoses and guiding treatment.The aim of this case report is to highlight non-classical presentations of HCL and the importance to have high index of suspicion to consider a diagnosis of HCL based on morphological features even in the absence of classical clinical features.

In conclusion, this case posed a diagnostic challenge as the patient had isolated thrombocytopenia. This case is important because it creates awareness of this uncommon presentation of HCL and emphasizes that the best approach in diagnosing HCL is to give careful attention to morphological details while interpreting peripheral blood, as in our case, which can prompt detailed evaluation with immunophenotyping in such cases for early diagnosis and management of the patient.

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