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Case Report

A rare confluence: chronic myeloid leukemia in a pediatric case of beta thalassemia

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ABSTRACT

Chronic myeloid leukemia (CML) is rare in children and extremely uncommon in patients with underlying hemoglobinopathies such as beta thalassemia. This report describes a 12-year-old beta thalassemia patient, who presented with fever, pallor and weakness, and was found to have marked hepatosplenomegaly and leukocytosis. Peripheral blood showed leukocytosis with increased myeloid precursors and bone marrow evaluation showed hypercellular marrow with myeloid hyperplasia and basophilia, consistent with a chronic myeloproliferative neoplasm. Karyotyping revealed balanced reciprocal translocation t (9;22) and reverse-transcriptase polymerase chain reaction (RT-PCR) detected BCR-ABL1 p210 fusion transcript, confirming the diagnosis of chronic phase CML in a background of beta thalassemia. The patient had elevated serum ferritin, indirect hyperbilirubinemia and mildly deranged transaminases, attributable to chronic transfusions and iron overload. After initiation of appropriate therapy, leukocyte counts normalized within one month along with disappearance of circulating immature myeloid precursors. This case highlights the importance of maintaining a high index of suspicion for CML in thalassemic patients with unexplained leukocytosis and splenomegaly, and underscores the need for integrated morphologic, cytogenetic and molecular work-up to distinguish disease progression from a second primary hematologic malignancy.

Keywords: Leucocytosis, Peripheral blood, Hepatosplenomegaly, RT(PCR), Iron overload, BCR-ABL1

INTRODUCTION

Chronic myeloid leukemia (CML) is a clonal myeloproliferative neoplasm characterized by the BCR-ABL1 fusion gene, accounting for a small proportion of childhood leukemias, with a reported incidence far lower than in adults.[1] Beta thalassemia major is a hereditary hemoglobinopathy requiring lifelong transfusions, and long-term survivors are increasingly recognized to develop secondary malignancies, although the coexistence of CML and beta thalassemia remains exceedingly rare.[2,3] The present study was conducted to study awareness and perception about dog bite among the population in the rural area of Maharashtra.

CASE REPORT

A 12-year-old boy, known case of transfusion-dependent beta thalassemia, presented with complaints of fever, generalized weakness and easy fatigability. On examination, he had marked pallor and hepatosplenomegaly.

Initial laboratory evaluation revealed hemoglobin 9.3 g/dl, total leukocyte counts 81,130/µl and platelet count 199,000/µl. Peripheral smear showed leukocytosis with left shift comprising of 3% blasts, 10% myelocytes, 7% metamyelocytes and 15% band forms, along with basophilia and background anemia, suggesting a

myeloproliferative process (Figure 1). Biochemical parameters showed serum ferritin of 4134 ng/ml, total bilirubin 2.4 mg/dl with indirect bilirubin 1.8 mg/dl, serum glutamic pyruvic transaminase (SGPT) 106 U/l, serum glutamic oxaloacetic transaminase (SGOT) 108 U/l and alkaline phosphatase 256 U/L, indicative of severe iron overload and mild hepatic dysfunction in a chronically transfused child.

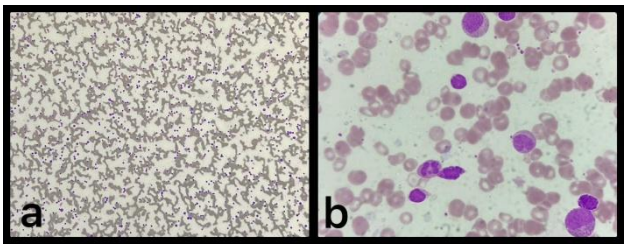

Figure 1: Peripheral smear showing leukocytosis with left shift, increased myeloid precursors and microcytic hypochromic RBCs: (a) 10x view (b) 100x view.

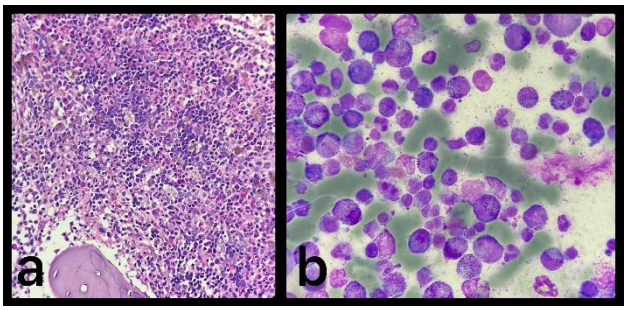

Figure 2: (a) Bone marrow biopsy showing increased myeloid precursors and hemosiderin-laden macrophages. (b) Bone marrow aspiration showing marked myeloid hyperplasia and basophilia.

Bone marrow biopsy revealed a hypercellular marrow with prominent myeloid precursors, reduced erythroid series and megakaryocytes, along with numerous hemosiderin-laden macrophages, suggestive of chronic myeloproliferative disorder with chronic transfusional hemosiderosis (Figure 2a). Bone marrow aspiration smears also showed similar findings, along with basophilia (Figure 2b). Blasts were not increased, in keeping with chronic phase disease.

Karyotyping revealed balanced reciprocal translocation between chromosomes 9 and 22 (Figure 3). Molecular evaluation by RT-PCR on peripheral blood leukocytes showed the presence of BCR-ABL1 p210 fusion transcript (72.76%), confirming the diagnosis of CML in a background of beta thalassemia. After initiation of appropriate tyrosine kinase inhibitor–based therapy and optimization of chelation, repeat hemogram at one month showed a total leukocyte count of 5,500/µl with no blasts or increased immature myeloid forms, indicating an early hematologic response. The patient continues to be followed for both CML response monitoring and iron overload management.

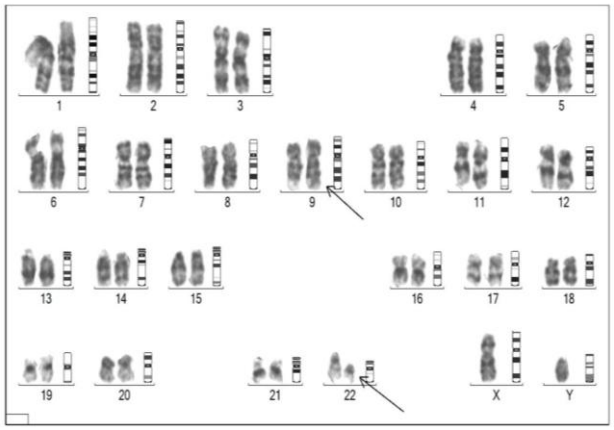

Figure 3: Karyotyping showing reciprocal translocation between chromosomes 9 and 22.

DISCUSSION

CML in children is uncommon, and its occurrence in patients with beta thalassemia is particularly rare, posing diagnostic challenges, as splenomegaly, anemia and even mild leukocytosis can be attributed to the underlying hemoglobinopathy.[2-4] In this case, the presence of marked leukocytosis with left-shifted myeloid cells and basophilia prompted bone marrow and molecular work-up, which demonstrated typical features of chronic phase CML and BCR-ABL1 p210 positivity in a marrow already altered by chronic transfusions and iron overload.[1]

Long-term survivors of beta thalassemia major are at significantly increased risk of developing secondary hematologic and solid malignancies, with lymphomas, acute leukemias, and hepatocellular carcinoma being the most commonly reported complications.[3] However, CML remains a very rare association and is usually reported as isolated cases.[3-5] The pathophysiology underlying this increased malignancy risk is multifactorial, involving chronic iron overload-related oxidative stress, marrow expansion, chronic viral infections (especially hepatitis C), transfusion-related immunomodulation, and potentially prior exposure to cytotoxic agents such as hydroxyurea used for fetal hemoglobin induction.[3-7] The recognition of numerous hemosiderin-laden macrophages and reduced erythroid precursors in this patient underlines how underlying thalassemia and iron overload may modify marrow morphology and mask a concurrent myeloproliferative neoplasm, emphasizing the need for careful integration of clinical history, morphology and targeted molecular testing.[2]

Timely confirmation of BCR-ABL1 fusion permits early initiation of tyrosine kinase inhibitor therapy, that can achieve rapid hematologic remission even in pediatric CML, while concurrent optimization of chelation is also

essential to limit further iron-related hepatic injury.[1-6] Regular monitoring of blood counts, liver function tests, ferritin levels and molecular response is mandatory in such dual-pathology patients, and multidisciplinary coordination between hematology, transfusion medicine and pediatric care is crucial.[4]

CONCLUSION

CML should be considered in the differential diagnosis when a thalassemic child presents with unexplained or disproportionate leukocytosis, basophilia and progressive splenomegaly. Early bone marrow examination with karyotyping and RT-PCR for BCR-ABL1 can distinguish disease evolution related to beta thalassemia from a second primary myeloproliferative neoplasm, allowing prompt therapy and improved outcomes in this complex clinical setting.

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