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Case Report

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Chronic myeloid leukemia in an asymptomatic young patient: a rare case report

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Abstract

Chronic myeloid leukemia (CML) is a clonal myeloproliferative neoplasm arising from the neoplastic transformation of pluripotent stem cells and is consistently associated with the BCR-ABL fusion gene located on the Philadelphia chromosome. It accounts for approximately 15% of all cases of leukemia in adults. Its annual incidence is 1-2 cases per 100,000 individuals. It is uncommon in children, and less than 3% of all patients with CML are younger than 20 years. Here, we report an uncommon case of CML in a young male presenting without symptoms.

The case was a 19-year-old male who came for a physical fitness checkup. He did not have any presenting symptoms or comorbidities. He arrived at SSG Hospital, where his blood sample was taken and sent to the Hematology Laboratory, Pathology Department, Medical College of Baroda, due to a persistently high leucocyte count for 1 month. On examination, the patient was conscious, oriented, and vitally stable. There was no pallor, icterus, pedal edema, or skin rash. There were no complaints of weight loss, anorexia, abdominal pain, or coughing. Cardiovascular and neurological examinations were normal. On abdominal examination, massive grade 4 splenomegaly was found. In view of the high WBC count and massive splenomegaly, he was advised to undergo cytogenetic examination to confirm or rule out the possibility of CML.

***Keywords*:** Asymptomatic, BCR-ABL fusion gene, Chronic myeloid leukemia, Philadelphia chromosome, Young adult

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Introduction

Chronic myeloid leukemia (CML) is a myeloproliferative neoplasm characterized by the presence of the Philadelphia chromosome, resulting from a reciprocal translocation between chromosomes 9 and 22 (1). This translocation leads to the formation of the BCR-ABL1 fusion gene, which encodes a constitutively active tyrosine kinase that drives the pathogenesis of CML (2). While CML typically affects older adults, with a median age at diagnosis of 64 years, it can occur at any age, including in children and young adults (3).

The importance of studying CML in younger populations cannot be overstated. Young patients with CML face unique challenges, including long-term treatment effects, fertility concerns, and psychosocial issues related to living with a chronic disease (4). Moreover, the presentation and progression of CML in younger individuals may differ from

those in older adults, potentially impacting diagnosis, treatment decisions, and outcomes (5).

Despite advances in CML treatment, particularly with the introduction of tyrosine kinase inhibitors (TKIs), early diagnosis remains crucial for optimal management and improved long-term outcomes (6). However, diagnosing CML in young, asymptomatic individuals can be challenging, as routine health check-ups may be the only opportunity to detect abnormalities in blood counts.

This case report describes an unusual presentation of CML in a 19-year-old male who was asymptomatic and diagnosed incidentally during a routine physical fitness checkup. The research method involved a comprehensive clinical examination, hematological and biochemical investigations, peripheral blood smear analysis, and cytogenetic confirmation through fluorescence in situ hybridization (FISH). By presenting this case, we aim to highlight the importance of considering CML in the differential diagnosis of young patients with unexplained leukocytosis, even in the absence of symptoms.

Our findings contribute to the growing body of literature on CML in young adults and underscore the need for heightened awareness among clinicians to ensure timely diagnosis and intervention in this age group. This case also emphasizes the value of routine health check-ups in detecting asymptomatic hematological disorders, potentially leading to improved patient outcomes through early initiation of appropriate treatment.

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In the hematological examination, there was a mild decrease in hemoglobin (Hb), and the total WBC count was significantly higher than normal, as shown in Table 1. Biochemical investigation findings were not significant. In the peripheral smear examination, RBCs were predominantly normocytic and normochromic, with occasional nucleated RBCs. The WBC differential showed a shift to the left with absolute leucocytosis and toxic granules in myeloid series cells (Figure 1). Blast cells were not seen. There were signs of mild basophilia (Figure 2). Myeloid precursors showed chromatin clumping and abnormal lobulation. Platelets were adequate. As advised, fluorescence in situ hybridization (FISH) was performed, which showed t (9,22) BCR/ABL1 fusion, confirming CML.

CML occurs mostly in the median age group of 55-65 years. In our case, a young male with no presenting complaints and a very ambiguous hematology report pointed towards two differentials. Age (19 yrs), total leucocyte count (78,700/cumm), absence of blasts, and normal platelet count favored leukemoid reaction, whereas massive splenomegaly, basophilia, and absence of any infection or inflammation favored CML.

A rare possibility of primary myelofibrosis was also considered in the differential diagnosis, as it can present with a moderate increase in WBC count that falls to subnormal levels secondary to poor production or increased destruction by the spleen.

Cytogenetic examination by FISH showed t(9,22) BCR/ABL1 fusion positivity (100% BCR-ABL fusion) which confirmed the diagnosis of CML. This case highlights that early diagnosis improves the disease-free survival of the patient. Proper monitoring through follow-up and appropriate treatment will improve patient outcomes.

Table 1. Results of various hematology parameters

Parameter	Value
Hemoglobin (Hb)	10.8 gm/dl
Red Blood Cell count (RBC)	3.59 *10^6/cmm
Packed Cell Volume (PCV)	32.8 %
Mean Corpuscular Volume (MCV)	91.4 fl
Mean Corpuscular Hemoglobin (MCH)	30.1 pg
Mean Corpuscular Hemoglobin Concentration (MCHC)	32.9 gm/dl
Red Cell Distribution Width Coefficient of Variation (RDWCV)	19 %
White Blood Cell count (WBC)	78700/cmm
Platelets	156000/cmm

Parameter	Value
Myelocytes	18
Metamyelocytes	3
Band cells	7
Neutrophils	54
Lymphocytes	14
Eosinophils	2
Basophils	2

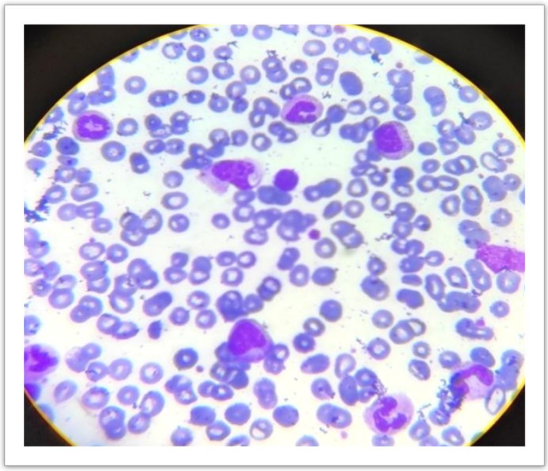

Fig. 1. Giemsa-stained peripheral blood smear showing shift to left with toxic granules in myeloid series cells (100x oil immersion)

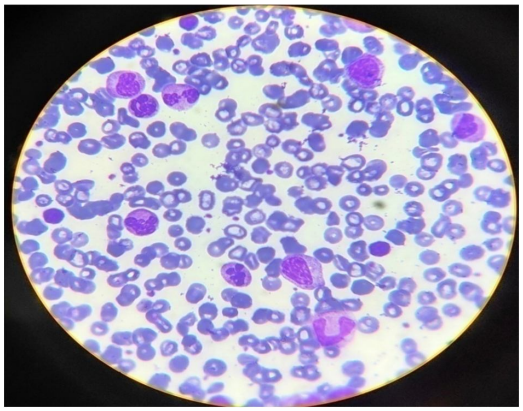

Fig. 2. Basophil on the peripheral blood smear (100x oil immersion)

Discussion

This case report presents several noteworthy findings that contribute to our understanding of CML in young adults and highlights important considerations for clinical practice.

1. Asymptomatic Presentation: The most striking aspect of this case is the patient's asymptomatic presentation. Despite having a significantly elevated WBC count (78,700/cumm) and massive splenomegaly, the patient reported no symptoms

typically associated with CML, such as fatigue, weight loss, or abdominal discomfort. This underscores the importance of routine health check-ups, even in young adults, as they can uncover serious hematological disorders before symptoms manifest (7). It also suggests that CML in younger patients may have a more indolent initial course, potentially delaying diagnosis if relying solely on symptom-based presentations.

2. Hematological Profile: The patient's hematological profile showed several characteristic features of CML:

- Mild anemia (Hb 10.8 g/dL)
- Marked leukocytosis (78,700/cumm)
- Normal platelet count (156,000/cumm)
- Presence of immature granulocytes in peripheral blood (shift to left)
- Basophilia

These findings are consistent with the chronic phase of CML (8). The absence of blast cells in the peripheral blood is particularly noteworthy, as it indicates that the disease was likely caught in its early stages, before progression to the accelerated or blast phase. This early detection is crucial for prognosis and treatment planning.

3. Cytogenetic Confirmation: The FISH analysis revealing 100% positivity for the BCR-ABL1 fusion gene confirms the diagnosis of CML. This high percentage of positive cells suggests that the leukemic clone had already become dominant in the bone marrow, despite the patient's lack of symptoms. This finding emphasizes the disconnect that can exist between molecular/cytogenetic findings and clinical presentation, particularly in younger patients (9).

4. Differential Diagnosis Considerations: The case highlights the importance of considering CML in the differential diagnosis of young patients with unexplained leukocytosis. While leukemoid reaction and primary myelofibrosis were initially considered, the absence of underlying infection or inflammation, along with the presence of basophilia and massive splenomegaly, favored a diagnosis of CML. This underscores the need for a thorough workup, including cytogenetic analysis, in cases of unexplained leukocytosis, even in young, asymptomatic individuals (10).

Conclusions

|This case of asymptomatic CML in a young adult emphasizes the importance of comprehensive health screenings, thorough diagnostic workups, and individualized patient care. It also highlights the need for further research into the unique aspects of CML in younger populations to optimize diagnosis, treatment, and long-term management strategies.

Acknowledgments

None.

Ethical statement

The case report is submitted. The consent of the patient was obtained for nondisclosure of identity.

Data availability

Central Hematology Lab, SSG Hospital, and Medical College Baroda.

Author contributions

Dr. Urja Bhatt- Data collection and writing the article

Dr Hiral Shah- Checked article details and provided the final version for publication after thoroughly examining the article's presentation and figures.

Conflict of interest

None.

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