



Case Report

Adult Type BCR-ABL Positive CML: A Case Report of Southern Punjab

Qazi Muhammad Irfan, Sana Khan, Faiza Sarwar, Ishfaq Ahmed

Sheikh Zayed Medical College/Hospital, Rahim Yar Khan

Abstract

Chronic Myeloid Leukemia is the commonest type of myeloproliferative neoplasm. Peak age group of this illness is 27 to 66 years. The purpose of this case report is to emphasize the occurrence of rare cases and to help clinicians in diagnosing diseases with uncommon presentation. Current case report study was performed in Sheikh Zayed Medical College / Hospital Rahim Yar Khan, Pakistan from february 2022 to March 2022. A 17 years old male patient was included in this study. Complete Blood Count with peripheral smear, bone marrow aspiration and trephine biopsy were performed in Hematology department Sheikh Zayed Hospital Rahim Yar Khan, while fluorecence in situ hybridization analysis for Bcr-Abl fusion gene was performed at Agha Khan University Hospital Laboratory Karachi. Translocation was detected (98%) amongst 200 nuclei scored. Based on this study, possibility of chronic myeloid leukemia in young patients must not be ignored.

Key words: Chronic myeloid leukemia, Fluorecence in situ hybridization, Bcr-Abl.

How to cite this:

Irfan QM, Khan S, Sarwar F, Ahmed I. Adult Type BCR-ABL Positive CML: A Case Report of Southern Punjab. J Pak Soc Intern Med. 2025;6(1): 86-87

Corresponding Author: Dr. Qazi Muhammad Irfan

Email: drirfanqazi@gmail.com

Received: 15-02-2024

Revised: 19-02-2025

Accepted: 24-02-2025

DOI: <https://doi.org/10.70302/jpsim.v6i1.2518>

This is an open access article under the CC BY4.0 license. <https://creativecommons.org/licenses/by/4.0/>

Introduction

The Philadelphia chromosomal translocation is the main feature of Chronic Myeloid Leukemia (CML), results in unchecked production of granulocytes. Myeloproliferative Neoplasm (MPN) are defined by the unchecked myeloid cell growth leading to increased number of various cell lines: erythrocytosis, leukocytosis, or thrombocytosis. Primary myelofibrosis (PMF), chronic myelogenous leukemia (CML), Polycythemia vera (PV), Myelofibrosis and Essential thrombocythemia (ET) are the four main disorders¹. CML accounts for 7–20% of all cases of leukemia and has an incidence of 1-2% per 100,000 each year². Peak age of disease is 27 to 66. Reciprocal translocation of chromosomes 9 & 22 causes the BCR-ABL1 fusion gene, main defect in CML that leads to the development of CML has an unregulated tyrosine kinase activity. CML is primarily a disease of middle age, with rare occurrence in pediatric population³. We present a young patient, age 17 years, who has CML, an uncommon occurrence.

Case Report

Physiological complaints and enlarged spleen (8 fingers) discovered during an examination, a 17-year-old male

patient admitted to Medical Unit III of the Sheikh Zayed Hospital in Rahim Yar Khan, with elevated white blood cell count (WBC) during his initial workup, referred to the Hematology Department. His WBC count was $383 \times 10^3/\mu\text{L}$, and Table 1 includes results from other blood tests. The peripheral blood smear revealed nucleated RBCs, anisocytosis, macrocytes, poikilocytosis and polychromasia. Platelet count was elevated. Mature neutrophils, myelocytes, metamyelocytes and blasts were also seen with Eosinophilia and basophilia.

Table 1: Complete blood count (CBC)

Parameter	Result	Reference range
Leukocytes	383 ,000/ μL	4000–11000/ μL
Thrombocytes	528,000/ μL	150,000–400,000/Ml
HB	9.7 g/dL	13.5–16.5 g/Dl
Lymphocyte	03%	15-25%
Neutrophils	64%	40-75%
Basophil count	03%	0–1 %
Eosinophil	02%	1-3%
Myelocytes	12%	
Metamyelocyte	06%	

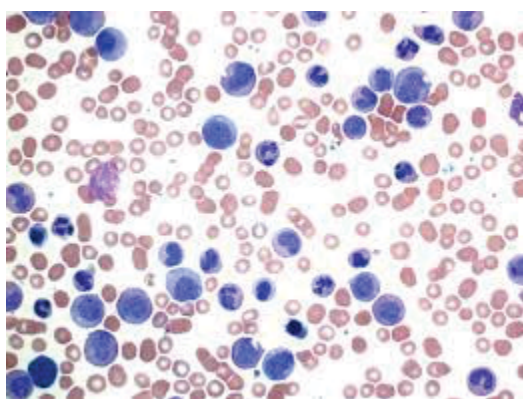


Figure 1: Peripheral Blood Picture showing neutrophilic leukocytosis with immature granulocytes and blasts

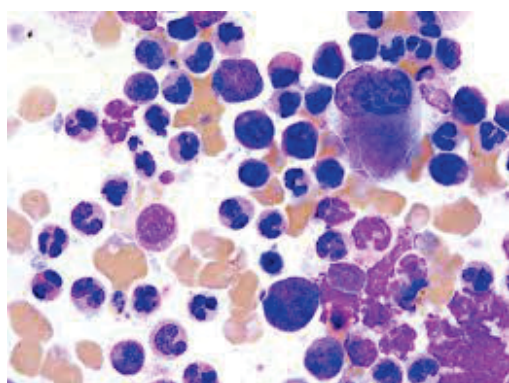
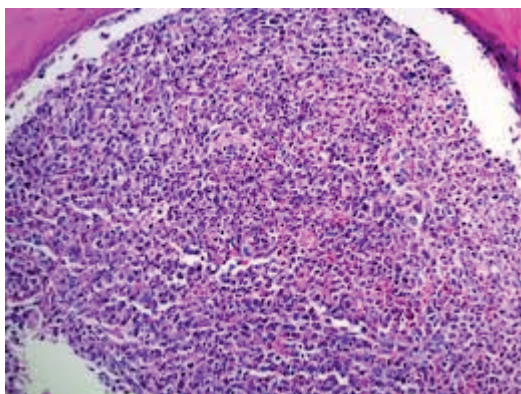


Figure:2 (a) Bone marrow biopsy



(b) Trephine biopsy

Discussion

Myeloproliferative neoplasm CML has a peak age range of 27-66 years. Constitutional signs and symptoms along with visceromegaly, leukocytosis ($>50,000/\text{mm}^3$), thrombocytosis, basophilia, and hypercellular bone marrow with a high proportion of myeloid cell population are present in $>50\%$ of CML patients during the chronic phase⁴. Up to 40% of CML patients in the chronic phase have no symptoms and found out incidentally during routine laboratory tests⁵. For CML diagnosis, (Bcr-abl) fusion gene/genomic translocation $t(9;22)(q34;q11)$ must be present. As BCR-ABL gene copies

were not measurable in cord blood sample and were detected only in 1 of (22) children but in 18 of (52) adults, Biernaux et al. found an increased incidence of positive BCR-ABL fusion gene transcripts with age 6. Consequences of increasing age may be described as mature cells have undergone increased cell divisions and consequently more likely to have accumulation of genetic lesions. An intricate discussion about the age group is sparked by our patient's presentation, which includes a noticeably increased WBC and splenomegaly. Our diagnosis was verified by Bcr-abl fusion gene by FISH. Based on our patient's exceptional age (less than 20 years), CML between the ages of 15-20 have not been reported yet. Clinicians will have more options for diagnosing unusual cases. People with CML usually present after age of 30. We are not familiar commonly with any report of CML <20 years with Bcr-abl positivity. Standard techniques should be used to diagnose, based on the rarity of this condition in adults to prevent misdiagnosis.

Conflict of Interest: None

Funding Source: None

Authors' Contribution

QMI: Conception

SK, FS: Design of the work

IA: Data acquisition, analysis, or interpretation

SK, IA: Draft the work

QMI, FS: Review critically for important intellectual content

QMI,SK, FS, IA: Approve the version to be published

QMI,SK, FS, IA: Agree to be accountable for all aspects of the work

References

- Coltro G, Loscocco GG, Vannucchi AM. Classical Philadelphia-negative myeloproliferative neoplasms (MPNs): A continuum of different disease entities. *International Rev Cell Mol Biol*. 2021;365(1):1-69.
- Shallis RM, Wang R, Davidoff A, Ma X, Podoltsev NA, Zeidan AM. Epidemiology of the classical myeloproliferative neoplasms: the four corners of an expansive and complex map. *Blood Rev*. 2020;42(1):100706.
- Kim JC. Origin of disease heterogeneity in BCR-ABL1 lymphoblastic leukemia [Doctoral dissertation, University of Toronto (Canada)].
- Tavares RS, Nonino A, Pagnano KB, Nascimento AC, Conchon M, Fogliatto LM, Funke VA, Bendit I, Clementino NC, Chauffaille MD, Bernardo WM. Guideline on myeloproliferative neoplasms: Associação Brasileira de Hematologia, Hemoterapia e Terapia Celular Project guidelines: Associação Médica Brasileira-2019. *Hematol Trans Cell Therap*. 2019;41(Suppl 1):1-73.
- Jabbour E, Kantarjian H. Chronic myeloid leukemia: 2014 update on diagnosis, monitoring, and management. *Am J Hematol*. 2014;89(5):547-56.
- Risques RA, Kennedy SR. Aging and the rise of somatic cancer-associated mutations in normal tissues. *PLoS Genet*. 2018;14(1):e1007108.