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Case Report

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

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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 fluorescence 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, Flourescence in situ hybridization, Bcr-Abl.

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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 disorders[1]. CML accounts for 7–20% of all cases of leukemia and has an incidence of 1-2% per 100,000 each year[2]. 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 population[3]. 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 383×103/µl, 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%	

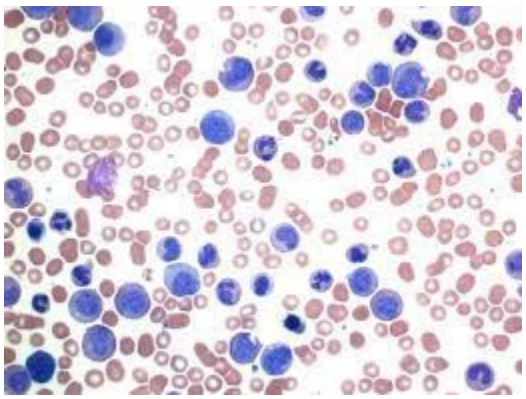


Figure 1: *Peripheral Blood Picture showing neutrophillic leukocytosis with immature granulocytes and blasts*

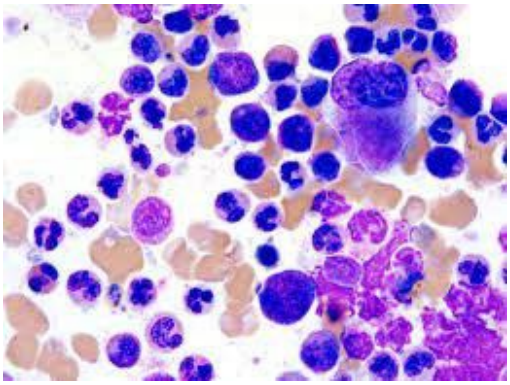
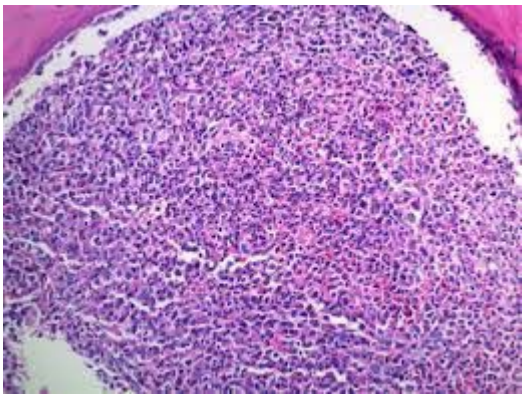


Figure:2 *(a) Bone marrow biopsy*



(b) Trephine biopsy

Discussion

Myeloproliferative neoplasm CML has a peak age range of 27-66 years. Constitutional signs and symptoms alongwith visceromegaly, leukocytosis (>50.000/ 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 phase[4]. Up to 40% of CML patients in the chronic phase have no symptoms and found out incidentally during routine laboratory tests[5]. For CML diagnosis, (Bcr-abl) fusion gene/genomic translocation t (9; 22) (q34; q11) must be present. As BCR-ABL gene copies were not measureable 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.

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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

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