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ACUTE MYELOID LEUKEMIA WITH BASOPHILIA WITH t(6;9) (p23;q34) – A RARE SUBTYPE OF AML.

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INTRODUCTION

Acute Myeloid Leukemia (AML) with the translocation t(6;9) (p23;q34), involving breaks at band 23 of the short arm of chromosome 6 and band 34 of the long arm of chromosome 9, is a rare subtype, representing 0.5% to 4% of AML cases.1 This translocation leads to the creation of a chimeric fusion gene (DEK-NUP214) on the derivative chromosome 6, known as der(6). The translocation was first discovered in AML by Rowley and Potter in 19762, marking a key development in understanding AML genetics. Most patients with AML and the t(6;9) (p23;q34) translocation are classified under the French-American-British (FAB) system as having either AML-M2 or AML-M4. WHO classification of Acute Leukemias 2017 categorized this under AML with recurrent genetic abnormalities. Many of these patients exhibit signs of underlying or preceding myelodysplasia, contributing to the poor prognosis. Complete remission is achieved in only about 50% of cases with conventional chemotherapy with most patients surviving just one year after diagnosis. Bone marrow transplantation may be the only treatment option capable of achieving a potential cure in these cases.

CASE REPORT

A 50-year-old male presented with a gradual onset of fever and a cough with expectoration lasting for one month, followed by episodes of hemoptysis and associated generalized weakness. He also reported history of blood transfusions and recurrent infections in the past. The persistent symptoms, along with the patient's medical history, raised concerns for an underlying chronic infection or hematological disorders.

Laboratory investigations - Hemoglobin levels : 8.7 g/dl

Peripheral blood smear (PBF) - Dimorphic red blood cell picture.

TLC- 58,900/cmm

DLC- 75% Blasts, 22% Basophils, 2% Lymphocytes, and 1% Polymorphs.

Platelet count -30,000/cmm.

Given these findings, particularly the high percentage of blasts and low platelets, the patient was advised to undergo a bone marrow biopsy for further evaluation and definitive diagnosis.

Another lab findings include- Increased serum alkaline phosphatase (S. Alk) levels of 152 IU/L, while SGOT, SGPT and RFT levels were within normal limits. There was no evidence of lymphadenopathy, splenomegaly, or hepatomegaly on examination.

BONE MARROW FINDINGS

Cellularity-Moderately cellular, Reaction- Mild megaloblastic

NE:E ratio- 98:2

Erythroid series: Markedly diminished with mild megaloblastic maturation and features of dysery-thropoiesis seen.

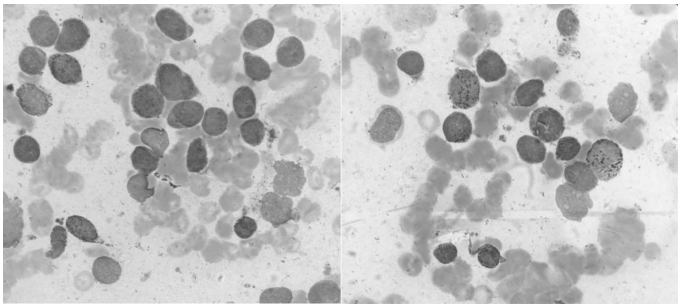
Myeloid series: Cellularity of marrow is mainly due to medium sized Blasts(70%) showing basophilic cytoplasm with 1-4 conspicuous nucleoli. Scattered cells are seen covered with basophilic granules. Basophilic precursors (17%) are increased in number. Rest of the myeloid series is markedly diminished.

Rare Megakaryocyte seen.

Impression:- Acute Myeloid Leukemia (AML) with Basophilia.

Advised: 1. FLOW CYTOMETRY

A cytogenetic study performed at a higher institute confirmed that the patient tested positive for the t(6;9) translocation.



DISCUSSION

The bone marrow in cases of t(6;9) acute myeloid leukemia (AML) is typically hypercellular for the patient's age and shows increased number of blasts (≥20%), which may contain Auer rods or granules. Additionally, bone marrow basophilia and dysplasia, either unilineage or multilineage, can be observed in the residual hematopoietic cells. According to Alsabeh et al.3, there is also an increased incidence of ringed sideroblasts, suggesting features of myelodysplasia in these patients. The largest retrospective study4 to date, involving five cooperative groups (Southwest Oncology Group, Cancer and Leukemia Group B, Eastern Cooperative Oncology Group, Children's Oncology Group, and Children's Cancer Group), reported a prevalence of 44% for marrow basophilia and 67% for evidence of myelodysplasia in t(6;9) AML cases. Immunophenotypically, the blast cells in t(6;9) AML are positive for CD9, CD13, CD33, and HLA-DR; they are usually positive for CD45 and CD38 and may also express CD15, CD34, and terminal deoxynucleotidyl transferase, contributing to its diagnostic profile.5 Alsabeh et al. noted that blasts, initially presenting as CD34-ve, often relapsed as CD34+ve. The t(6;9) translocation is most commonly associated with acute myeloid leukemia (AML) of the FAB-M2 subtype and is sometimes considered a distinct disease entity due to its unique clinical and morphologic features include marrow basophilia and evidence of myelodysplasia, as well as its poor prognosis.

CONCLUSION

Patients diagnosed with t(6;9) acute myeloid leukemia (AML) generally have a very poor prognosis, as current chemotherapy options have not shown significant improvement in overall survival rates. However, early and accurate diagnosis is essential, as these patients may benefit from early allogeneic stem cell transplantation, which can improve outcomes. There is a growing need to explore novel therapies, such as anti-CD33–based treatments and FLT3 inhibitors, to potentially enhance treatment efficacy for t(6;9) AML patients. Furthermore, molecular monitoring of minimal residual disease (MRD) is a valuable tool for assessing risk stratification and guiding disease management. The World Health Organization's classification of hematopoietic tumors highlights the importance of understanding the prognostic implications of cytogenetic abnormalities in hematologic mali-gnancies. Given its distinct morphologic, cytogenetic, and clinical characteristics, AML with t(6;9) (p23;q34) should be recognized as a separate entity within the classification of AML with recurrent cytogenetic abnormalities. This recognition emphasizes the need for tailored treatment approaches and continued research to address the unique challenges associated with this rare but aggressive subtype of AML.

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