SOHO ANNUAL MEETING 2024

Molecular analysis always wins. A case report of Philadelphia positive Myelofibrosis

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INTRODUCTION

Myelofibrosis is a myeloproliferative neoplasm characterized by replacement of bone marrow with fibrous tissue which is typically associated with mutations in genes as JAK2, CALR, and MPL. The presence of the Philadelphia chromosome (Ph) is generally indicative of chronic myeloid leukemia rather than myelofibrosis. The occurrence of Philadelphia chromosome-positive myelofibrosis (Ph+ MF) is rare, representing a diagnostic and therapeutic challenge due to its unusual clinical presentation and overlapping features with CML.

AIM

This abstract presents a detailed case study of a patient diagnosed with Ph+ MF, exploring the clinical manifestations, diagnostic criteria, and therapeutic approach. By discussing this rare entity, we aim to enhance the understanding of its pathophysiology, highlight the importance of molecular and cytogenetic analysis in accurate diagnosis, and review potential treatment strategies.

METHODS

A case report which includes diagnostic history taking, physical examination, full blood count, Molecular markers , Bone marrow aspiration and biopsy .

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RESULTS

Our 45 year old Egyptian Lady presented with leucocytosis, thrombocytosis(and mild splenomegaly .

No characteristics of CML were found in blood film. Bone marrow biopsy showed a hypercellular marrow (90% cellularity) with increased myeloid series (70% of cellularity) along with excess immature megakaryocytes with grade III reticulin fibrosis. JAK II and CALR mutation were negative as well as mild elevated LDH.

Initially our patient started Hydroxyurea with anagrelide with no improvement until Ruxolitinib 15 mg BD was started with progression of platelet count to exceed 1.5 million/ul . This did not improve on adding hydroxyurea.

PCR for BCR –ABL 210 was done to come as positive 38% and 29% on repeat. The patient then was shifted on Imatinib 400mg which led to normalization of her platelet count. PCR for BCR-ABL 210 was repeated after 3months to be back as 0.1%. Our lady continues the same dose with no added side effects.

CONCLUSIONS

Molecular and cytogenetic studies are corner stones in haematological diagnosis especially MPN. Despite Philadelphia positive cases of MF are very rare but needs to be considered with refractory or progressive cases after Ruxolitinib therapy.

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