Introduction

Chronic myelomonocytic leukemia (CMML) is a clonal stem cell disorder defined as a myeloproliferative/myelodysplastic disease (MPN/MDS) by the World Health Organization based on five diagnostic criteria which include: persistent ≥3 months peripheral monocytosis (monocytes ≥1,000/µL, accounting for ≥10% of the WBC count), no Philadelphia chromosome or BCR-ABL1 fusion gene, no PDGFRα, PDGFRβ, or FGFR1 rearrangement, fewer than 20% blasts in the blood and bone marrow, and dysplasia in one or more myeloid lineages. If dysplasia is not seen, then cytogenetic abnormalities or exclusion of other causes of chronic monocytosis such as malignancy, infection, or inflammation, qualifies.

Patient History

We describe the case of a 68 year-old female with a past medical history of untreated HCV cirrhosis and chronic kidney disease who presented with persistent (>6 months) thrombocytopenia, monocytosis, and neutrophilia. There was no lymphadenopathy or splenomegaly noted. Bone marrow biopsy and aspiration were obtained to rule out malignancy.

Materials and Methods

Peripheral blood and bone marrow aspirate smears were reviewed in conjunction with bone marrow biopsy and clot sections. Iron stores were evaluated in the aspirate smear using Prussian Blue. Additional material was submitted for flow cytometry, karyotype analysis, and fluorescence in situ hybridization (FISH).

Bone Marrow and Peripheral Blood

Peripheral Blood and Bone Marrow: The peripheral blood showed macrocytic anemia, left-shifted leukocytosis with absolute neutrophilia, eosinophilia, and monocytosis (A). The bone marrow was markedly hypercellular (90-100%) (B). The aspirate contained increased granulocytes, monocytes, and megakaryocytic dysplasia with 2% blasts (C). The CD61 immunohistochemical stain highlights dysplastic megakaryocytes (D).

Flow Cytometry: Monocytes showed decreased expression of CD14 and aberrant increased expression of CD56. There was no monotypic B-cell, abnormal T-cell populations, or increase in CD34-positive blasts.

Genetic Testing: Normal karyotype (46, XX). The qualitative PCR assay was negative for V617F JAK2 mutation. No assay specific abnormalities detected by AML, MDS, BCR/ABL and MPN with eosinophilia probes. There was no deletion or insertion detected with the analyzed regions of the calreticulin (CALR) gene.

Discussion

CMML is a rare entity with a highly variable clinical presentation which can lead to a delayed diagnosis. This case illustrates a helpful and practical learning point to consider CMML in the management of elderly patients with persisting unexplained peripheral monocytosis.

Conclusion

References