

p210 BCR-ABL1 CML with CMML Clones: A Rare Presentation

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Abstract : Introduction: p190 BCR-ABL1 in CML is often associated with monocytosis. In the case described here, monocytosis is associated with coexisting p210 BCR-ABL and CMML clones. Mutation analysis using next-generation sequence (NGS) in our case showed TET2 and SRSF2 mutations. Aims & Objectives: A 75-year male was evaluated for monocytosis and thrombocytopenia. CBC showed Hb-11.8g/dl, TLC-12,060/cmm, Monocytes-35%, Platelets-39,000/cmm. Materials & Methods: Bone marrow examination showed a hypercellular marrow with myeloid series showing sequential maturation up to neutrophils with 30% monocytes. Immunophenotyping by flow cytometry from bone marrow had 3% blasts. Making chronic myelomonocytic leukemia as the likely diagnosis. NGS for myeloid mutation panel had TET2 (48.9%) and SRSF2 (32.5%) mutations. This report further supported the diagnosis of CMML. To fulfil the WHO diagnostic criteria for CMML, a BCR ABL1 by RQ-PCR was sent. The report came positive for p210 (B3A2, B2A2) Major Transcript (M-BCR) % IS of 38.418. Result: The patient was counselled regarding the unique presentation of the presence of 2 clones- P210 CML and CMML. After discussion with an international faculty with vast experience in CMML. It was decided to start this elderly gentleman on Imatinib 200mg and not on azacytidine, as ASXL1 was not present; hence, his chances of progressing to AML would be less and on the other end, if CML is left untreated then chances of progression to blast phase would always be a possibility. After 3 months on Imatinib his platelet count improved to 80,000 to 90,000/cmm, but his monocytosis persists. His 3rd month BCR-ABL1 IS% is 0.004%. Conclusion: After searching the literature, there were no case reports of a coexisting CML p210 with CMML. This case might be the first case report. p190 BCR ABL1 is often associated with monocytosis. There are few case reports of p210 BCR ABL1 positivity in patients with monocytosis but none with coexisting CMML. This case highlights the need for extensively evaluating patients with monocytosis with next-generation sequencing for myeloid mutation panel and BCR-ABL1 by RT-PCR to correctly diagnose and treat them.

Keywords : CMML, NGS, p190 CML, Imatinib

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