Adult Hematology Abstract Categories

Chronic Myeloproliferative Diseases

PP 07

BCR-ABL1 MINOR (P190, E1A2) POSITIVE CHRONIC MYELOID LEUKEMIA: A RARE CASE REPORT

Songül Beskisiz Dönen [1,*], Miray Nilgün Yazok [2], Vehbi Demircan [1], Abdullah Karakuş [1], Mehmet Orhan Ayyıldız [1]

[1] *Dicle University, Faculty of Medicine, Department of Hematology, Türkiye*
[2] *Dicle University, Faculty of Medicine, Department of Internal Medicine, Türkiye*

INTRODUCTION: Chronic myeloid leukemia (CML) is a myeloproliferative neoplasm characterized by the BCR-ABL1 fusion gene and accounts for approximately 15–20% of all leukemias. While the majority of cases harbor the p210 (major) transcript, the p190 (minor, e1a2) transcript is exceedingly rare, representing only about 1–2% of CML cases. This subtype may exhibit distinct hematologic features compared to p210-positive cases, particularly peripheral monocytosis and marked splenomegaly. In the literature, responses to tyrosine kinase inhibitor (TKI) therapy in p190-positive CML have been reported to be variable, and long-term outcomes are described only in limited case reports. Therefore, presenting the clinical and laboratory features of this uncommon subtype is of particular importance. **CASE PRESENTATION:** A 23-year-old female patient presented with complaints of fatigue and dyspeptic symptoms. Complete blood count revealed WBC: $70 \times 10^3/\mu L$, neutrophils: $58.5 \times 10^3/\mu L$, monocytes: $7.38 \times 10^3/\mu L$, hemoglobin: 10.4 g/dL, and platelets: $871 \times 10^3/\mu L$. Abdominal ultrasonography demonstrated splenomegaly with a longitudinal diameter of 175 mm. Peripheral blood smear and bone marrow aspiration-biopsy findings were consistent with chronic myeloid leukemia, with blasts reported as <5%, and the overall evaluation was described as a "myeloproliferative neoplasm." Molecular testing showed negative results for the major BCR-ABL1 transcript, whereas the minor BCR-ABL1 (e1a2) transcript was detected at 3.4%. The patient was started on first-line therapy with imatinib. At the third month of treatment, BCR-ABL1 (minor) was 10.51%, although hematologic parameters had improved. With continuation of imatinib, the sixth-month evaluation showed a decrease in BCR-ABL1 (minor) to 1.56%, with a normalized blood count (WBC: $5.31 \times 10^3/\mu L$, Hb: 10.6 g/dL, platelets: $226 \times 10^3/\mu L$). The patient's clinical symptoms had resolved, and she remains on imatinib therapy with ongoing follow-up. **DISCUSSION AND CONCLUSION:** While the p210 (major) transcript is the most frequently detected form in chronic myeloid leukemia (CML), the p190 (minor, e1a2) transcript is exceedingly rare, occurring in only about 1–2% of cases. In the literature, this subtype has been associated with peripheral monocytosis and marked splenomegaly, and responses to tyrosine kinase inhibitors (TKIs) have been reported as variable. In some patients, imatinib therapy may not achieve sufficient molecular response, whereas deeper responses have been described with second-generation TKIs. In our patient, early hematologic response was achieved with imatinib, and by the sixth month a marked molecular reduction was observed. Through this case, we aim to highlight the clinical and laboratory characteristics of p190-positive CML and to emphasize the importance of close molecular monitoring and careful evaluation of treatment response in this rare subtype.

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Adult Hematology Abstract Categories

Coagulation Disorders

PP 08

AN UNUSUAL DIAGNOSIS IN A TODDLER PRESENTING WITH MASSIVE GASTROINTESTINAL BLEEDING: A CASE OF ANGIODYSPLASIA AND TYPE 3 VON WILLEBRAND DISEASE

Arzu Akyay [1,*], Nezihe Köker [2], Bengü Macit [3], Yurday Öncül [3], Emre GÖk [4], İlknur Varol [4], Şükrü Güngör [4]

[1] *Inonu University Department Of Pediatric Hematology, Türkiye*
[2] *Inonu University Department Of Pediatrics, Türkiye*
[3] *Inonu University Department Of Pediatric Hematology and Oncology, Türkiye*
[4] *Inonu University Department Of Pediatric Gastroenterology, Türkiye*

Case report: Von Willebrand disease (VWD), caused by a deficiency or dysfunction of the von Willebrand protein (VWF), presents with a wide range of clinical manifestations. VWF is known to play a role in both platelet adhesion and angiogenesis. Consequently, defective angiogenesis can lead to angiodysplasia, particularly in the gastrointestinal system, occurring in 2-5% of VWD cases, typically in adults. Herein, we present what is, to our knowledge, the youngest reported case of a patient diagnosed with VWD following a presentation of gastrointestinal bleeding secondary to angiodysplasia. A 2-year and 10-month-old female patient was admitted to our hospital for melena and hematemesis. Her medical history was unremarkable, with no reported fever, diarrhea, or use of anti-inflammatory medications. There was no consanguinity between the parents, and no known family history of bleeding diathesis, Türkiye. Upon physical examination, the patient was lethargic, weak, and pale. A cardiac murmur was noted. Several 0.5 cm ecchymoses were present on her legs, though petechiae were absent. Initial laboratory tests revealed severe anemia (hemoglobin 3.5 g/dL). Her platelet count was within the normal range, as was her INR (0.96; normal range: 0.8-1.2). However, a prolonged aPTT (46.4 s; normal range: 20-34 s) and a bleeding time greater than 5 minutes were noted. An erythrocyte transfusion was immediately administered. Treatment with somatostatin and tranexamic acid was initiated. Despite this, the patient experienced three