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.

https://doi.org/10.1016/j.htct.2025.106141

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

more episodes of bright red bleeding and required two additional erythrocyte transfusions. An upper endoscopy was performed, revealing no esophageal varices. A 2 × 3 cm angiodysplastic lesion was observed in the gastric corpus and was cauterized with an argon laser. A scintigraphy scan confirmed increased activity in the same area. Following the procedure, a fresh frozen plasma transfusion was administered and propranolol treatment was started. With the bleeding controlled, and given the concurrent angiodysplasia, a detailed work-up for coagulopathy was performed. The patient's von Willebrand factor activity was below 5%, the von Willebrand factor antigen was below 3%, and Factor VIII was less than 3.5%. Platelet aggregation tests were normal. Genetic analysis of the VWF gene identified a novel homozygous mutation, c.7176T>G p.(Tyr2392Ter), and a heterozygous mutation, c.817C>G p.(Ar273Gly). Considering the clinical presentation, laboratory findings, and genetic analysis in a patient with no parental bleeding history, a diagnosis of Type 3 von Willebrand disease was established. Gastrointestinal bleeding due to angiodysplasia in VWD is a well-known complication that typically arises in adults. The only previously reported pediatric case was by Aggoune et al., who described a 14-year-old with Type 3 VWD and duodenal angiodysplasia who required surgical resection for recurrent bleeding. In contrast, our patient's initial bleeding episode was successfully managed with a combination of argon laser cauterization and medical therapy. This case highlights the importance of considering VWD in pediatric patients who present with severe gastrointestinal bleeding, especially when routine coagulation tests show a prolonged aPTT and bleeding time in the absence of risk factors for esophageal varices. It also serves as a crucial reminder that angiodysplasia is a known complication of VWD that can present even in the youngest of patients

https://doi.org/10.1016/j.htct.2025.106142

PP 09

Marrow-Dominant Marginal Zone Lymphoma with Plasmacytic Differentiation in a Frail and old patient: Immunophenotypic Pitfalls and Rituximab-Only Strategy

Naciye Nur Tozluklu *, Birol Güvenç

Çukurova University, Dept.of Hematology, Balcali_Adana,Turkiye

Introduction: Marginal zone lymphoma (MZL) with plasmacytic differentiation can mimic other B-cell entities—particularly CLL/SLL and lymphoplasmacytic lymphoma (LPL)—and often presents in the elderly with cytopenias rather than bulky nodal disease. Correct classification is critical, as comorbidity and frailty frequently constrain treatment intensity. We report a bone marrow–dominant, plasmacytoid MZL in an 84-year-old woman successfully managed with rituximab monotherapy. **Methods:** We conducted a single-patient, retrospective case review of prospectively collected clinical, laboratory, pathology, and imaging data. Diagnostic workflow integrated complete blood count, immunoglobulin quantification, bone marrow histology with immunohistochemistry (IHC), multiparameter flow cytometry, and FDG-PET/CT. Treatment selection followed a frailty-adapted decision process. **Results:** An 84-year-old woman presented with progressive fatigue and dyspnea on exertion. Baseline labs showed leukocytosis with marked lymphocytosis and macrocytic pancytopenia (WBC 22.2 × 10^9/L; absolute lymphocytes 18.4 × 10^9/L; Hb 8.1 g/dL; MCV 105 fL; platelets 42 × 10^9/L). Immunoglobulins were not suggestive of LPL/WM (IgM 0.73 g/L; IgG 13.6 g/L; IgA 1.7 g/L). FDG-PET/CT revealed mediastinal/abdominal lymphadenopathy, splenomegaly, and a sternal cortical irregularity suspicious for osseous involvement. Bone marrow biopsy was hypercellular (∼95%) with ∼90% interstitial/patchy small-to-intermediate B-cell infiltration; reticulin fibrosis 0/4; Congo red negative. IHC supported a non-CLL, non-mantle phenotype: CD20 strong positive; CD5, CD23, CD10, cyclin D1, annexin A1, TRAP all negative. Flow cytometry demonstrated a clonal mature B-cell population (CD19+, CD20+, CD38+, cCD79a+) without CLL-type markers (CD5/CD23 negative). Plasmacytic differentiation was present, yet serum IgM remained normal, arguing against LPL/WM. Overall, findings established marginal zone lymphoma with plasmacytic differentiation, stage IV-A (marrow ± splenic/possible bone involvement). Given advanced age, cytopenias, and frailty, cytotoxic chemo-immunotherapy was deferred. The patient received rituximab monotherapy with antiviral prophylaxis and supportive care (transfusion as needed). Treatment was well tolerated; early follow-up showed clinical improvement with rising hemoglobin and platelet counts and reduction in lymphocytosis. **Discussion:** This case highlights three practice points. First, plasmacytic differentiation in MZL can masquerade as CLL or LPL/WM; a disciplined panel—CD5/CD23/cyclin D1 negativity with strong CD20 and compatible flow cytometry—prevents misclassification. Second, serological context matters: normal IgM helped exclude LPL/WM despite plasmacytoid histology. Third, in the very elderly/frail, rituximab monotherapy is a rational, lower-toxicity strategy that can reverse cytopenias and improve function when marrow disease predominates. The possible osseous signal on imaging further underlines the heterogeneity of MZL dissemination. Educationally, the case underscores integrating morphology, IHC, flow, and serology to secure diagnosis and tailor therapy beyond one-size-fits-all chemo-immunotherapy. **Conclusion:** Bone marrow-dominant MZL with plasmacytic differentiation presents diagnostic challenges but can be accurately classified through integrated morphology, IHC, flow cytometry, and serology. In very elderly or frail patients, rituximab monotherapy represents a rational and effective treatment strategy, offering hematologic recovery and functional benefit while minimizing toxicity.

https://doi.org/10.1016/j.htct.2025.106143