PEGINTERFERON ALFA-2A FOR THE TREATMENT OF PAEDIATRIC ESSENTIAL THROMBOCYTHEMIA - A CASE REPORT

Teov B[1], Jovanovska A[1], Kocheva S[1], Trajkova-Antevska Z[1]
1) University Children's Hospital Skopje, North Macedonia

Corresponding author
Teov Bojan
University Children's Hospital Skopje, North Macedonia
Department for hematology and oncology
teov.bojan@gmail.com
0038975213009

Essential thrombocythemia (ET) is a myeloproliferative disease characterized by high platelet count that cannot be attributed to other causes and megakaryocytic lineage hyperplasia in the bone marrow. Although less common in children, it generally presents with a more benign course compared to adults.

This study describes a 17-year-old male with asymptomatic thrombocytosis observed at the age of 9. The patient's platelet count gradually escalated to nearly $3.000x10^9$/L. Past medical history and clinical examination were unremarkable, ruling out secondary thrombocytosis. Genetic analysis failed to identify mutations in JAK2, MPL and CALR genes, nor the BCR-ABL fusion transcript. A bone marrow trephine biopsy was consistent with ET. Notably, acquired vWD was detected. Treatment with hydroxyurea resulted in only a partial response, prompting the initiation of second-line therapy with subcutaneous PEGinterferon alfa-2A. The patient has been followed on this regimen for an 80-month period during which the platelet count has remained stable at $400\text{-}600x10^9$/L. Signs of hemorrhagic or thrombotic complications have not been observed.

This case emphasizes the diagnostic and treatment challenges experienced in paediatric ET. JAK2-V617F, MPL and CALR mutations, commonly associated with myeloproliferative neoplasms in adults, are not clearly correlated with paediatric ET. Many paediatric patients with clinical and histological features of ET carry a molecular triple wild-type status.

In contrast to well-defined recommendations in adults, there is no standard approach for risk stratification and management in pediatric ET. Long-term follow-up is crucial to optimize therapeutic strategies and gain a comprehensive understanding of paediatric ET.