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XIX Convegno Triennale sugli Aspetti Clinici e Sociali dell'Emofilia ABSTRACT N. CO-82

3.04 Acquired coagulation disorders

ACQUIRED SEVERE FACTOR XIII DEFICIENCY AS THE INITIAL PRESENTATION OF PAEDIATRIC ACUTE PROMYELOCYTIC LEUKAEMIA

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Background. Acute Promyelocytic Leukemia (APL), representing 10% of AML cases, is characterized by the t(15;17) translocation. Despite cure rates exceeding 90% with ATRA and ATO, early hemorrhagic death remains a critical challenge, occurring in 10-30% of patients. APL coagulopathy is multifactorial, involving DIC and primary hyperfibrinolysis. Malignant promyelocytes trigger these pathways via tissue factor and annexin A2 expression. Furthermore, ATRA-induced ETosis—the release of extracellular chromatin traps (NETs)—exacerbates the condition by discharging potent proteases like elastase. Crucially, these enzymes degrade Factor XIII (FXIII), a transglutaminase vital for fibrin cross-linking. This acquired FXIII deficiency destabilizes hemostasis, significantly contributing to the severe bleeding diathesis. We present an APL case where severe FXIII deficiency and related bleeding preceded the onset of characteristic APL coagulopathy.

Case Report. A 12-year-old male with no prior history of bleeding disorders presented to the emergency department with a soft, 15 mm scalp hematoma in the right parietal region, triggered by minor hair pulling. He also reported recent fatigue and low-grade fever. Coagulation tests showed a PT ratio of 1.4, a PTT ratio of 1.15, and fibrinogen levels of 151 mg/dL. Given that the severity of the hemorrhage appeared disproportionate to these mild laboratory abnormalities, Factor XIII (FXIII) levels were assessed, revealing a severe deficiency (7%). No other bleeding manifestations were present. FXIII levels improved following administration of fresh frozen plasma and subsequently normalized with purified FXIII concentrate. Over the following days, the patient developed progressive pancytopenia; peripheral blood and bone marrow investigations led to the diagnosis of Acute Promyelocytic Leukemia (APL).

Conclusions. The coagulopathy associated with Acute Promyelocytic Leukemia (APL) is traditionally attributed to disseminated intravascular coagulation (DIC) and primary hyperfibrinolysis. However, acquired Factor XIII (FXIII) deficiency represents a critical, yet often under-recognized, component of this hemostatic failure. Diagnosing acquired FXIII deficiency is challenging because routine screening tests, such as PT and aPTT, remain normal in cases of isolated deficiency. Therefore, quantitative FXIII activity assays should be prioritised in APL patients presenting with severe or unexplained bleeding. Despite the severity of these mechanisms, current international guidelines—including those from ASH, ELN, EHA, and NCCN—are highly rigorous regarding APL management but do not yet include specific recommendations for FXIII monitoring. Our case highlights a unique clinical scenario where severe acquired FXIII deficiency and its subsequent hemorrhagic manifestations preceded the formal diagnosis of APL and the onset of its characteristic coagulopathy by several days. The early detection of this deficiency allowed for the prompt initiation of replacement therapy with FXIII concentrate, effectively preventing further life-threatening bleeding complications. This observation suggests that proteolytic degradation of FXIII can act as a primary and independent driver of hemorrhage in the earliest stages of the disease. In conclusion, integrating FXIII assessment into the initial diagnostic workup of suspected APL may be crucial for early stabilization and improved clinical outcomes.

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