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ABSTRACT N. PO75

PROPHYLAXIS WITH RFXIII: A CLINICAL CASE IN A PEDIATRIC PATIENT WITH CONGENITAL FXIII DEFICIENCY AND DELAYED DIAGNOSIS.

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Background: Congenital factor XIII (FXIII) deficiency is a rare coagulation disorder characterized by muscular or mucocutaneous bleeding with life-threatening intracranial hemorrhages (ICHs), especially in cases with severe disease. The best treatment is the use of prophylactic plasma-derived or recombinant FXIII (rFXIII). All routine coagulation tests are normal in FXIII deficiency (FXIIID), which complicates the diagnosis of this disorder.

Case reports: A 6 years old girl affected by severe deficiency of FXIII. The patient was born at term at 36 weeks, two days after delivery she was admitted to the NICU for respiratory failure, anemia and acute thrombocytopenia, acute renal failure and hepatic hematoma. She was treated with dialysis for 2 days, assisted ventilation, transfusion with concentrated red blood cells, platelets and plasma and discharged after 40 days. In August 2021 she had a new access to the ER of Moscati Hospital in Avellino for left gluteal hematoma with difficulty in walking, treated with paracetamol and antibiotics. In August 2022 she was admitted to our hospital for the detection of an iliopsoas muscle hematoma, with severe difficulty in deambulation. Coagulation tests were normal, therefore, factor XIII assay was performed, resulting 1%. The patient was treated with rFXIII at a dose of 35 IU/kg, with resolution of the clinical symptoms in about 2 weeks. The patient has been on prophylaxis with rFXIII every 4 weeks at a dose of 35 IU/kg since August 2022 and has only experienced minor bleeding episodes (superficial post-traumatic hematomas and ecchymoses). The activity of FXIII is on average about 120% after 1 hour of drug administration. The trough level at 4 weeks is on average about 6%.

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Conclusions: Prophylaxis with rFXIII appears to be safe and effective in our patient with congenital FXIII deficiency: from 2022 to today, no significant haemorrhagic episodes have been recorded, only superficial manifestations of mild severity.

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