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ABSTRACT N. PO45

ROUTINE PROPHYLAXIS WITH PD-FX CONCENTRATE IN SEVERE INHERITED FX DEFICIENCY: A CASE REPORT.

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Background: Severe or moderate Inherited factor X (FX) deficiency is a very rare autosomal recessive bleeding disorder affecting 1:1,000,000 individuals. FX protein is synthesized by the liver and is encoded by a gene (F10) of 27 kb located on chromosome 13, containing 8 exons. One hundred eighty mutations, most of which missense, have been identified to date. FX plasma levels above 20% are infrequently associated with bleeding and heterozygotes are usually asymptomatic. In moderate (FX 1-5%) or severe (FX <1%) deficiency bleeding manifestations can be serious including hemarthroses, hematomas, umbilical cord, gastrointestinal, and central nervous system bleeding. Severe FX deficiency may benefit from routine prophylaxis. Treatment options include the use of fresh frozen plasma (FFP) or prothrombin complex concentrate (PCC). Recently a specific plasma-derived (pd) FX concentrate (Coagadex, Kedrion Biopharma) has become available for on-demand, routine prophylaxis and perioperative treatment in people of all ages with inherited FX deficiency. We present the case of an 18-year-old boy with severe FX deficiency switched to routine prophylaxis with pd-FX.

Case report: In negative family history for bleeding, the propositus was born with cesarean delivery due to fetal distress. In the first 12 hours of life multiple petechiae in chest and lower limbs appeared, followed by pallor, tachypnea, irritability, and evidence of large abdominal subcutaneous hematoma, suspected retroperitoneal bleeding and hematuria. He was admitted to neonatal intensive care and treated with packed red cells and FFP. Laboratory tests showed Hb 8.5 g/dL, metabolic acidosis (pH 7.13, HCO3- 15 mmol/L), PTT 109 sec (NR 22 – 38 sec), PT INR 11.7, FX < 1 U/dL with F10 mutation p.Gly173Trp/c.139delGfsp.Glu47X (exon 2). After diagnosis of severe FX deficiency and central venous catheter (CVC) insertion, the patient started treatment with PCC 50 U/kg every 8-12 hours, then 30-35 U/kg every 72-96 hours up to 2 years, then every 96-120 hours. For reluctance to switch to prophylaxis 2 times a week, once the CVC was removed, he continued with weekly infusions of PCC 40-50 U/kg with a FX trough 2-2.5 U/dL. He remained asymptomatic, except for a slight bruising tendency (he plays soccer), and continued a weekly prophylaxis with PCC 30-40 U/kg. From 2020, due to post-infusion unspecific symptoms (nausea, sometimes dizziness) he performs premedication with Betametasone 1 mg, infusing PCC 25-30 U/kg weekly and FX trough between 1-1.8 U/dL. With the availability of pd-FX, in February 2025, a therapeutic switch is proposed to the patient with the aim of infusing only FX and making possible home therapy (CPP can be infused only in hospital) with an improvement in daily life. An infusion of 30 U/kg of pd-FX was made with the following FX troughs: basal 1.5 U/dL, +1 h 52.3 U/dL, + 24h 27.6 U/dL, +96h 5.3 U/dL, +168 h 1.9 U/dL. He continued prophylaxis with pd-FX 30 U/kg per week and 3 months after the switch FX trough is 2.1 U/dL. No side effects were reported. Since the patient continues to refuse biweekly prophylaxis, an increase to 50 U/kg is aimed to achieve higher FX trough (5 U/dL).

Conclusions: The availability of effective and safe high-purity pd-FIX concentrate is an important therapeutic option for the treatment of people with inherited FX deficiency.

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