A Case of an Isolated Factor X Deficiency

Shea Gallus, DO[1], Maggie Vogel, DO[1], Dr. Marie-Ellen Sarvida, MD[1,2]
[1]Advocate Children's Hospital Department of Pediatrics, [2]Department of Pediatric Hematology

Advocate Children's Hospital
Tomorrow starts today.

Introduction

Factor X deficiency is a rare blood disorder that causes prolonged bleeding due to a disruption in the clotting cascade. Factor X is a necessary clotting factor that is essential for proper clotting to stop bleeding (Fig. 1). Factor X deficiency is inherited in autosomal recessive pattern and usually due to a mutation in the F10 gene. The variable signs and symptoms makes this rare deficiency hard to diagnose.

Case

- 17yo previously healthy female with recent wisdom tooth extraction who presented for recurrent, persistent bleeding.
- Wisdom teeth removal 2 weeks prior to arrival, bleeding started 1 week after removal. Patient saw oral surgeon who sutured area and applied topical anticoagulant, bleeding stopped.
- Two days after, bleeding started again and increased over the next two days, which prompted visit to OSH ED
- Arrived at OSH with oozing of blood from left lower molar. Initial labs at OSH significant for Hbg 11.6, elevated PTT to 46, INR 1.7
- Transferred to ACH-OL for heme consult where she was taken to OR with OMFS to contain bleeding
- OMFS packed area with resolution of bleeding
- Hbg recheck <12hr later was 8.3
- Received IV iron and 1u pRBCs. Patient also completed 5 day course of TXA.
- Labs drawn showing isolated Factor X deficiency with 21% activity
- Pt discharged with no further bleeding
- Three months later, labs repeated which confirmed Factor X deficiency with 19% activity

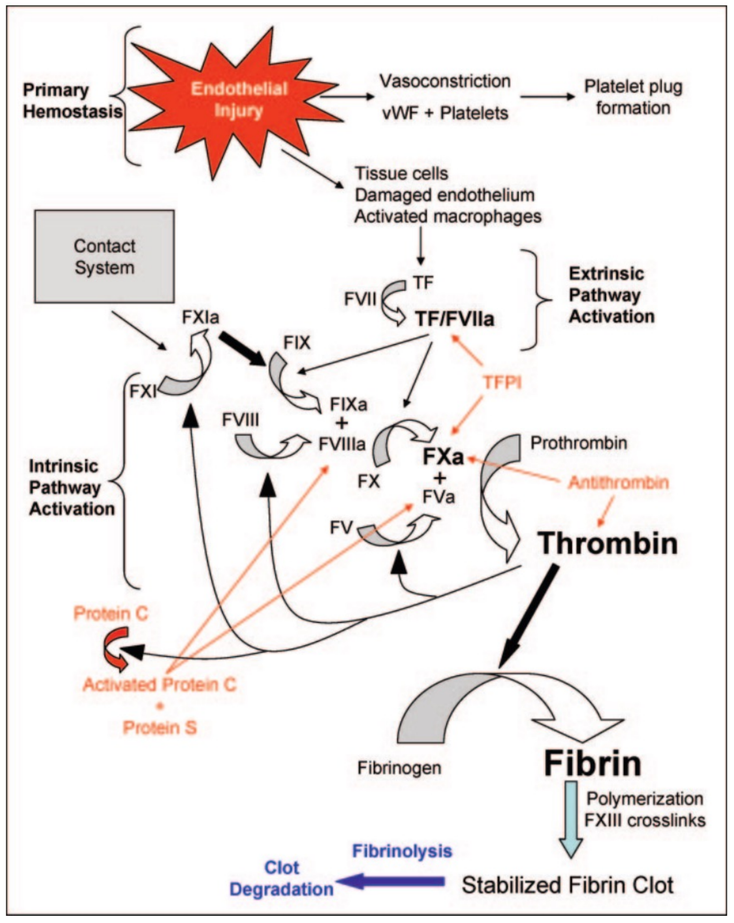

Fig. 1 – clotting cascade showing factors and their importance in creating a clot to control bleeding

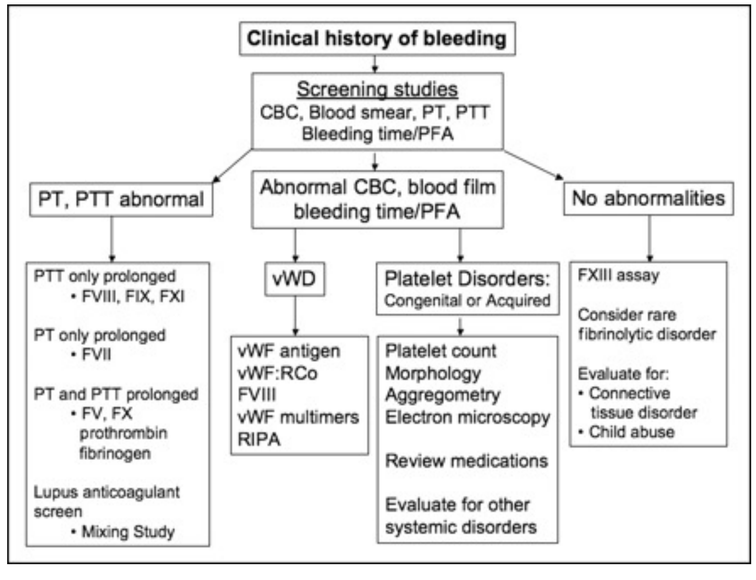

Fig. 2- Initial work up for persistent bleeding and how different abnormalities point to different clotting disorders

Case (cont.)

- Past medical history is significant for bleeding for <10 min post dental extraction as a child. Resolved with black tea bag, no further medical care needed. Patient and mother denied bleeding gums, heavy menstrual bleeding, or any other issues with bleeding
- Family history significant for mother with prolonged bleeding after tooth extraction as a child and father with blood transfusion as a child for unknown reason. Patient has no siblings.

Discussion

Initial work up for any persistent bleeding should include basic labs, including coagulation studies. In this case initial labs showed a prolonged PTT and PT (Fig. 2) which is seen with Factor X deficiency. Ultimately Factor X activity will need to be measured. Mild factor X deficiency is defined as >40% activity of the enzyme. Most patients with mild deficiency are asymptomatic but have bleeding during trauma, surgery, or childbirth. Moderate factor deficiency is defined as 10-40% of enzyme activity. In addition to bleeding during trauma, surgery, and childbirth, patients can have spontaneous bleeding. Severe deficiency is defined as <10% activity and these patients have life threatening bleeding. Our patient had moderate deficiency as defined by her 19% enzyme activity. This case illustrates the importance of a complete thorough history and potential for further bleeding if prompt diagnosis is not made. Although factor X deficiency is a rare autosomal recessive genetic disorder that occurs in 1:500,000 to 1:1,000,000 individuals it should be considered in any individual who presents with prolonged bleeding. Recognition of this rare bleeding disorder is essential for prevention of further bleeding by receiving the proper treatment, especially prior to procedures with bleeding as a known risk.

References

- Factor X deficiency. *Genetics Home Reference*. January 2015; http://ghr.nlm.nih.gov/condition/factor-x-deficiency.
- Schwartz, RA. Factor X Deficiency. *Medscape Reference*. Updated Mar 23, 2018; http://emedicine.medscape.com/article/209867-overview.
- Sharathkumar, Anjali A., and Steven W. Pipe. "Bleeding Disorders." *Pediatrics in Review*, The American Academy of Pediatrics, 29 Apr. 2008.