

Management of intrapartum bleeding risk in a case of Factor XI deficiency

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Background

Factor XI (FXI) deficiency is a congenital clotting disorder affecting 1 in 100,000 people worldwide. Severe deficiencies are defined as patients with FXI $\leq$ 20% of the normal, normal being defined between 60 -70 IU/dl. However levels are often not representative of level of activity. Due to this weak correlation between FXI levels and bleeding tendency, patients are often categorized into ‘bleeder’ phenotypes using bleeding symptoms such as heavy menstrual bleeding, bleeding after dental/surgical procedures, mucous membrane bleeding and easy bruising. However, as is often the case in medicine, FXI levels and historical care can often become the primary guide for treatment particularly in acute situations such as happened in this case. This is compounded in pregnant populations where interpreting the stability of FXI levels is difficult as studies show either stable concentrations or small fluctuations making levels even more inaccurate for care planning.

Introduction

We present a case of Factor XI (FXI) deficiency in pregnancy which demonstrates the poor correlation between FXI serum levels and bleeding propensity. Severe FXI deficiency often has a longer aPPT than severe Haemophilia A or B yet will bleed less. This apparent contradiction compared to more common haemophilia's can lead to lack of prognostic clarity making management in emergencies challenging.

Given that approximately 20% of FXI deficient women will experience pregnancy related bleeding, this rare disease can pose a significant management challenge to the obstetric team overseeing their care.

Case Report

A 33-year-old with mild FXI deficiency, diagnosed after episodes of epistaxis in childhood, was admitted for induction of labour (IoL) following a low risk antenatal period (aside from a minor first trimester bleed).

She had a history of bruising easily but reported no heavy menstrual or post-operative bleeding (a previous miscarriage had been treated via surgical management with no complications).

During her third trimester, Haematology MDT recommended that she receive Tranexamic acid (TXA) during labour as this was previously used to cover her miscarriage surgery and her FXI level had declined from 53 IU/dl to 41 IU/dl, despite being labelled as a non-bleeder in her pre-natal haematology clinics. The normal range for FXI levels is 70 – 150 IU/dl with severe disease exhibiting levels $<15-20$. Severe disease generally occurs in homozygous individuals, while a partial/moderate deficiency (levels of 20 -70) is more typical of compound heterozygous individuals

In labour she required syntocinon (oxytocin) augmentation, epidural and continuous CTG. Following a prolonged second stage, a Caesarean section was performed under general anaesthetic on the basis of bleeding risk posed to baby from a difficult instrumental and concerns for maternal bleeding risk.

She received further prophylactic Tranexamic acid peri-operatively and as a precaution, a Robinson's drain was left in situ for 24 hours. Her total EBL was 1000ml

Postpartum, she received a customized regime of tranexamic acid and LMWH with FXI monitoring by Haematology team.

Inheritance pattern

Factor XI deficiency is caused by mutations of the F11 gene resulting in a reduced plasma concentration of the activated factor X. FXI deficiency can be inherited in an autosomal recessive and dominant pattern. However majority of inheritance is autosomal recessive.

Though our patient's parents have never been tested for the deficiency, all of her siblings have been confirmed as Factor XI deficient and therefore it was safe to assume her family's inheritance pattern is dominant and therefore she is likely to have it to.

Conclusion

This case demonstrates how high-level interventions can occur for seemingly low-level issues when treating rare diseases that show weak correlation between measurable facets of the disease and more subjective elements. This was compounded by her unclear inheritance pattern requiring fetal precautions such as instrumental delivery avoidance.

By highlighting this case we hope to showcase a rare but prognostically complex disease which requires specialist MDT input from an early stage to assess bleeding phenotype rather than FXI levels and historical treatment to ensure personalised care planning, correct levels of clinical intervention and education of the caregiving team.