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PUBLISHED ABSTRACT

FXI Deficiency Found on Expanded Carrier Screening: A Quandry in Routine Obstetric Management

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Background

The routine use of prenatal carrier screening has resulted in increased identification of carriers for Factor XI (FXI) deficiency, a rare inherited coagulation disorder with variable bleeding phenotypes. The objective of this study is to describe the management and pregnancy outcomes of patients with no bleeding history who screened positive for FXI deficiency.

Methods

This is a prospective analysis of data obtained from asymptomatic patients who tested positive for FXI deficiency through expanded carrier screening between 01/2014 and 05/2018. We describe characteristics of this population and clinical history that may be associated with an increased risk of bleeding complications, management of these patients, and pregnancy outcomes.

Results

46 patients, of whom 1 was homozygous (FXI level < 11%) and 45 were heterozygous for FXI deficiency, were included in the analysis. 76% of the heterozygous patients reported a prior hemostatic challenge (major surgery or wisdom tooth extraction) with only 2 having had excessive bleeding. Median FXI level for the 43 heterozygous patients with no history of increased bleeding was 48% (IQR: 41:58). None of these patients had levels below 15%; 16% were between 26–35% and 28% between 36 and 50%. The rest of this cohort had FXI levels >50%. 6 patients received FFP (1–3 Units) prior to regional anesthesia or delivery based on hematologist recommendation and none received TXA prophylaxis. Median FXI levels for the 6 patients that received FFP prophylaxis was 34% (IQR: 30, 42), compared to 48% (IQR: 41, 58) in the group that did not. None of the remaining patients required FFP or TXA at the time of delivery for excessive bleeding and 26 of them (70%) received regional anesthesia uneventfully. Only 1 patient required FFP after c-section due to a subcutaneous hematoma (FXI level 32%). Other antenatal and delivery outcomes of the 43 heterozygous patients with no bleeding history are shown in **Table 1**.

Conclusions

Patients found incidentally to have FXI deficiency during carrier screening have a wide range of FXI levels and guidance on the obstetric management of these patients is non-existent. The majority of patients have an uncomplicated course of labor and delivery and do not require prophylactic treatment with FFP or TXA. More studies are needed to further establish the optimal approach to these patients.

Table 1: Antenatal and delivery outcomes of the 43 heterozygous patients with no bleeding history.

	N	FXI levels (%)
Antenatal bleeding	0	
Subchorionic hematoma	2	31% 42 %
Postpartum hemorrhage	1	43%
Late postpartum hemorrhage	0	0

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