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CASE REPORT

A Rare Case of Symptomatic Factor XII Deficiency Manifesting as Intraventricular Haemorrhage and Hydrocephalus in a Term Neonate

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Abstract

Background: Factor XII (Hageman Factor) is the initiating factor for the Intrinsic Pathway of Coagulation cascade. Literature describing bleeding tendencies in Factor XII deficiency is scarce.

Case: An 18-day-old boy presented with symptoms suggestive of raised intracranial tension and new onset convergent squint. CT scan revealed intraventricular bleeding with hydrocephalus. Platelet count, PT and aPTT were within normal limits. Coagulation profile revealed severe factor XII deficiency.

Keywords

Factor XII deficiency, Intraventricular haemorrhage, Hydrocephalus

Standard Abbreviation

PT: Prothrombin Time; aPTT: Activated Partial Thromboplastin Time; INR: International Normalised Ratio

Introduction

Factor XII (FXII) deficiency, which is also known as Hageman factor deficiency, was first described in the medical literature by Dr. Oscar Ratnoff and Dr. Jane Colopy in 1955. It has an Autosomal Recessive inheritance. The precise incidence of the disorder in the common population is still unknown, but it is found approximately one in a million [1].

FXII plays a dual role in the cascade reactions of coagulation. It can stimulate FXI, which, in turn, activates FIX (previous intrinsic pathway). Factor XII auto activates by contact with a variety of artificial or biologic negatively charged surfaces (activation of contact), resulting in blood coagulation and activation of the inflammatory kallikrein - kinin and complement systems. Most biologic surfaces that activate factor XII become expressed in disease states, though the major function of factor XII is the initiation of fibrinolysis and clot stability [2,3].

Asymptomatic prolongation of aPTT is the most commonly reported manifestation of factor XII deficiency [4]. Immune Thrombocytopenic Purpura like picture was described by Kumar, et al. [5]. Association with congenital abnormalities [6] and ischemic strokes have also been reported [7,8]. Factor XII deficiency has been associated with recurrent abortions in pregnant women [9,10]. Literature describing bleeding tendencies in Factor XII deficiency is scarce.

Trauma and perinatal asphyxia are the most common causes of Intraventricular Haemorrhage in term neonates [11,12]. Though Haemophilia B has been described to cause extracranial bleed [13], coagulation factor deficiencies have rarely been described as the etiology for intraventricular haemorrhage in this age group.

In this article we report a rare case of intraventricular haemorrhage and hydrocephalus secondary to Factor XII deficiency in a term neonate.

Case

An 18-day-old term boy baby with a birth weight of

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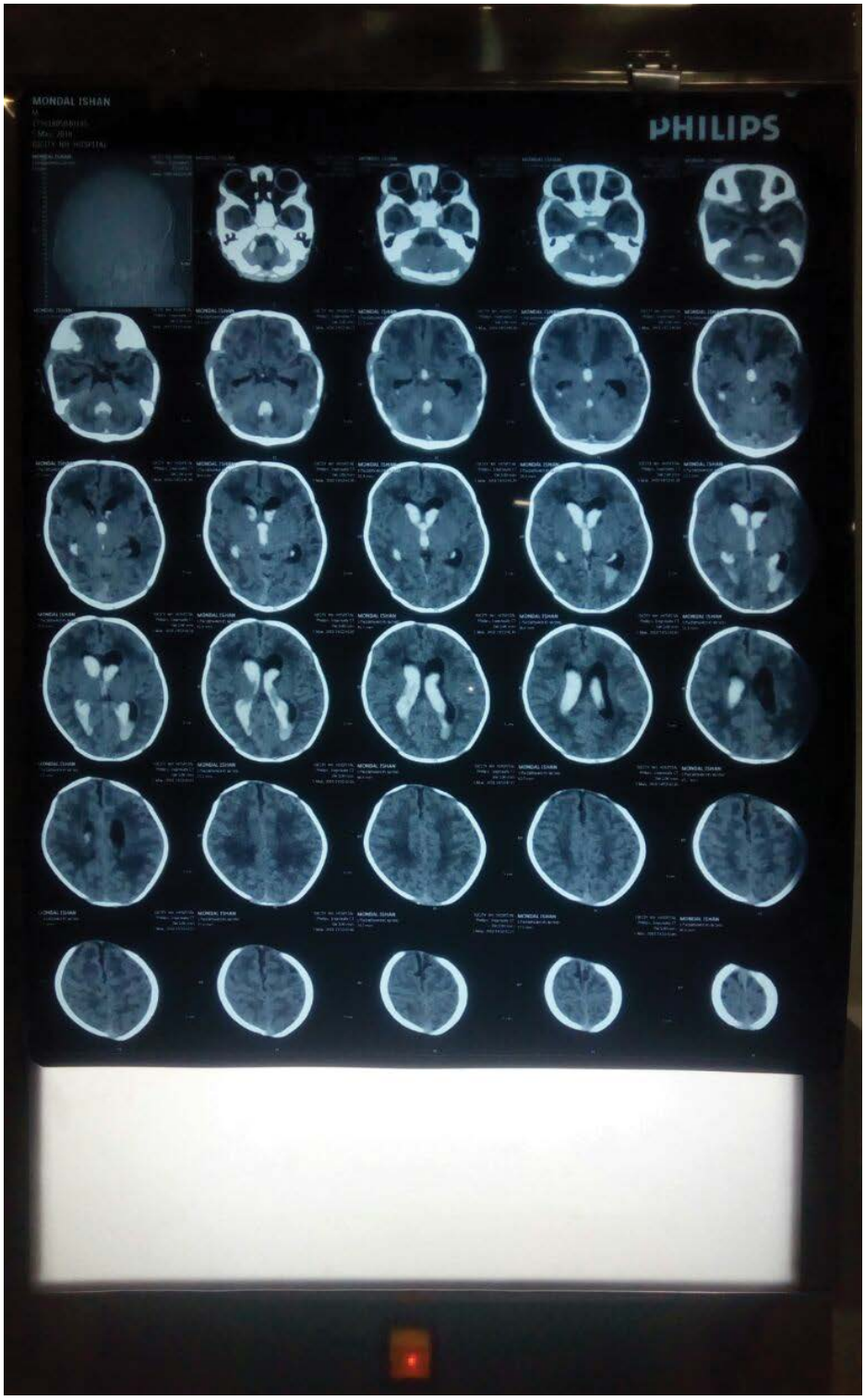

Figure 1: Grade III Intraventricular haemorrhage with dilation of ventricles.

Reference: Department of Pediatrics, IQ City Medical College and Narayana Multi-specialty Hospital, CT Scan done on 5/4/2018.

3 kg, presented with refusal of feeds, shrill cry, intermittent stiffening of limbs and new onset squint. The mother had history of recurrent abortions. Otherwise, the antenatal history was insignificant. Examination revealed increased tone, bulging anterior fontanelle, sutural diastasis and convergent squint. Lumbar puncture resulted in haemorrhagic tap. CT scan revealed intraventricular bleeding with hydrocephalus (Figure 1). Blood, urine and cerebrospinal fluid culture were negative. Normal platelet count, PT and aPTT reports were normal. Coagulation profile revealed severe factor XII deficiency (Observed value: 33.1%). Mixing studies were done, results were negative. Other coagulation factor values were within normal levels. This case was managed conservatively till hemodynamic stability was achieved and referred for definitive management of the hydrocephalus.

Discussion

The baby did not have any history of perinatal asphyx-

ia or birth trauma. Thus congenital thrombocytopenia and coagulopathy were the next possibilities. However, the platelet count and PT, aPTT and INR reports were within normal limits. This prompted the team to investigate for other coagulation factors of which factor XII which was deficient. The mother was asked to get her factor XII assay done in view of bad obstetric history.

Intraventricular haemorrhage secondary to Haemophilia B has been described by Bhattacharya, et al. [14]. But extensive literature search could not locate Factor XII deficiency as an etiology for Intraventricular haemorrhage.

Severe deficiency of factor XII in this baby may have caused abnormalities in intrinsic pathway leading to intraventricular haemorrhage.

Conclusion

Factor XII deficiency is asymptomatic with *in-vitro* prolongation of aPTT. We would like to conclude by emphasizing that factor XII deficiency may also present with haemorrhagic manifestations as in our case.

Disclaimer

None.

Sources of Support

None.

Declaration on Competing Interests

None.

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