Case Report

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A Case of Infant with Factor VII Deficiency Presenting as ICH

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Abstract

ICH can be spontaneous or traumatic. the most common cause of ICH in adults is trauma(road traffic accident or fall from height) and CVA. In neonates and infants ICH is caused by trauma associated with labor and delivery. Factor VII deficiency presenting as ICH on an infant is a rare entity. Here we present a case of 40 day old male child presenting as seizure which was later diagnosed to have ICH due to severe factor VII deficiency.

Keywords: Intracranial Haemorrhage; Factor VII Deficiency; Seizure; Prothrombin Time; Haemorrhage.

Introduction

The most common cause of ICH in adults is trauma and CVA.

Whereas the causes of ICH in neonates and infants include:

- Trauma associated with labor and vaginal delivery
- Acidaemia
- Hypoxia
- Hypercarbia
- Immaturity of the coagulation system, hereditary disorders/syndromes.

The majority of neonates with intracranial haemorrhage have no clinical symptoms, including some with moderate to severe haemorrhages. Term newborns with intracranial haemorrhage may manifest with a neonatal seizure, decreased level of consciousness, or both.

Bleeding/clotting disorders are among the rare causes of ICH. Among these, Factor VII deficiency is the most common among rare inherited Autosomal recessive bleeding disorders. In spite being the most common, prevalence is estimated to be 1 case per 500,000 persons in the general population.

Factor VIIa can be detected in plasma by a sensitive assay using a recombinant soluble form of tissue factor. The mean plasma concentration is 3.6 ng/mL in healthy individuals. The half-life of factor VIIa is relatively long (2.5 h) compared with other activated coagulation factors.

Factor VII deficiency is an autosomal recessive disease, unlike haemophilia (X-linked recessive). Only homozygote or compound heterozygote patients with factor VII deficiency are symptomatic. Heterozygote who have partial factor VII deficiency may not exhibit hemorrhagic manifestations, even following trauma. In symptomatic patients, clinical phenotypes vary from mild to severe and do not necessarily correlate with factor VII levels. A multicenter European study of patients who are congenitally factor VII deficient showed that clinical symptoms did not vary with the frequency of functional polymorphisms and that homozygote with the same mutation presented with striking differences in severity of bleeding.

The most frequently reported bleeding symptoms among "platelet-like" FVII deficiency are-

- Epistaxis (60%),
- Gum bleeding (34%),
- Easy bruising (36%),
- Menorrhagia (69% of females).

Bleeding Risk	Factor VII (%)	Personal History	Family Hisotry
High risk	<2	CNS bleed, umblical stump bleed, hemarthrosis, GI bleed	Life-threatening bleeding, death for hemorrhage in first degree relatives
Low risk	>20	Negative for spontaneous bleed	Negative for spontaneous bleeding

Among the severe forms-

- Recurrent hemarthrosis (19%)
- Gastrointestinal bleeding (15%)
- Central nervous system bleeding (2.5%)

Case Report

Forty days male child presented to ER with complains of (Historian-mother) abnormal movement of the body from 1 day. There was no history of trauma/fall/fever/cold/cough/loss of consciousness. The patient was admitted in another hospital for 1 day where NCCT head was done which was suggestive of large hyperdense heamorrhage in right fronto-accipital region with perifocal edema and mass effect on right lateral ventrical and midline shift to left side. The patient was managed conservatively.

On arrival, the child was conscious, playful, and all vitals were within normal range according to age. The systemic examination was unremarkable except increased tone and brisk deep tendon reflexes. There was history of prolonged umbilical bleed after birth. Patient has a positive family history of death of elder brother at 6 months of age with history of patecheal spots all over the body.

MRI brain with contrast was done which was suggestive of Intraventricular hemorrhage in left lateral ventricle and fourth ventricle, Supra and infratentorial subdural and subarachnoid hemorrhages. Neurosurgery consult was taken and patient was admitted in PICU after starting antiepileptics, measures to decrease ICP and Inj Vit K. Routine investigations were sent which included complete hemogram, liver function test, renal profile, coagulation profile. Investigations revealed Hemoglobin of 9.8gm/dL, Prothrombin time >1min. peripheral smear for type of anaemia was suggestive of normocytic normochromic anaemia.

The initial investigation was suggestive of anaemia and prolonged PT. Accordingly, factor VII assay was sent and plan to replace factor VII was made. Lab values showed factor VII to be <1%, Hematology consult was taken and so accordingly factor VII was transfused. The patient was discharged 21 days after admission in a stable condition with no new bleed. A follow-up CT of the brain at 1 month showed a resolving ICH.

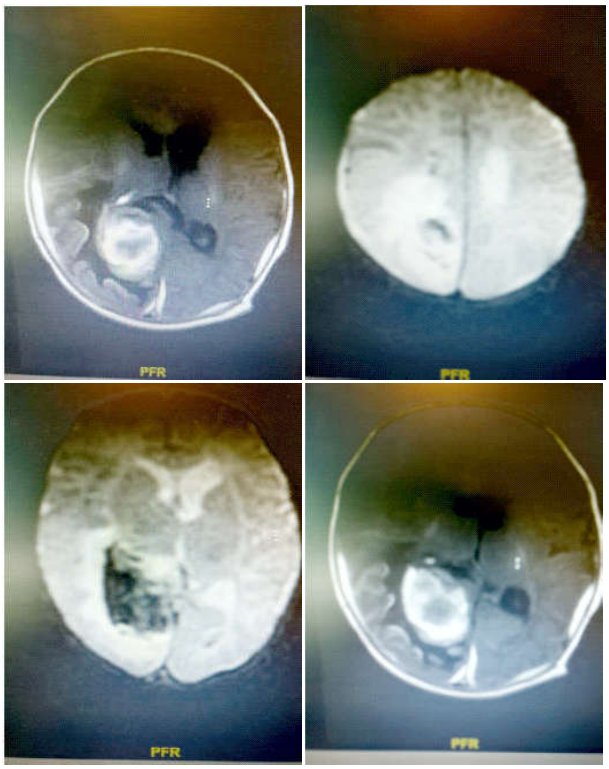

Fig. 1:

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