Case Report

Posterior Fossa Hematoma Following Minor Trauma in an Infant with Rare Combined Factor V and Factor X Deficiency

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Congenital combined deficiency of factor V and factor X deficiency is extremely rare. We report this for the first time in literature in an infant who developed acute subdural hematoma in posterior fossa leading to hydrocephalus.

KEYWORDS: Factor deficiency, factor V, factor X, hydrocephalus, posterior fossa hematoma

INTRODUCTION

Both factor V (proaccelerin or labile factor) and factor X (Stuart–Prower factor) are synthesized in the liver. The synthesis of factor X is vitamin K dependent, whereas factor V synthesis does not need vitamin K. Both factors act in the common pathway of coagulation to form thrombus. Combined deficiency of both factor V and factor X is extremely rare and it is reported once earlier in the literature where a 20-year-old lady developed secondary postpartum hemorrhage in her first pregnancy.[1] Our case is the first report in infant and second overall.

CASE REPORT

This 10-month-old girl presented with history of minor trauma in head followed by repeated episodes of vomiting without any episodes of seizure and loss of consciousness. She was brought to our emergency department after 3 days of that event and on examination she was drowsy but arousable, opening eyes, and crying on mild-to-moderate painful stimulus. Her anterior fontanelle was full. Non-contrast computed tomography (NCCT) scan showed posterior fossa acute subdural hematoma causing obstructive hydrocephalus [Figure 1]. Her blood test showed prolonged prothrombin time (PT) of 58 s, international normalized ratio (INR) of 6, activated partial thromboplastin time (aPTT) of 128 s with normal platelet count and liver function test. Hematologist opinion was taken and on further workup for coagulopathy, deficiency of factor V and factor X (both test result 9%, normal value 70–120) was detected. She was managed with several units of fresh frozen plasma (FFP) transfusion and injection of vitamin K to correct the PT, INR, and aPTT levels. She was monitored in intensive care unit, and serial ultrasonography (USG) of head was done to know the change in ventricular size. Surgical intervention was the last option in our mind considering her coagulopathy. Fortunately, she did not deteriorate clinically and her head circumference was stable. After 5 days of conservative treatment, her anterior fontanelle was soft on palpation. Repeat NCCT brain after 10 days showed resolving hematoma and hydrocephalus with prominent sulci and cisterns [Figure 2]. A detailed family history and past medical history were obtained which revealed that there is a history of consanguineous marriage among the parents and mother had bleeding tendency in the form of hematemesis, malena, and erythema, which were treated conservatively without detailed coagulation workup. Mother had multiple missed abortions in the past and this is their precious baby after 10 years of infertility treatment in different form. The baby girl was born by normal delivery at term. Following birth, there is no history of abnormal umbilical cord bleeding. Coagulation workup of both parents was
normal. The parents were offered genetic counseling and, after genetic workup, both of them showed the carrier status of factor V and X deficiency.

**DISCUSSION**

The clinical manifestation or bleeding tendency depends upon percentage of factor deficiency. Severe form of deficiency may be incompatible with life whereas others bleed very early in their life. In clotting factor deficiency, even a minor trauma can produce bigger hematoma as the bleeding continues for longer than it should be. In our case, the minor trauma might have caused tearing of the underlying bridging vein in posterior fossa and factor deficiency has aggravated the bleeding.

Bleeding disorders are generally carried from parents to child in autosomal inheritance pattern. Both factor V and factor X are autosomal recessive coagulation disorders. Autosomal recessive means that each parent must pass on a defective gene in order for the child to manifest the disorder. Hence the incidence of Factor deficiency is higher in consanguineous marriages like in our case. factor V and factor X deficiency are due to mutation on chromosome 1 (1q23) and chromosome 13 (13q34), respectively. The incidence of isolated factor V deficiency is one in one million. Whereas, the incidence of isolated factor X deficiency
is one in two million.\textsuperscript{4} There are some case reports of combined deficiency of vitamin K-dependent factors, but combined deficiency of both factor V and factor X is extremely rare.\textsuperscript{1}

There is no concentrate containing only factor V available.\textsuperscript{5} So, FFP transfusion is the main mode of treatment. Prothrombin complex concentrate is the other alternative to treat factor X deficiency. One needs to be cautious during correction period because over correction of these factors can produce hypercoagulable complications.\textsuperscript{6} factor V level ranging 10–20\% of normal\textsuperscript{1} and factor X level ranging 10–40\% of normal\textsuperscript{6} are sufficient enough to cause hemostasis.

Regarding the management of posterior fossa acute subdural hematoma with hydrocephalus, there are different surgical options including only shunt procedure or external ventricular drainage, and posterior fossa craniotomy with or without shunt procedures.\textsuperscript{7} In our case, risk of surgery was more because of coagulopathy. In infants, serial USG of head is a very useful tool to monitor progression of hydrocephalus while waiting for surgery or being managed conservatively.\textsuperscript{8}

Genetic counseling should be offered to such families to identify existing factor deficiency and possible gene mutation.

\textbf{Conclusion}

Combined deficiency of clotting factor V and X is a rare congenital coagulation disorder. It is very challenging to manage such case when presents with brain hemorrhage. A dedicated multispecialty team involving neurosurgeon, pediatrician, hematologist, radiologist, and trained nursing team in critical care unit is essential for good outcome.

\textbf{Declaration of patient consent}

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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\textbf{Conflicts of interest}

There are no conflicts of interest.

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