Case Report

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Quick Response Code:

Website: www.ijhonline.org

DOI: 10.4103/ijh.ijh_6_21

Factor VII deficiency-related recurrent hemarthrosis in a female child – When to suspect?

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Abstract:

Recurrent hemarthrosis is a common entity in children. Although recurrent hemarthrosis most often associated with hemophilia (VIII or IX deficiency), but rarely it can be associated with factor VII deficiency (FVIID). It is a strong mimicker of hemophilic hemarthrosis. Once hemophilia is excluded as a cause of recurrent hemarthrosis, congenital FVIID needs to be considered for long-term planning of treatment and avoiding unnecessary transfusion of factor concentrates. Clinical presentation of FVIID has a varied spectrum and does not correlate with factor levels. Here, we present a case of recurrent hemarthrosis secondary to FVIID.

Keywords:

Factor VII deficiency, female, recombinant factor VIIa, recurrent hemarthrosis

Introduction

Recurrent hemarthrosis is mostly seen in children with factor VIII or factor IX deficiency. Rarely, it can be caused by other congenital coagulation disorders. Factor VII deficiency (FVIID)-associated recurrent hemarthrosis is even more uncommon. It is a strong mimicker of hemophilic hemarthrosis. Once hemophilia is excluded as a cause of recurrent hemarthrosis, congenital FVIID needs to be considered for long-term planning of treatment and avoiding unnecessary transfusion of factor concentrates. Clinical presentation of FVIID has a varied spectrum and does not correlate with factor levels. Here, we present a case of recurrent hemarthrosis secondary to FVIID.

Case Profile

A 7-year-old girl presented to our OPD with a 6-month history of spontaneous recurrent afebrile painful swelling of the right knee joint associated with limitation of movements. There was no other significant past or family history of bleeding or any other disorder. On local examination, there was swelling of the right knee and right elbow joints, along with warmth over both joints and moderate tenderness in the right knee joint [Figure 1a-c]. Her systemic examination was noncontributory. Initial clinical diagnosis of recurrent spontaneous hemarthrosis secondary to von Willebrand Disease (vWD) and hemophilia was considered. Her complete blood count showed normal hemoglobin, total leukocyte count, and platelet count with normal morphology. Her coagulation profile showed abnormal prothrombin time (PT) (68.8 s), international normalized ratio (5.73), and normal activated partial thromboplastin time (APTT) (24.9), hence clinical possibility of inherited FVIID was considered. Her factor assays were normal for factors VIII and IX while functional factor VII levels were found to be <2.0 IU consistent with severe FVIID. Acquired FVIID was ruled with no history of any drug intake and no evidence of any acute or chronic systemic illness, and her liver function tests were

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Submission: 08-02-2021
Revised: 17-03-2021
Accepted: 18-03-2021
Published: 21-06-2021

How to cite this article: Didel S, Purohit AH, Krishna D, Vyas V, Singh K. Factor VII deficiency-related recurrent hemarthrosis in a female child – When to suspect? Iraqi J Hematol 2021;10:82-3.

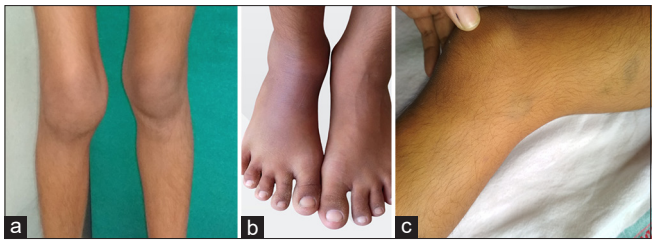

Figure 1: (a-c) Right knee (chronic stage) and right ankle (acute stage) joint hemarthrosis and ecchymotic patches on the right thigh

normal (alanine aminotransferase – 24 IU/L [13–45], aspartate aminotransferase – 20 IU/L [9–24], and serum albumin – 3.9 g/dl [3.4–5.4]) except isolated PT prolongation. She was given fresh frozen plasma (FFP) for 5 days and P.R.I.C.E. (Protect, Rest, Ice, Compression and Elevation) regimen therapy in view of nonavailability of recombinant activated factor VII (rFVIIa), and she responded well to FFP infusion. Parents were counseled about the need of rFVIIa infusion during recurrent attack of hemarthrosis.[1]

Discussion

After hemophilia and vWD, FVIID is the third most common cause of recurrent hemarthrosis, and probably the most common cause in females.[1,2] Depending on the age of onset, FVII level, and type of bleeding, it can be classified into three groups: mild, moderate, and severe bleeding.[3] Life-threatening bleeding like intracranial bleeding also may be presenting feature in 20%–25% of cases. It is more common during early neonatal or infantile period.[4] Congenital FVIID can be caused by more than 250 mutations, and the majority of them are missense mutations.[2] Risk of severe bleeding is more common in cases with compound heterozygous and homozygous mutation and can lead to premature death.[2] Early suspicion starts with isolated prolonged PT with normal APTT. FVIID can have similar clinical features of the musculoskeletal system as seen in cases with hemophilia.[5] The severity and frequency of joint bleed are less in case of FVIID hemarthrosis, although severe case of hemarthrosis has been reported in FVIID also. Nevertheless, common join involvement described in FVIID-induced hemarthrosis includes knee, ankle, hip, and shoulder joint hemarthrosis.[5] rFVIIa is the most preferred therapy in acute symptomatic patients. This case highlights that FVIID should be considered in any female child with recurrent hemarthrosis or bleeding from other sites along with other coagulation disorders.

Conclusion

- Spontaneous hemarthrosis always needs detailed evaluation to establish specific cause to plan further, and there is an urgent need to develop multidisciplinary treatment facilities for holistic care of these case
- FVIID is a specific cause which should be considered in any female child with recurrent hemarthrosis or bleeding from other sites after vWD.

Compliance with ethical standards

- Written informed consent was taken for publication of this manuscript.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the parents have given their consent for their child's images and other clinical information to be reported in the journal. The parents understand that their child's name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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