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Combined Factor V and Factor VIII Deficiency: A Case Report

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Abstract

Background: Combined factor V and factor VIII deficiency (F5F8D, OMIM 227300) is a rare autosomal recessive bleeding disorder. It seems more common among Jews and Iranians, particularly in regions with frequent consanguineous marriages. **Case Report:** We describe a 5-year-old girl born out of consanguineous marriage with a complaint of prolonged bleeding after dental extraction. There were no history of spontaneous bleeding and other coagulation defect symptoms. **Conclusion:** In frequent consanguineous marriage regions, inherited deficiency of factor V and VIII should be considered in patients with coagulation defect symptoms. [GMJ. 2016;5(1):42-44]

Key words: Combination; Factor V; Factor VIII

Introduction

Combined factor V and factor VIII deficiency (F5F8D, OMIM 227300) is a rare autosomal recessive bleeding disorder first described in 1954 by Orieri and colleagues [1]. Since then, more than 200 cases were reported all over the world. Noticeable number of these patients belonged to Mediterranean and Asian countries in particular the Middle East. Regarding autosomal recessive inherited pattern of this disorder, parents of these patients are obligatory heterozygotes. So, the prevalence of this rare mutation increases in regions with higher rate of consanguineous marriages like afibrinogenemia [2-4]. Simultaneous decrease of plasma factor V and

factor VIII as the underlying F5F8D characteristic leads to mild-to-moderate bleeding symptoms including: epistaxis, postsurgical bleeding, menorrhagia, hemarthrosis and muscular hematomas [5-7]. We report a patient suffering from F5F8D with no evidence of common symptoms before diagnosis.

Case Presentation

A 5-year-old girl born out of consanguineous marriage referred to our pediatric hematology clinic of Baqiyatallah hospital, Tehran, Iran for clarifying the reason of her extraordinary bleeding after dental extraction procedure. She did not have a history of spontaneous bleeding like epistaxis and gum bleeding or

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prolonged bleeding after skin rupture. But her history revealed the bruising after falls. Admission examination did not show ecchymosis or any joint swelling, and review of systems was remarkable. She did not have a history of spontaneous bleeding like epistaxis and gum bleeding or prolonged bleeding after skin rupture; although, her past history revealed the bruising after falls. The patient was hemodynamically normal with a temperature of 36.8°C, blood pressure of 110/ 58 mmHg, heart rate of 95 beats/min, and respiratory rate of 22 breaths/min. Laboratory investigations demonstrated an activated partial prothrombin time (aPTT) of 86.9s (control 35.5s), a prothrombin time (PT) of 21s (control 13.2s) and a bleeding time (BT) of 3 minutes. Because of prolonged PT and PTT, we doubted a common pathway factor deficiency including factor V and factor X, congenital Vitamin K deficiency and vitamin K related factor deficiency including factor II, factor VII, factor IX and factor X as differential diagnoses. The patient’s hemostatic assessment showed the level of factor II at 67%, factor V 10%, factor VIIIc 9%, factor IX 64%, factor X 45%, vWF activity (RCo) 48%, vWF antigen 55%, and fibrinogen 249 mg/dl. Our patient belonged to a 4-member family comprising of an older brother and consanguinity parents. There were no positive history and symptoms in fa-

vor of hematologic disorders. Even though, her mother had a PT of 14.7s (control 13.6s) simultaneously with factor VII 39% in her coagulation assessment (Table1).

After definitive diagnosis of combined factor V and factor VIII deficiency, the patient was asked on regular visits. The patient was also recommended to receive factor injection before any surgery procedure.

Discussion

Combined factor V and factor VIII deficiency is known as the most prevalent combined plasma coagulation factor deficiency [8]. The affected person was a case of F5F8D without classic manifestation described for this disorder. We reported this case because Iran is located in the most prevalent area for this disorder based on our literature review [8, 9]. So, each physician might be faced with patients suffering from this disorder. In the United Kingdom, Haemophilia Centre Doctors’ Organization guideline has been recommended to use fresh frozen plasma (FFP) as a part of treatment regimen which can provide FV and FVIII concentrate. Some previous reports have demonstrated that FFP containing regimen could successfully control bleeding during dental extraction, male circumcision and preparation for labor [10].

Table1: Laboratory Findings of Patient’s Family

Laboratory Parameters	Patient	Patient’s mother	Patient’s father	Patient’s brother
aPTT (control)	86.9s (35.5s)	36.1s (33.7s)	38.9s (33.7s)	42.8s (33.7s)
PT (control)	21s (13.2s)	14.7s (13.6s)	12s (13.6s)	12.7s (13.6s)
Factor V (Normal range)	10% (60-130)	71% (60-130)	96% (60-130)	107% (60-130)
Factor VII (Normal range)	-	39% (55-170)	-	-
Factor VIIIc (Normal range)	9% (60-150)	70% (60-150)	64% (60-150)	96% (60-150)

So, in patients with coagulation defect symptoms along with a history of parents with kindred marriage, inherited deficiency of factor V and VIII should be considered due to autosomal recessive inherited pattern of F5F8D. More investigations are needed to establish valuable guidelines for hemorrhage management in individuals with combined factor V and VIII deficiency.

Conclusion

Combined factor V and VIII deficiency seems more common among Iranians particularly in

frequent consanguineous marriage regions. So, inherited deficiency of factor V and VIII should be considered in patients with coagulation defect symptoms. Finally, we recommend the evaluation of factor VIII level in patients with prolonged PT and PTT and factor V deficiency.

Conflict of Interest

The authors report that there is no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

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