Scientific Research Publishing

Open Journal of Blood Diseases, 2026, 16(1), 35-39
https://www.scirp.org/journal/ojbd
ISSN Online: 2164-3199
ISSN Print: 2164-3180

Deficiency of Factor V of Coagulation: About a Case

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How to cite this paper: Djibrilla-Almoustapha, A., Malam-Abdou, B., Mousatapha, M.-B., Elhadji-Chefou, M., Adamou-Chaibou, O., Bouwe-Abdou, M., Abdoulaye-Soumana, O., Amadou-Adamou, H., Bassirou-Garba, A., Hama-Moussa, A., Ibrahim-Oumara, O., Maikabi-Nomaou, M., Mamoudou-Idrissa, B. and Samna-Kona, I. (2026) Deficiency of Factor V of Coagulation: About a Case. *Open Journal of Blood Diseases*, **16**, 35-39.
https://doi.org/10.4236/ojbd.2026.161005

Received: January 4, 2026
Accepted: March 16, 2026
Published: March 19, 2026

Abstract

Context: Factor V deficiency is a rare abnormality of coagulation as parahaemopilia, originally described by Owren in 1947, it is transmitted in an autosomal and recessive fashion. Its incidence is estimated at 1/10,000 in France. Clinical expressive is usually asymptomatic in the homozygous state. The diagnosis is based on a low prothrombin rate and normal TCK associated with a decrease in the factor of PFC in our context. **Results:** This was a 16-year-old male patient of normal build, born to consanguineous parents. He had sustained a head injury with the formation of an occipito-parietal hematoma. On admission, he was conscious, with moderate pallor of the skin and mucous membranes. The Complete Blood Count (CBC) showed normocytic anemia, a normal white blood cell and platelet count, PT at 59.5% and Activated Partial Thromboplastin Time (aPTT) Normal. Factor assays using a chronometric method revealed a factor V deficiency with normal factor VII. The patient received six transfusions of Fresh Frozen Plasma (FFP). The outcome was favorable, with progressive resorption and disappearance of the hematoma within a few days. **Conclusion:** Factor V deficiency is a very rare blood clotting disorder. It affects men and women equally and can, in some cases, cause significant bleeding.

Keywords

Factor V Deficiency, Onco-Hématology, National Hospital of Niamey

1. Introduction

Factor V is an essential cofactor in the conversion of prothrombin to thrombin by

activated factor X. Its deficiency, commonly known as parahemophilia, is rare. It is a congenital or acquired factor V coagulopathy that slows thrombin generation, thus delaying fibrin formation and leading to a tendency to bleed [1]. It is a disorder of the common coagulation pathway. Its incidence is 1/10,000 and its prevalence is 1/1,000,000 in the French population [1] [2]. The rare clinical manifestations are dominated by a hemorrhagic syndrome associated with a biological decrease in proaccelerin [3]. Biologically, it is distinguished by a disturbance in hemostasis parameters, namely a decrease in the Prothrombin Time (PT) without a disturbance in the activated partial thromboplastin time (aPTT). Its management consists of administering the missing factor. Therefore, we report a case of isolated factor V deficiency revealed by a post-traumatic intracranial hemorrhage.

2. Observation

Identity and History: The patient was a 16-year-old male of normal build, born to consanguineous parents, the seventh of ten children, all living, with a history of intermittent nosebleeds. There was no significant family history of illness. Following a road traffic accident that resulted in a head injury with the formation of an occipito-parietal hematoma (**Figure 1**).

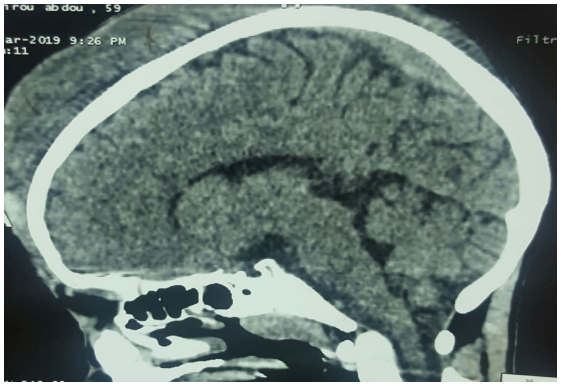

Figure 1. Subdural hematoma.

He underwent neurosurgical intervention, which was complicated by persistent bleeding at the suture sites. Due to the continued hemorrhage, he was transferred to hematology.

Diagnostic Evaluation: On admission, he was conscious, with moderate pallor of the skin and mucous membranes, and a blood pressure of 110/70mmHg.

A respiratory rate (RR) of 18 breaths/min, a pulse of 87 bpm, weight: 58 kg, and height: 1.60 m. A complete blood count (CBC) revealed normocytic anemia (MCV: 88.2 fl oz), normochromic anemia (MCHC: 34.5 g/dL), a normal white blood cell count (WBC: 8500/mm^3) and platelet count (Plq: 253,000/mm^3), a normal prothrombin time (PT): 59.5%, and a normal activated partial thromboplastin time (aPTT). Factor assay using a chronometric method revealed a factor V deficiency of 17% (normal range: 62% - 150%) with a normal factor VII level of 121% (normal range: 67% - 143%).

Therapeutic Data: Due to the unavailability and high cost of recombinant factor V in Niger, the patient received 6 transfusions of Fresh Frozen Plasma (FFP). The outcome was favorable, with progressive resorption and disappearance of the hematoma within a few days.

Discussion: Factor V deficiency is a rare coagulation disorder, more common in regions with high levels of consanguinity. In 2018, only 150 cases were reported in the literature [4] [5]. In France, its incidence is 1/10,000, with a prevalence of homozygous forms of 1/1,000,000 inhabitants [6]. Other cases have been reported in India, Japan, North America, and Europe [7]-[11]. It can occur in both men and women and can manifest at any age, but generally, severe forms appear early in childhood. The reported case is a symptomatic male with the condition, aged 16 years. Clinical manifestations are variable and the most frequent are epistaxis, post-traumatic ecchymoses, bleeding of mucous membranes or soft tissues, hemarthrosis, menorrhagia in young girls and women or hemorrhages after an invasive procedure (circumcision or tooth extraction, surgical interventions). More rarely, deep hematomas occur. According to the literature, bleeding associated with factor V deficiency is generally benign [12]. In severe cases, antenatal or postnatal cerebral hemorrhages are exceptional [13]-[15]. No ethnic predisposition has been reported [16]. Our patient presented with a post-traumatic subdural hematoma that led to the diagnosis of the deficiency. The diagnosis is suggested by a decreased prothrombin time (PT) and a normal activated partial thromboplastin time (aPTT), indicating involvement of the common pathway. It is confirmed by a decreased factor V level. Molecular analysis is possible but not necessary for diagnosis. This was the case for our patient, with a factor V level of 17%. According to the literature, a specific factor V assay is necessary to confirm the diagnosis. Factor V deficiency is diagnosed if the factor V level is below normal [17]. Veterinary factor (VF) concentrate remains the treatment of choice for managing this condition. In its absence, fresh frozen plasma (FFP) is the only recommended treatment [18]. It can be administered once daily for seven days at the time of surgery, and in extreme cases of severe hemorrhage, platelet concentrate transfusions may be useful in addition to FFP [3] [19]-[23]. According to the guidelines, even without obvious bleeding, a report should be filed. All questions related to coagulation tests will be addressed, which can facilitate management. This was the case for our patient, who received FFP transfusions and whose condition improved with desorption of the hematoma within a few days.

3. Conclusion

Factor V deficiency is a rare, autosomal recessive coagulopathy that can manifest as a hemorrhagic syndrome of varying severity.

Ethical Aspects

This publication complied with ethical and professional standards, in particular the protection of patient identity and their approval regarding the use of images

for educational purposes.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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