World Journal of Biology Pharmacy and Health Sciences
eISSN: 2582-5542
Cross Ref DOI: 10.30574/wjbphs
Journal homepage: https://wjbphs.com/

(CASE REPORT)

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Congenital afibrinogenemia and splenic rupture: A case report

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World Journal of Biology Pharmacy and Health Sciences, 2026, 25(01), 204-208

Publication history: Received on 09 December 2025; revised on 19 January 2026; accepted on 21 January 2026

Article DOI: https://doi.org/10.30574/wjbphs.2026.25.1.0032

Abstract

Congenital afibrinogenemia is an extremely rare coagulation disorder affecting 1-2 individuals per million. We report the case of a 15-year-old male with known congenital afibrinogenemia who developed hemorrhagic shock following minor soccer ball trauma to the abdomen. Clinical examination revealed hemodynamic instability with hypotension (90/40 mmHg) and tachycardia (140 bpm), associated with abdominal defense. Laboratory tests showed severe anemia (Hemoglobin 7 g/dL), undetectable fibrinogen (<0.2 g/L), and prolonged coagulation times. Abdominal computed tomography revealed massive hemoperitoneum with splenic involvement and a hematoma of the greater gastric curvature. Emergency exploratory laparotomy with gastrosplenic disconnection and surgical hemostasis was performed. The patient received blood transfusions, fresh frozen plasma, and 9 grams of fibrinogen concentrate, with full recovery and discharge on day 7. Splenic rupture occurs disproportionately in afibrinogenemia patients despite the rarity of the condition. Management requires prompt diagnosis, fibrinogen replacement therapy, and surgical intervention when indicated, along with preventive education regarding high-risk activities.

Keywords: Congenital Afibrinogenemia; Fibrinogen; Hemorrhagic Shock; Splenic Rupture; Case Report

1. Introduction

Congenital afibrinogenemia is an extremely rare and serious hematologic pathology, the clinical manifestations of which vary in onset and severity, ranging from minimal bleeding to hemorrhagic shock at birth or later. Nowadays, just over 250 cases have been published. We report the observation of a 15-year-old male adolescent, suffering from congenital afibrinogenemia discovered at birth, hospitalized in medical intensive care for hemorrhagic shock because of a minor abdominal trauma. The aim of this article is to establish the diagnostic and therapeutic approach for this condition in a surgery departement.

2. Case Report

This is a 15-year-old adolescent, the only child of non-consanguineous parents, suffering from congenital afibrinogenemia discovered on the 5th day of life following a persistent umbilical hemorrhage and an undetectable fibrinogen level.

He was admitted to the emergency department for hemorrhagic shock with acute abdominal pain and vomiting occurring 48 hours after minor trauma to the epigastric and left hypochondriac regions caused by a direct soccer ball impact.

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It is noted that the patient benefited from infusions of fresh frozen plasma during 2 bleeding episodes, without taking any prophylactic treatment.

On admission, the patient was conscious, afebrile but hemodynamically unstable with mucocutaneous pallor, hypotension (90/40 mmHg) and tachycardia at 140 bpm. Abdominal examination revealed abdominal distension related to fluid effusion and diffuse abdominal defense.

Laboratory evaluation demonstrated microcytic hypochromic anemia with a hemoglobin level of 7 g / dl, hematocrit of 22%, hyperleukocytosis at 23,470 / mm3 predominantly neutrophils and thrombocytosis at 439,000 / mm3. The hemostasis assessment objectified a profoundly reduced prothrombin time at <15%, markedly prolonged activated partial thromboplastin time at > 4 and a fibrinogen at <0.2 g / l.

Abdomino-pelvic ultrasound showed a very abundant effusion with finely echogenic content. The CT scan confirmed the hemoperitoneum at the perisplenic level, left parietocolic and hypogastric groove which is associated with heterogeneous splenomegaly and subhepatic air bubbles related to pneumoperitoneum, suggesting a perforation of a hollow organ (Figure 1).

Faced with hemodynamic instability, the patient was hospitalized in intensive care where he received a transfusion of 3 red blood cells and 6 fresh frozen plasma, then was sent to the operating room for an exploratory laparotomy and a gesture of hemostasis. Exploration of the abdominal cavity above and below transverse mesocolic did not show any perforation at the level of the antro-pyloro-duodenal outlet. In addition, there was a large hematoma of the greater curvature extending to the gastro-splenic meso and which seemed related to the origin of the bleeding. A gastro-splenic disconnection by section and ligation of the gastrosplenic meso and short vessels was performed in addition to a hemostasis suture on the greater curvature and placement of hemostatic material in contact with the spleen.

Postoperatively, the patient was hospitalized in intensive care where he received 6 red blood cells, 5 infusions of fresh frozen plasma and 9 grams of human fibrinogen with good clinical outcome, drains reporting only traces of blood, without any bleeding or thromboembolic complication, and biological with normalization of the hemoglobin level at 11.1 g/dl and the fibrinogen level at 0.80 g/l. The patient was declared discharged on D7 and received therapeutic education including exclusion from high-risk sports and banned drugs, regular monitoring and what to do with bleeding. The follow-up after 1 month showed a hemoglobin level of 11.8 g / dl and a fibrinogen level of <0.22 g / l (Tableau 1).

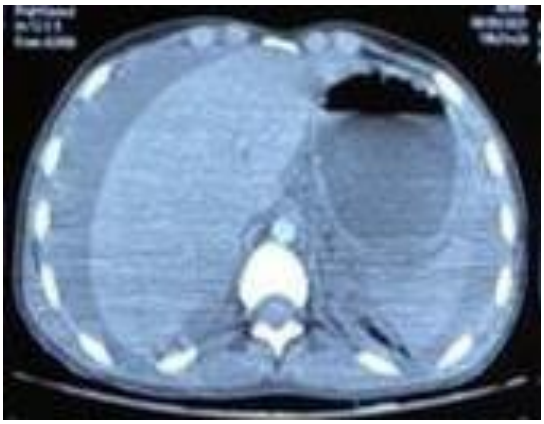

Figure 1 Abdominal computed tomography scan showing massive hemoperitoneum with splenic involvement following minor abdominal trauma in a patient with congenital afibrinogenemia

Table 1 Laboratory findings on admission and after treatment

Parameter	On admission	After treatment
Hemoglobin (g/dL)	7.0	11.1
Hematocrit (%)	22	34
White blood cells (/mm^3)	23,470	Normal
Platelets (/mm^3)	439,000	Normal
Prothrombin time (PT, %)	<15	Normalized
Activated partial thromboplastin time (aPTT)	>4	Normalized
Fibrinogen (g/L)	<0.2	0.80

3. Discussion

Fibrinogen is a large glycoprotein made by parenchymal cells in the liver. It is the third most common protein in plasma, with a concentration of 1.4 - 4 mg / ml. It is a multifunctional molecule endowed with many biological activities, the most important of which are undoubtedly those which give it an essential role in the general process of primary hemostasis which includes all the events and reactions which contribute to the formation of the hemostatic nail at the site of the vascular injury [1].

Congenital afibrinogenemia is an exceptional hereditary disease, with a notion of consanguinity in approximately 50% of the families studied [2,3]. Its frequency is estimated internationally at 1-2 cases per 1 million people. In our country, there are no data on the prevalence of afibrinogenemia. Unlike hemophilia A where the prevalence is stable around 1/5000 male subjects, afibrinogenemia affects both sexes with a sex ratio of 1 and its incidence varies according to the regions, explained mainly by endogamy, frequent in Iran and Lebanon [4,5,6].

The diagnosis is reminiscent of hemophilia and can therefore be suspected in the neonatal period by the occurrence of hemorrhage from the umbilical cord, or later by ecchymosis, hemorrhage following circumcision, bleeding gums, epistaxis, gastrointestinal hemorrhage, genitourinary or certain locations that can make the disease serious, such as rupture of the spleen or liver or intracranial hemorrhage. The fibrinogen level is often undetectable [7].

In front of an abdominal trauma, some areas are more vulnerable than others to be prone to lesions such as the liver because of its volume and its location and the spleen because of its fragility, which can bleed profusely and cause hemorrhagic shock in the absence of effective coagulation.

We report the case of a known patient with congenital afibrinogenemia admitted for epigastric pain and vomiting after a benign trauma to the abdomen revealing a splenic fracture associated with a hematoma of the greater gastric curvature resistant to resuscitation measures.

Note that ruptured spleen appears to be more common in patients with afibrinogenemia than in subjects with other congenital coagulation disorders. Indeed, despite a 100 times higher incidence of hemophilia [8], there is an equal number of reported cases of splenic fracture in hemophiliacs [9,10] than in patients with afibrinogenemia.

Compared to a normal subject, the management is different. That said, in the face of any serious trauma, management is mainly symptomatic by vascular filling, blood transfusions and, if necessary, angioembolization or even direct hemostasis surgery.

In our case, the treatment was essentially based on substitution therapy in the first place [11], the source of which was whole blood which contains 0.1 to 0.2 g / 100mL of fibrinogen, which remains insufficient to ensure correct coagulation and plasma fresh frozen which contains approximately 0.5 g of fibrinogen, a dose proving to be temporarily beneficial for coagulation while awaiting the availability of the fibrinogen concentrate. It is currently the most frequently used in the United States and Europe [12].

It is recommended to administer loading doses of 20 to 25 mg / kg of fibrinogen for 24 hours and then give the third of this dose on the following days [13]. The most common side effect associated with this treatment is the risk of

thrombosis. It is therefore preferable to give small doses of fibrinogen at short intervals. DDAVP, by increasing Von Willebrand factor, and anti-fibrinolytics are also two adjunct treatments that may have a beneficial effect, but this has not been observed by all authors.

Otherwise, surgical hemostasis decisions must be taken without delay in the face of hemodynamic instability in order to avoid the complications of massive transfusion. It is imperative to proceed with the rules of "damage control" requiring a shortened laparotomy and control of bleeding sites. It is preferable to opt for conservative surgery in children and young subjects, the main purpose of which is to avoid post-splenectomy infectious complications and restrictive preventive measures for the splenectomized patient.

Regarding the prevention of hemorrhagic events, an intramuscular injection or the taking of an NSAID should be avoided. It is also important to educate the child and parents to integrate the disease into their daily life by avoiding sports at risk of falling such as boxing, judo, football and domestic accidents, by learning what to do in the event of bleeding, and by consulting quickly in the event of severe symptoms. Finally, genetic counseling and awareness for screening in the event of a new birth are essential.

It is also recommended to consult a dentist every six months to prevent dental problems and gingivitis and to contact a hemophilia center if surgery or tooth extraction is scheduled in order to plan adequate preventive treatment.

Wearing an afibrinogenemia bracelet or card containing essential information on the proper management of the patient is necessary in the event of an accident, for the rescue teams.

All these preventive measures have been established in the face of the vital prognosis which is darker than in hemophilia.

Close collaboration between the patient and / or his relatives, the CRTH or CTH and pediatricians, general practitioners or specialists is essential for optimum follow-up.

4. Conclusion

In conclusion, hematoma of the spleen secondary to benign trauma to the abdomen is frequently observed in patients with congenital afibrinogenemia and is a real challenge for clinicians and surgeons. The diagnosis can be made in the face of acute epigastralgia, hypotension or even a state of shock. Supportive treatment, administration of fibrinogen and shortened laparotomy, where "damage control" and selective embolization have been shown to be effective, should help overcome hemorrhagic abdominal trauma.

Compliance with ethical standards

Acknowledgments

The authors would like to thank the medical and nursing staff of the Cheikh Khalifa International University Hospital for their contribution to patient care.

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of ethical approval

The study was conducted in accordance with ethical standards. Approval was obtained from the appropriate ethics committee.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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