Case Report

Bleeding Without a Wound: Spontaneous Splenic Rupture in a Child With Congenital Afibrinogenemia

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Congenital afibrinogenemia, a rare bleeding disorder (1–2 per million) characterized by a complete absence of fibrinogen, presents a high risk for life-threatening spontaneous splenic rupture. It is an autosomal recessive disease and occurs as a result of mutation in one of the three genes which code the three polypeptide chains of fibrinogen. In this case report, we present a 13-year-old boy who presented with abdominal pain, vomiting, and giddiness. On examination, he had pallor, abdominal guarding, tachycardia, and hypotension. Laboratory investigations indicated severe anemia with low hemoglobin, prolonged prothrombin time (PT) and activated partial thromboplastin time (APTT), and a normal platelet count. Contrast-enhanced computed tomography (CECT) revealed splenic hematoma with hemoperitoneum secondary to splenic rupture, while Histopathological examination (HPE) findings suggested a congested spleen. Bedside point-of-care ultrasound (POCUS) showed free intraperitoneal fluid, and Rotational thromboelastometry (ROTEM) reported absent clot firmness suggestive of severe coagulopathy. Cryoprecipitate transfusion was administered, and emergency splenectomy was subsequently performed after achieving ROTEM correction. The patient was prescribed aspirin, paracetamol, penicillin, omeprazole, and vitamin C supplementation on discharge. This case highlights the critical role of early bedside POCUS and ROTEM assessment in enabling prompt diagnosis and effective surgical and therapeutic management.

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Introduction

Congenital afibrinogenemia is a rare, genetically inherited fibrinogen disorder in which blood fails to clot normally because of the absence of fibrinogen, a blood protein essential for coagulation [1]. This disorder follows an autosomal recessive pattern, indicating that two unaffected parents may have a child affected with the disorder [2]. In patients with afibrinogenemia, umbilical bleeding, soft tissue and mucosal bleedings, menorrhagia, gingival bleedings, and bleeding in the mouth may be observed frequently, with bleeding episodes being excessive and, at times, controllable [3].

In the case of congenital afibrinogenemia, spontaneous splenic rupture occurs due to minor or unnoticed microvascular injuries within the highly vascular splenic parenchyma; because the complete absence of fibrinogen prevents stable blood clot formation, these trivial micro-bleeds expand uncontrollably into subcapsular hematomas until the stretched splenic capsule tears under pressure [4]. Spontaneous splenic rupture is an unusual finding in patients with congenital afibrinogenemia [5-7].

Point-of-care ultrasound (POCUS) is a rapid, non-invasive bedside modality that plays an important role in the early detection of splenic injury by identifying hemoperitoneum and splenic abnormalities, particularly in hemodynamically unstable patients, whereas contrast-enhanced CT remains the gold standard for definitive diagnosis and grading of splenic injury [4,8]. Rotational thromboelastometry (ROTEM) is an emerging, rapid bedside diagnostic modality in the management of spontaneous splenic rupture (SSR), especially for evaluating coagulopathy in patients with severe hemorrhagic shock [9].

Splenic rupture is frequently overlooked in the differential diagnosis of abdominal pain in the absence of trauma, and in most cases, splenectomy is required for treatment [10,11]. Mortality associated with this condition

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Table 1: Clinical examination findings at presentation

System/Parameter	*Findings*
General examination	Pallor present
Blood pressure	100/60 mmHg
Pulse rate	140/min (tachycardia)
Clinical impression	Findings suggestive of severe anemia with hemorrhagic shock
Abdominal examination	Tenderness in the right hypochondrium and suprapubic region
Guarding	Present on palpation
Per rectal examination	Normal

is mainly related to delays in diagnosis and management, as well as to risks associated with the clinical condition and severity of the underlying pathology [12,13].

In this case, we present a 13-year-old male child with spontaneous splenic rupture, a known case of congenital afibrinogenemia, effectively diagnosed using bedside POCUS and ROTEM, emphasizing their critical role in early diagnosis and emergency management.

Case Presentation

A 13-year-old boy with known congenital afibrinogenemia presented to the emergency department with sudden-onset abdominal pain for 1 day, associated with three episodes of vomiting and giddiness, followed by a fall. The patient had a history of recurrent mucosal bleeds in the past and had previously undergone multiple transfusions for the same. There was no past history of trauma, seizures, or loss of consciousness (LOC). He was diagnosed with congenital afibrinogenemia at the age of 1.5 years and had a history of multiple admissions in the recent past.

On clinical examination, the child was conscious, oriented, afebrile, and had no icterus. Pallor, low blood pressure (100/60 mmHg), and tachycardia (140/min) were suggestive of severe anemia with hemorrhagic shock. On abdominal examination, tenderness was present in the right hypochondrium and suprapubic region, with guarding elicited on palpation. Per rectal examination was normal [Table 1].

Laboratory investigations revealed severe anemia, with a hemoglobin level of 6.7 g/dL on admission, suggestive of acute blood loss. The total leukocyte count was 6,700 cells/mm^3. The platelet count was within the normal range (1.92 lakh/mm^3), implying that the bleeding tendency was primarily due to coagulation factor deficiency rather than thrombocytopenia. Liver function tests revealed an elevated AST level of 121 U/L, while ALT (23 U/L) and ALP (172 U/L) were within normal limits. Total protein and albumin levels were reduced (4.6 g/dL and 2.9 g/dL, respectively), likely associated with blood loss and acute illness. Blood urea was mildly elevated at 45 mg/dL, whereas serum creatinine remained within normal limits (0.7 mg/dL), indicating preserved renal function. Coagulation studies demonstrated significantly prolonged PT (>180 seconds) and APTT (>180 seconds), consistent with severe coagulopathy due to congenital afibrinogenemia. Viral markers including HIV, HBsAg, and HCV were negative, and the patient's blood group was O positive [Table 2].

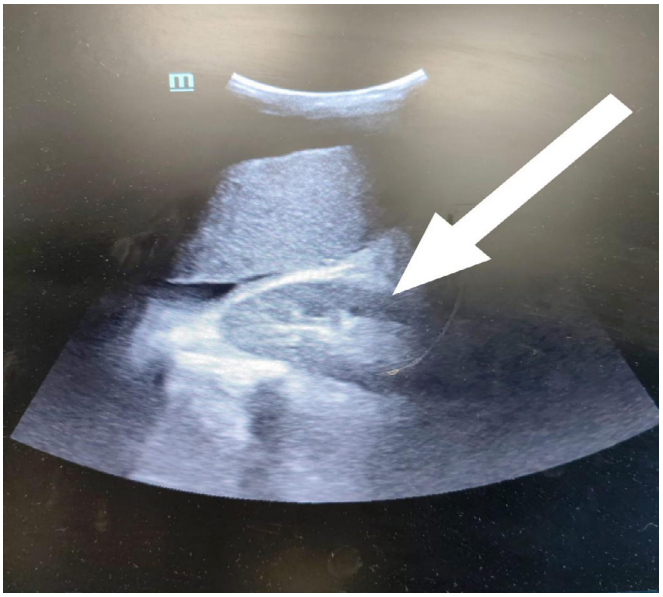


Figure 1: Bedside POCUS showing free intraperitoneal fluid

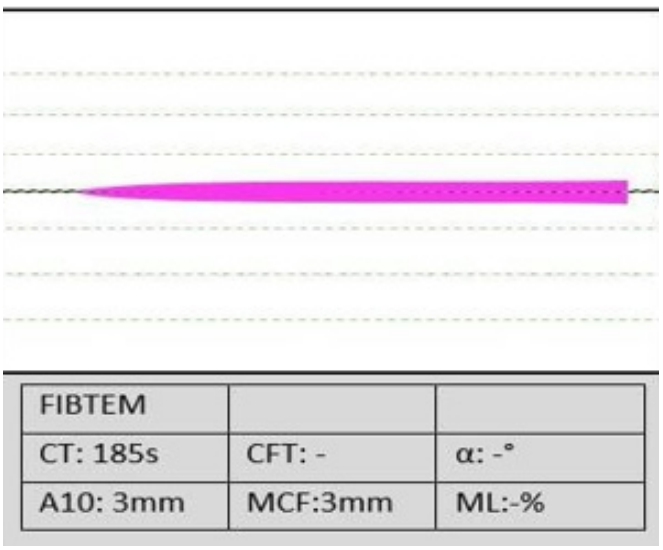


Figure 2: ROTEM-FIBTEM showing absent clot firmness

Table 2: Laboratory investigations

Investigations	*Patient value*	*Reference range*
Hemoglobin	6.7 g/dL	12-16 g/dL
Total Leukocyte Count (TC)	6,700 cells/mm^3	4,500-13,500 cells/mm^3
Platelet Count (PLT)	1.92 lakh/mm^3	1.5-4.5 lakh/mm^3
Random Blood Sugar (RBS)	111 mg/dL	70-140 mg/dL
Total Bilirubin (TB)	0.3 mg/dL	0.2-1.2 mg/dL
Direct Bilirubin (DB)	0.1 mg/dL	0-0.3 mg/dL
Aspartate Aminotransferase (AST)	121 U/L	10-40 U/L
Alanine aminotransferase (ALT)	23 U/L	10-45 U/L
Alkaline phosphatase (ALP)	172 U/L	130-340 U/L
Total protein (TP)	4.6 g/dL	6.0-8.0 g/dL
Albumin (ALB)	2.9 g/dL	3.5-5.0 g/dL
Blood urea	45 mg/dL	7-20 mg/dL
Serum creatinine	0.7 mg/dL	0.5-1.0 mg/dL
Serum sodium	140 mEq/L	135-145 mEq/L
Serum potassium	4.9 mEq/L	3.5-5.0 mEq/L
Prothrombin time (PT)	>180 seconds	11-15 seconds
Activated partial thromboplastin time (APTT)	>180 seconds	25-35 seconds
Human immunodeficiency virus (HIV)	Negative	-
Hepatitis B surface antigen (HBsAg)	Negative	-
Hepatitis C virus (HCV)	Negative	-

In our patient, bedside POCUS performed in the emergency department demonstrated free intraperitoneal fluid, raising suspicion of hemoperitoneum due to splenic rupture [Figure 1]. ROTEM analysis demonstrated absent clot firmness with a near-flat clotting curve, indicating severe functional coagulation impairment due to congenital afibrinogenemia [Figure 2].

CECT abdomen demonstrated an ill-defined hypodense collection measuring 6.1 × 4.9 × 6.6 cm in the spleen, along with moderate free fluid in the abdomen and pelvis (HU +43), indicating hemoperitoneum. Radiological findings revealed grade 4 splenic rupture with dense ascites and peritoneal thickening. Histopathological examination (HPE) revealed a congested spleen [Figure 3].

The patient was managed by resuscitation with intravenous fluids and fibrinogen concentrate. Emergency splenectomy was performed after achieving target ROTEM correction as the patient was hemodynamic instability. He had grade IV splenic injury, massive hemoperitoneum, and persistent bleeding despite correction. Blood was transfused to compensate for intraoperative blood loss and blood (nearly 2 liters) lost into the hemoperitoneum.

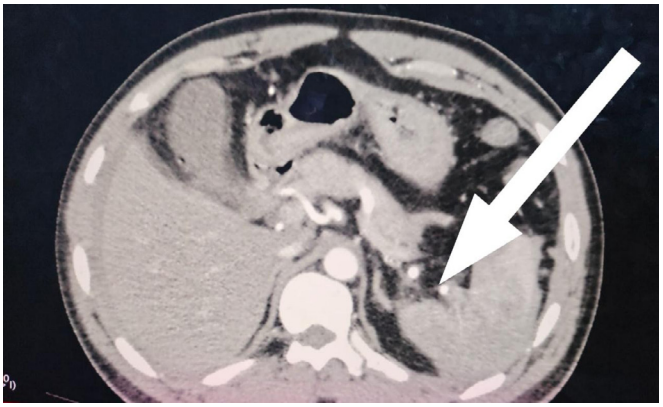


Figure 3: CECT abdomen demonstrating splenic hematoma with hemoperitoneum

Postoperative cardiology and hematology opinions were obtained for further management and monitoring of complications related to coagulopathy and hemodynamic instability. Cryoprecipitate was transfused intraoperatively and postoperatively to correct fibrinogen deficiency and ensure effective hemostasis. The patient developed thrombocytopenia due to postoperative consumption and transfusion-related changes, for which

Table 3: Medications prescribed after discharge

S. No.	*Medication*	*Dose*	*Frequency*	*Duration*	*Indication**
1	Vitamin B Complex with Vitamin C	1 tablet	Once daily	15 days	Vitamin supplementation
2	Paracetamol	500 mg	Twice daily	5 days	Fever/pain relief
3	omeprazole	20 mg	Twice daily	5 days	Gastric protection/acidity
4	Penicillin	400 mg	Twice daily	15 days	Antibacterial therapy
5	Aspirin	75 mg	Once daily	15 days	Antiplatelet therapy

tablet aspirin 75 mg once daily and low-molecular-weight heparin 0.4 mL subcutaneously were prescribed for 3 days.

On discharge, the patient was prescribed tablet B-complex with vitamin C once daily for 15 days, tablet Paracetamol 500 mg twice daily for 5 days, capsule omeprazole 20 mg twice daily for 5 days, tablet Penicillin 400 mg twice daily for 15 days, and tablet Aspirin 75 mg once daily for 15 days [Table 3].

Discussion

Congenital afibrinogenemia is an extremely rare, autosomal recessive bleeding disorder characterized by a near-complete absence of fibrinogen in the blood. It is reported to have an incidence of approximately 1 in 1 million individuals [14]. In patients with afibrinogenemia, umbilical bleeding, soft tissue and mucosal bleedings, menorrhagia, gingival bleedings, and bleeding in the mouth may be observed frequently [15,16]. Although bleedings may occur after trauma and surgery, spontaneous bleedings are rare [16].

A total of 25 cases of spontaneous splenic rupture have been described in patients with afibrinogenemia [17]. Spontaneous splenic rupture in congenital afibrinogenemia is a rare, life-threatening complication, often affecting children and teenagers [18]. In this case report, a 13-year-old boy presented with complaints of severe abdominal pain, vomiting, and giddiness. There is no past history of trauma, seizure, or LOC. Pallor, tachycardia, and hypotension are crucial clinical indicators of acute blood loss and evolving hemorrhagic shock in spontaneous splenic rupture [18]. Our patient presented with pallor, tachycardia (140/min), and low blood pressure (100/60 mmHg), which were consistent with the clinical manifestations of SSR. Severe anemia (Hb: 6.7 g/dL) along with significantly prolonged PT and APTT (>180 sec) indicated severe coagulation dysfunction secondary to congenital afibrinogenemia in our patient.

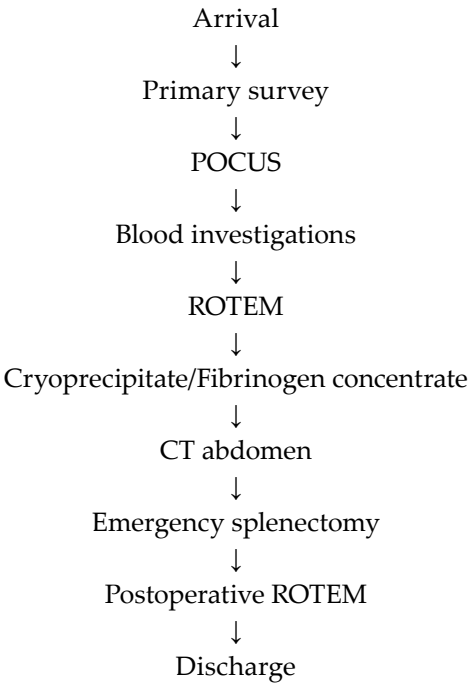
Anyfantakis et al. reported that SSR is often misdiagnosed because of its atypical clinical presentation and the absence of a preceding history of trauma. Patients may present with symptoms resembling other causes of acute abdomen, which can delay the diagnosis and management [19]. Our patient presented with abdominal pain, vomiting, giddiness, pallor, tachycardia, and guarding, which can be mistaken for many acute abdominal conditions. Due to the absence of trauma history, the diagnosis depends on increased clinical suspicion and rapid bedside evaluation.

According to Blaivas and Quinn, emergency POCUS is crucial in the early diagnosis of SSR, enable rapid bedside identification of intraperitoneal free fluid and splenic injury, specifically in unstable patients where quick decision-making is essential [20]. In our patient, POCUS performed in the emergency department demonstrated intraperitoneal free fluid with splenic abnormalities suggestive of SSR, which necessitated early stabilising measures and further confirmatory test. CECT is the preferred radiological modality in SSR, as it provides rapid and detailed evaluation of hemoperitoneum, splenic parenchymal injury, and active bleeding, consequently facilitating timely diagnosis and management [19]. In our patient, CECT revealed SSR associated hemoperitoneum, confirming the diagnosis suggested by POCUS.

Schöchl et al. stated ROTEM is a valuable point-of-care tool for rapid assessment of coagulation abnormalities. In SSR associated with coagulopathy, ROTEM can facilitate early detection of fibrinogen deficiency and guide timely blood product replacement [21]. In our patient, ROTEM testing demonstrated significant coagulation abnormalities suggestive of severe fibrinogen deficiency, which enabled rapid targeted blood product replacement and guided hemostatic management.

Previous literature by Renzulli et al. suggests effective management of atraumatic splenic rupture requires early diagnosis, resuscitation with blood product transfusion, correction of underlying clotting disorder, and early surgical intervention, with splenectomy

persisting as the most commonly performed treatment in hemodynamically unstable patients [22]. In our patient, resuscitation with intravenous fluids and fibrinogen concentrate was initiated, secondary to ROTEM-guided correction of coagulopathy. Emergency splenectomy was subsequently performed due to ongoing intra-abdominal hemorrhage with hemodynamic instability, in order to achieve curative hemorrhage control and prevent further clinical deterioration.



Timeline of Clinical Evaluation and Management

Despite its clinical significance, this case report has certain limitations. Plasma fibrinogen levels, Clauss fibrinogen assay, thrombin time, reptilase time, and genetic mutation analysis were not available. Detailed information regarding the emergency management could not be obtained. Further follow-up information was not available after discharge.

Future advancements in similar cases may include earlier diagnosis using advanced point-of-care imaging and coagulation monitoring, improved targeted fibrinogen replacement strategies, and emerging gene-based therapies for congenital afibrinogenemia.

Conclusion

In conclusion, this case is a rare presentation of spontaneous splenic rupture in a patient with congenital afibrinogenemia. Early POCUS aided immediate recognition of intra-abdominal bleeding in shock, while ROTEM enabled goal-directed transfusion and precise hemostatic therapy.

Integration of POCUS and ROTEM shortens the diagnosis time and improves outcome in rare bleeding disorders. This case highlights the importance of rapid multidisciplinary management and targeted hemostatic therapy in improving outcomes in life-threatening hemorrhagic emergencies.

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