Spontaneous splenic rupture in a patient with congenital afibrinogenemia

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Abstract

Afibrinogenemia is a rare bleeding disorder which is observed with an incidence of 1:1 000 000. 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. Basic clinical findings include spontaneous bleeding, bleeding after minor trauma or due to surgery. Splenic rupture in afibrinogenemia has been reported only in 6 cases so far. In this article, we present a 15-year old congenital afibrinogenemia patient with spontaneous splenic rupture.

(Türk Ped Arş 2014; 49: 247-9)

Key words: Spleen rupture, congenital afibrinogenemia, bleeding disorder

Introduction

Afibrinogenemia is a rare bleeding disorder which is observed with an incidence of approximately 1:1 000 000 (1, 2). It is an autosomal recessive disease (1, 2). There is consanguineous marriage between many parents (2). Afibrinogenemia occurs as a result of mutation in one of the three genes (FGA, FGB, FGG) which code the three polypeptide chains of fibrinogen on the fourth chromosome (2, 3). These mutations disrupt the synthesis, intracellular interactions, release or stability of fibrinogen (2). In these patients, hemorrhagic diathesis is present from the childhood and the clinical picture of hemorrhage may range between minor hemorrhage and life-threatening hemorrhage (1). We found spontaneous splenic rupture which developed in a patient with congenital fibrinogenemia. We presented this case, since a limited number of similar cases have been reported in the literature and we wanted to emphasize our treatment approach.

Case

A 15-year old female patient presented to our emergency outpatient department with complaints of abdominal pain and malaise. In her history it was learned that she was born from the fourth pregnancy of the parents who were third degree relatives as the fourth living child, oozing of blood occurred in the umbilical site after birth, she was hospitalized for one week because of bleeding in the mouth after falling from the armchair at the age of 7 months, she had occasional gingival hemorrhages and long-term bleedings after finger cuts. The patient who presented to the Pediatric Hematology Outpatient clinic because of unstoppable gingival bleeding for the first time in 2006 was diagnosed with congenital afibrinogenemia with findings including a prothrombin time (>120 s) and an activated partial thromboplastin time (>120 s) which were too long to be measured and a fibrinogen level which was too low to be measured. During the investigations, the mother was found to have a fibrinogen level of 113 mg/dL, the father was found to have a fibrinogen level of 83 mg/dL and her older brother was found to have a fibrinogen level of 83 mg/dL (heterozygous). Our patient had a fibrinogen level which was too low to be measured. It was also learned that the first child of the family was lost at the age of five days because of uncontrollable umbilical bleeding. The son...
of her paternal aunt was lost at the age of five years and the
daughter of her paternal uncle was lost at the age of 2 years
because of bleeding during the operation which was being
performed because of hip dislocation.

The patient presented to the pediatric emergency department
with complaints of abdominal pain, nausea and pain in the
left shoulder and leg which had been lasting for one week.
There was no history of trauma. On physical examination,
there was tenderness on abdominal palpation. There was no
defence and/or rebound tenderness. Examination of the sys-
tems was normal. Her laboratory tests were as follows: Hb: 9.1
g/dL, Hct: 28.1%, WBC: 12 200/mm³, platelets: 366 000/mm³,
erythrocyte sedimentation rate: 3 mm/h, CRP <0.34 mg/dL,
urea: 34 mg/dL, creatinine: 0.7 mg/dL, Na: 140 mmol/L, K:
4.4 mmol/L, Cl: 104 mmol/L, Ca: 8.6 mg/dL. On abdominal
ultrasonography, a structure with dimensions of 64x59 mm
compatible with abscess with a dense content was observed in
the spleen and free fluid was observed in the abdomen; ce-
fazolin, amikacin and ornidazole treatment was started with
a prediagnosis of splenic abscess, but a probable hemorrhage
could not be excluded, since she was being followed up with a
diagnosis of bleeding disorder. Two days later, severe abdom-
inal pain developed and abdominal tenderness, defence and
rebound were found on physical examination. Abdominal
imaging could not be performed, since her poor general sta-
tus continued. During this period, active bleeding continued
from the wound site and from the site of the catheter placed
before. In the follow-up, 15 units of erythrocyte suspension,
14 units fresh frozen plasma and 24 fibrinogen concentrates
(1 g) were given. The patient was discharged on the 29th
day of hospitalization.

Discussion

Fibrinogen transforms to fibrin with the effect of thrombin
and has an important role in formation of coagulum (2). In
addition, it is involved in primary hemostasis by binding
glycoprotein IIb/IIIa on the surface of activated platelets
and contributing to aggregation of platelets (2). In afibrino-
genemia, prothrombin time, thrombin time and activated
partial thromboplastin time are prolonged and there is no
fibrinogen in the blood (2). While thrombin-induced and
collagen-induced platelet aggregation are normal, platelet
adhesion and ADP(adenosin diphosphate)-induced platelet
aggregation are disrupted (2). In patients with afibrinogen-
emia, umbilical bleeding, soft tissue and mucosal bleedings,
menorrhagia, gingival bleedings and bleeding in the mouth
may be observed frequently (2, 3). Gastrointestinal and ur-
inary tract bleedings are observed less frequently. Intracranial
bleedings occur rarely (2). Although bleedings may occur af-
ster trauma and surgery, spontaneous bleedings are rare (3-5).

Splenic rupture is a rare finding in patients with a fibrinogen-
emia (6-8). 6 cases have been reported in the literature
until the present time (9). Splenic rupture is observed more
commonly in congenital afibrinogenemia compared to the
other congenital bleeding disorders. For example, although
hemophilia is a bleeding disorder which is observed with a
100-fold higher frequency compared to afibrinogenemia, the
number of cases with splenic rupture are similar (9).

Bleeding attacks in patients with afibrinogenemia are treat-
ed with fibrinogen concentrates, cryoprecipitates and fresh
frozen plasma (3, 10, 11). Fibrinogen concentrates are the
main options, because they are virally inactivated, they can
be infused with small volumes and have a low allergy risk
(2). Cryoprecipitate and fresh frozen plasma are the other
options, when fibrinogen concentrates are unavailable and only
in urgent conditions (10, 11). Bleedings in previous periods
were controlled with fresh frozen plasma in our patient. In
treatment guidelines, the targeted fibrinogen level in pa-

tients with afibrinogenemia is recommended to be >1 g/L
until hemostasis is provided and >0.5 g/L until the bleeding
surface heals (6). Many clinicians aim to reach high fibrino-
gen levels to provide hemostasis, but this treatment approach
increases the risk of thrombosis (2). It is known that patients
with afibrinogenemia have a tendency to thrombosis as well
as bleeding (2). In addition, use of antifibrinolytic drugs in
combination with concentrates used in treatment are a cause
of tendency to thrombosis. Therefore, one should be care-
ful clinically when monitoring these patients (10, 11). In our patient, we thought that respiratory distress which lasted for four days after splenectomy could be related with pulmonary emboly. However, respiratory distress regressed in four days and we did not find it necessary to perform imaging methods including computerized thoracic tomography and/or ventilation-perfusion scintigraphy. Fibrin glues can be used in cases of regional bleedings, dental interventions and superficial wounds (10). Tranexamic acid can be used in mucosal bleedings. It may be given intravenously, orally or as mouth wash (10, 11). In our patient, tranexamic acid was primarily given intravenously and then orally during supportive treatment for bleeding. In terms of inexpensiveness and convenience, tranexamic acid is efficient in reducing the number and frequency of factor requirement in severe bleedings rather than being used only for local bleeding control in patients with these types of rare factor deficiencies.

The first-line treatment is fibrinogen concentrates in patients with afibrinogenemia with splenic rupture. However, pictures of recurrent splenic rupture and following severe intra-abdominal bleeding, hypotension and shock have been reported in these patients in the literature. Therefore, splenectomy should be performed, although bleeding is controlled with medical treatment in patients with congenital afibrinogenemia with splenic rupture, since the risk of recurrence is high (8, 9). In our patient, we decided to perform splenectomy, although the bleeding was controlled with medical treatment on the 9th day. Regular use of fibrinogen concentrates has been reported after life-threatening bleedings and during pregnancy (2, 10, 11). It was planned to administer protective treatment to our patient once a week after discharge.

Conclusively, spontaneous splenic rupture can be observed in patients with afibrinogenemia. Splenic rupture should be considered in patients who present with a picture of abdominal pain, acute abdomen and hypotension/shock. In treatment, supportive treatment and splenectomy is performed.

Informed Consent: Written informed consent was obtained from patient’s parents.

Peer-review: Externally peer-reviewed.


Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

References