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Full Length Research Paper

Uncommon Presentation of Factor XIII Deficiency: Two Cases of Spontaneous Splenic Rupture in Young Patients

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Factor XIII deficiency is a rare bleeding disorder, and spontaneous spleen rapture is not reported in cases of this disorder. We report 2 cases of spontaneous ruptured spleen in 2 young Pakistani boys who were known factor XIII deficiency who were presented at the same time within 24 h. Both cases were recovered and one was successfully treated conservatively while the other treated surgically by spleenectomy. It should also remind physicians that bleeding disorders may have unusual presentations and should keep in mind ruptured spleen in the differential diagnosis of patient with factor XIII deficiency presenting with acute abdomen. The treatment is either surgery or conservatively depending on the progress of the bleeding.

Key words: Factor XIII deficiency, ruptured spleen, bleeding diathesis.

INTODUCTION

Congenital factor XIII (FXIII) deficiency is a rare autoso-mal recessive disease usually associated with a severe bleeding diathesis. The incidence is about 1 case per 2 - million populations. The male-to-female ratio is 1:1. FXIII is a plasma transglutaminase that catalyzes the final step in the coagulation cascade, cross-linking the loose fibrin polymer into a highly organized structure. In addition, FXIII covalently binds fibronectin, 2-plasmin in-hibitor, and other molecules to the fibrin plug; this en-hances adherence to the wound site, resistance to fi-brinolysis, and wound healing. Deficiency of factor XIII results in bleeding diasthesis. Physical manifestations related to bleeding may include the following: Persistent bleeding from the stump of the umbilical cord, soft tissue bleeding, neurologic findings commensurate with central nervous system hemorrhage, bleeding in the oral cavity, hemarthroses or periarticular bleeding, poor wound hea-ling The principal sites of bleeding are intracranial, um-bilical cord, superficial bruising and hematoma forma-tion, oral cavity (Ernest Beutler et al.,1994). Diagnosis is made by normal coagulation screening tests and a de-tailed family history. Specific factor XIII assays can confirm the diagnosis. The condition can also be defined by a clot solubility test. Because of the severity of the bleeding diathesis, prophylaxis is desirable and has been shown to be very effective as the *in vivo* half-life of plasma XIII is long, and low plasma levels are sufficient for haemostasis (Anwar et al., 2002).

Case 1

A.S is a 19-year -old patient presented in May, 2007 to A&E with severe abdominal pain, started all of a sudden at home when he was inside bathroom, the pain was agonizing, increasing in intensity, without history of neith-er constipation nor diarrhea, involving the whole abdo-men but mainly left hypochondia associated with difficulty of breathing and the patient was anxious. Also, he repor-ted left side basal pleuritic chest pain. He was denying hi-story of trauma. Urgent CT scan of the abdomen (Figure 1 showed subcapsular splenic tear and hematoma with intraabdominal collection, which was hemorrhagic. The patient was admitted to medical intensive care unit, managed conservatively by fresh frozen plas-ma (FFP) and tranexamic acid. No surgical intervention done. CT angio showed no bleeding but capillary blee-ding which is not feasible for arterial embolization. The patient received 10 - 20 ml/kg FFP q12H for first 5 days and also he was transfused 5 units PRBC. After five days from his admission, his hemoglobin became stable around 10 g/dL, FFP stopped and maintained on ferrous sulfate treatment, because of shortness of breath and ta-chypnea and chest pain, he was kept on IV Tazobactam and Piperacillin (Tazocin) on assumption of chest infec-tion by chest x-ray. Five days later, he had oxygen desaturation for which spiral CT chest showed

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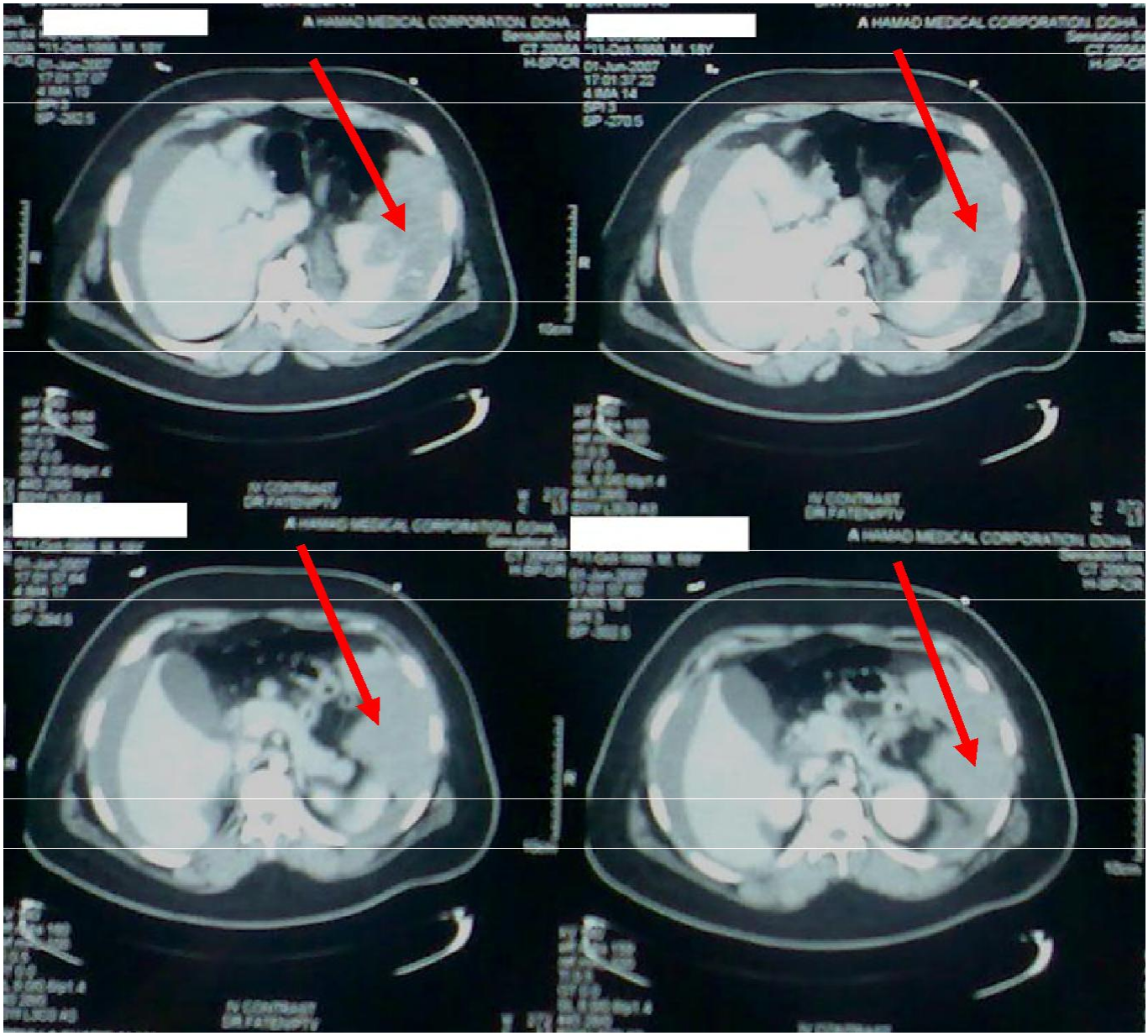

Figure 1. Case number 1, CT scan abdomen for case number 1 showing the ruptured spleen.

suspicious small pulmonary embolism in one of the left basal pulmo-nary arteriole. But the patient was not started on any treatment especially he was feeling much better, without pain, and no more shortness of breath.

He had been diagnosed as factor XIII deficiency, presented at 1-month-old with hematoma post vaccination and with later post circumcision bleeding then followed at Pediatric Hematology out patient department for frequent intra- articular and paraarticular soft tissue bleeding even after trivial trauma. Then after the age of 14, his care transferred to us as Adult Hematology Team. The patient continued to have same type of bleeding mainly involving the large joints like elbows, hips, and knees. In one admission in 2005, he had intra-abdominal bleeding with abdominal hematoma and one of other severe episodes; he had huge hemiarthrosis of the knee joint. During all these episodes, he was managed by fresh frozen plasma 10 - 20 ml/kg /day for two to three days in addition to IV or oral tranexamic acid 1 gm 8 hourly and given ferrous sulfate for his constantly low hemoglobin 100 - 120 g/L.

After the incident of knee joint hemiarthrosis early 2006 and at age of 18 years old, the patient was started on fresh frozen plasma prophylaxis 5 ml/kg every two to three weeks and for about one year, his condition was

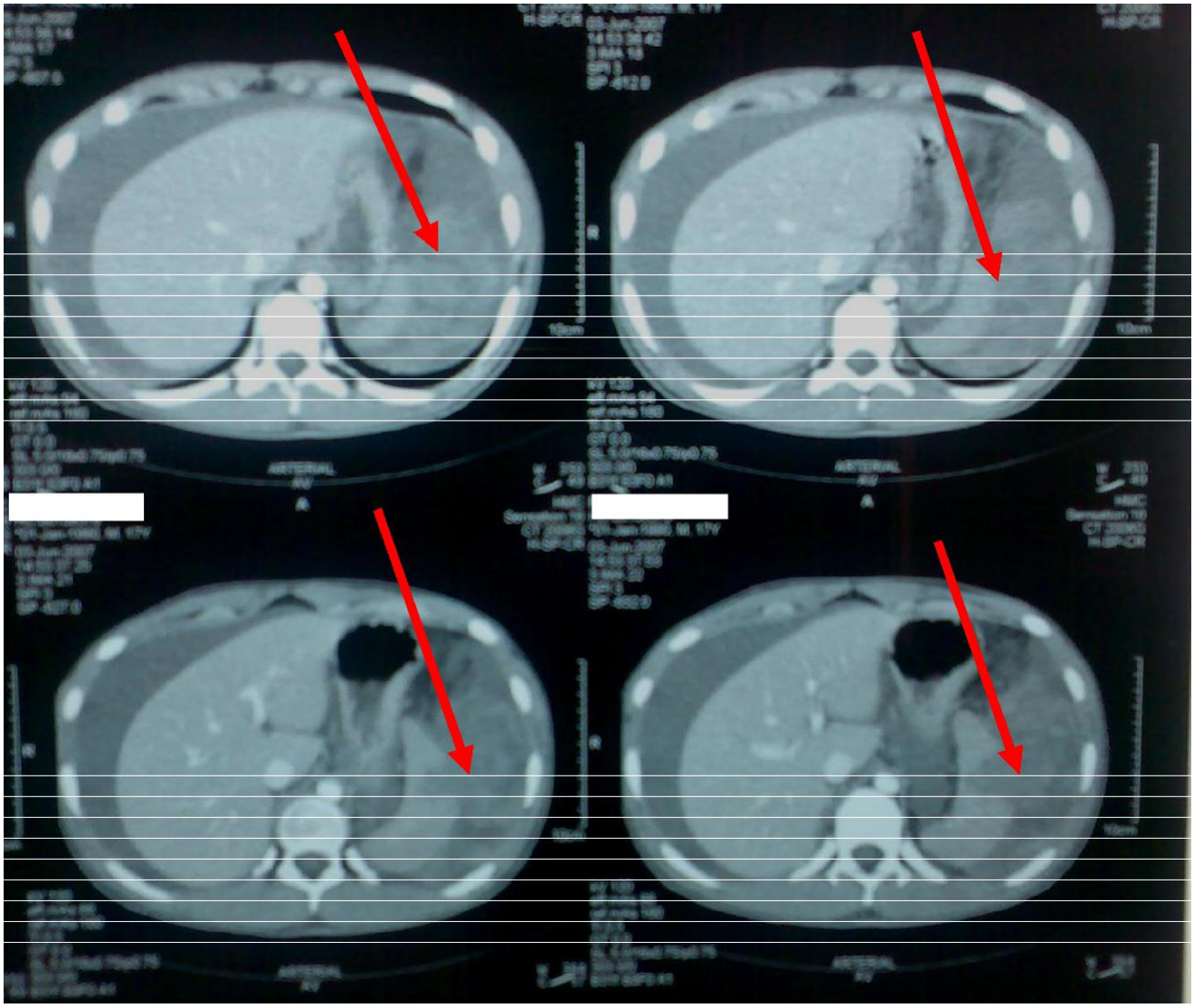

Figure 2. CT scan of abdomen for case number 2, showing the ruptured spleen.

very well without bleeding. His hemoglobin was trending to be more than 120 g/L but over the last two months, he stopped receiving FFP prophylaxis.

Social history: Nonsmoker nonalcoholic low to medium social class. Finished his secondary school two years ago and he is not employed. Not athletic.

Family history: The patient's father has no chronic illness. No bleeding tendencies. The mother has no bleeding tendency but history of migraine and convulsing disorder. The patient has two little brothers. The youngest has factor XIII deficiency and the other has no bleeding tendency so far and three sisters one older than the patient and 2 younger, none had bleeding tendency and screening test for factor XIII deficiency were negative.

Case 2

F.M.B a 17 years old Pakistani male patient had been diagnosed as factor XIII deficiency at age of 2 years when he developed bleeding after circumcision, he had history of repeated hematomas in the thigh, lips and bleeding after tooth extraction, presented on 3rd of June, 2007 presented to the emergency department with sudden onset severe abdominal pain of few hours duration, an urgent CT scan of the abdomen (Figure 2) revealed raptured spleen, patient received 2 unit PRBC and 5 units of FFP and underwent splenectomy, continued FFP after surgery until stablised. Post operatively patient developed shortness of breath and fever CXR showed bilateral infiltration, patient started on antibiotic and had smooth recovery.

In family history, his parents are first degree cousin he

had 2 sisters one of them diagnosed factor XIII deficiency and he had 3 brothers one died at age of 6 years from unknown reason.

DISCUSSION

Factor XIII (FXIII) deficiency is a rare inherited disease usually associated with a severe bleeding diathesis, with very low incidence which is about 1 case per 2 - 5 million populations and equal incidence in both male in female. Signs of bleeding include the following: CNS hemorrhage is frequent (25 - 30%) and may occur spontaneously or after minor trauma. Prevention of this complication is the major rationale for initiating prophylactic therapy. Infants are at risk of bleeding immediately after birth; the greatest risk is due to CNS hemorrhage. Soft tissue bleeding and bruising are very common, as is bleeding into the mouth and gums during teething. Hemarthroses occur in 20% of cases; however, incidence is less frequent than in severe hemophilia. Bleeding that is delayed (that is 12 - 36 h) after trauma or surgery is pathognomonic of FXIII deficiency. Recurrent spontaneous abortions are very common in women with FXIII deficiencies who do not receive FXIII replacement. Wound healing is abnormal in a small number of patients; therefore, FXIII may have a role in tissue repair (Waks et al., 1989; Anwar et al., 1999; Shaikh and Khurshid, 1993). We report 2 cases presented with spontaneously ruptured spleen in 2 young male Pakistani patients with in 24 h. There is high incidence of consanguinity was observed in affected families of factor XIII deficiency among Pakistanis (Board et al., 1993). It know that rupture of solid organ as liver , spleen kidney can occur after trauma but in a search of the Medline the occurrence of spontaneous rapture of spleen is not reported in the literatures.

Conclusion

Factor XIII deficiency is a rare bleeding disorder characterized by variable bleeding manifestations but consistent laboratory findings. We have reported an unusual clinical presentation in 2 patients with FXIII deficiency, which is ruptured spleen the treatment depended on the individual patient's situation whether to go for conservative line or surgical interference depending on the progressing of the bleeding needed awareness.

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