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A rare case of Factor XIII Deficiency and spontaneous rupture of the Spleen

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ABSTRACT

Introduction Factor XIII deficiency is an extremely rare occurrence & a patient presenting with spontaneous rupture of the spleen is challenging to diagnose and manage. Case Report We present here a case report of a patient diagnosed to have factor XIII deficiency since early childhood, presenting with acute onset abdominal pain with hemorrhagic shock at middle age. On evaluation, the Patient was found to have spontaneous splenic rupture which was managed by emergency splenectomy with adequate blood products transfusion. Conclusion Factor XIII plays a major role in the final step of the coagulation cascade in stabilizing fibrin clot & factor XIII deficiency is a very rare inherited or acquired disorder. Manifestations of factor XIII deficiency vary from being asymptomatic to life-threatening hemorrhagic conditions. Patients may have to be dependent on repeated specific blood product transfusions in order to avoid bleeding diathesis.

Keywords: *Factor XIII Deficiency, Spleen Rupture, Splenectomy*

1. INTRODUCTION

Coagulation Factor XIII (FXIII), also known as fibrin stabilizing factor, comes to play its role for stabilizing fibrin clot in final step of coagulation cascade. FXIII is a pro-enzyme & a tetrameric molecule made up of 2 alpha and 2 beta subunits linked by non-covalent bonds. [(1)] The activated FXIII acts on fibrinogen and plasmin inhibitors to make fibrin clot strong & stable. Such clot is resistant to degradation and helps in stopping further bleeding.

The half life of Factor XIII is long (11– 14 days) & hence small quantity (2–5%) is sufficient to prevent bleeding. Thus prophylatic therapy is very effective in these patients with known factor XIII deficiency.[(2)]

Genetic coagulation factor XIII deficiency is a rare inherited autosomal recessive disease, with an incidence of around one in two million.[(2)] Splenic rupture usually is associated with trauma (blunt or penetrating injuries) and imaging studies can show exact extent & grading of splenic damage. However, Spontaneous rupture of spleen can be easily confused with other abdominal pathologies like hematological & neoplastic diseases, tropical endemic disease or other causes of spontaneous rupture of the spleen. Spontaneous rupture of spleen is a rare but potentially life threatening condition which warrants immediate management.

Hereby we present the case of spontaneous rupture of spleen in a patient with hereditary factor XIII deficiency.

2. CASE REPORT

A 26 years young lady was brought to Emergency Room(ER) with complaints of diffuse abdominal pain, vomiting & fever of acute onset & 1 day duration. Patient had a significant past history in the form of umbilical stump bleeding at 3rd to 8th day of life & compartment syndrome due to large hematoma secondary to left forearm injury at 15 years of age. Patient was evaluated in CMC hospital, Vellore,India for excessive post partum haemorrhage and diagnosed to have Factor XIII deficiency responsible for recurrent bleeding manifestations. Patient had a positive family history of consanguineous marriage & elder brother was having similar recurrent bleeding manifestations. Patient had history of multiple times cryo-precipitates transfusion in the past. On arrival to ER, patient was found to be hemodynamically unstable & in hypotensive shock , blood pressure being 80/60 & Hemoglobin dropped from 10.1 gm/dl to 5gm/dl. BT(Bleeding time), CT(Clotting time), PT(Prothrombin time), INR(International normalized ratio) & APTT(Activated partial thromboplastin time) were found to be within normal limits.

Patient underwent FAST (Focussed Assessment Sonography of Trauma) abdomen which was suggestive of cavernous transformation of main portal vein, mildly enlarged spleen with heterogenous echotexture with ill-defined hetero-echoic lesion of 5.7cm*5.5cm in splenic region with minimal vascularity.

Patient underwent emergency Contrast Enhanced CT abdomen which was suggestive of perisplenic hematoma with splenic rupture with moderate ascitis. [Figure 1 and 2]

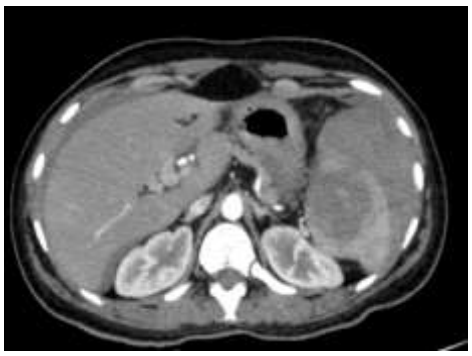

Fig. 1: Contrast enhanced CT abdomen showing perisplenic hematoma, splenic rupture and moderate ascitis

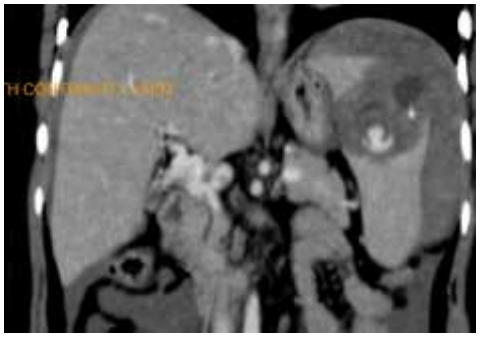

Figure 2. Coronal CT abdomen showing perisplenic hematoma with splenic rupture

Patient was diagnosed to have non-traumatic non-pathological splenic rupture with hemodynamic instability and hence patient was taken up for emergency laparotomy.

Intra-operatively 2 litre of hemoperitoneum with ruptured splenic hematoma was found.[Figure 3 and 4]
Intra-operatively patient was transfused 4 units of packed cells, 2 units of FFP (Fresh frozen plasma) & 12 units of cryoprecipitates in view of ongoing haemorrhage and coagulopathy.

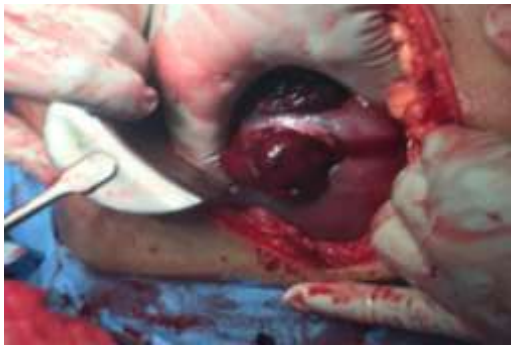

Fig. 3: Intraoperative picture of splenic rupture and blood clot

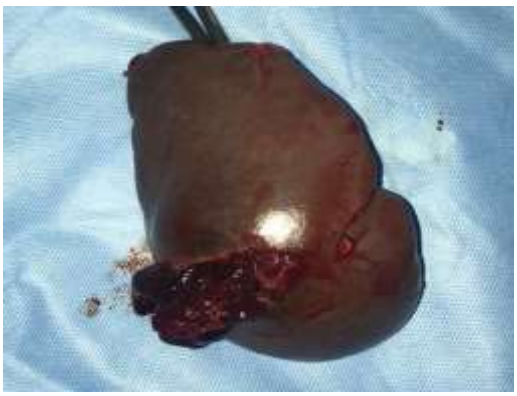

Fig. 4: Post-operative specimen of splenectomy.

Postoperative period was uneventful & patient was discharged 2 weeks after vaccination.
Pathological examination revealed no significant pathologic process in spleen.

3. DISCUSSION

One of the main constituent of the clot is fibrin which is stabilized by factor XIIIa that binds adjacent fibrin monomers & strengthens the clot. FactorXIII Val34Leu is a genetic determinant of fibrin structure/function.[3]

Previously spontaneous rupture has been reported in patients with afibrinogenemia proving that fibrinogen is an essential for splenic integrity & for preventing progression of minimal trival traumatic injury to disastrous consequences. There has only been one report in the English literature of a Factor XIII deficient patient who suffered from spontaneous splenic rupture.[4]

Non traumatic spontaneous rupture of spleen requires a high degree of clinical suspicion for immediate diagnosis, appropriate resuscitation and intervention.[5]

Orloff and Peskin suggested following criterias for diagnosing spontaneous rupture of spleen [6]

a. No history of trauma prior to operation or retrospectively after operation.
b. No evidence of disease that can affect spleen.
c. No evidence of perisplenic adhesions or scarring of spleen.
d. Spleen is normal on gross and histological examination.

Crate and Payne suggested fifth criterion that rise in viral antibody titres will not be seen in serum in acute & convalescent stage of the condition. [7]

The first cases of spontaneous splenic rupture were reported by Rokitansky et al in 1861 and Atkinson et al in 1874 [8]. Weidman in 1927 first used the term spontaneous rupture of spleen[8][9].

In 1966 Knoblich suggested that the term "spontaneous" should be replaced by "pathologic" in atraumatic rupture of the diseased spleen[9] Spontaneous rupture of apparently normal spleen or as a complication of infectious, inflammatory, hematologic, neoplastic causes have been reported.

The exact etio-pathogenesis of spontaneous rupture in normal spleen is not known but there are many theories without strong evidence to support them: [10]

1. Localized involvement of the spleen with a pathologic process, which upon rupture, all evidence of pathologic changes are destroyed.
2. Reflex spasm of splenic vein causing acute splenic congestion.
3. Portal venous congestion with chronic splenic congestion.
4. Abnormally mobile spleen that undergoes recurrent torsions and the resultant congestion leads to rupture.
5. Rupture of a degenerative or aneurysmal splenic artery.
6. Forgotten or unnoticed trauma
7. Inherited / Acquired coagulation factor deficiency.

Factor XIII deficiency is a very rare autosomal bleeding & Only 20% of patients experience bleeding in CNS, mouth and muscle or from lacerations and this occur usually without trauma [4]

Imaging can provide suspicion of immediate diagnosis but definitive diagnosis can be made on laparotomy only. Ultrasound can be confusing in the beginning as it happened in our case. Radiologists need to be aware of this possibility especially when confronted with background of hematological diseases. CT is a rapid diagnostic tool for reliable evaluation of the spleen.

There are no definitive guidelines for optimal management of spontaneous splenic rupture. In the scenario of splenic rupture secondary to blunt abdominal trauma, nonoperative management is recommended in hemodynamically stable by The Eastern Association for the Surgery of Trauma. Patient may need emergency laparotomy only in case of diffuse peritonitis or hemodynamic instability. [2]

FXIII deficiency confers a lifelong coagulopathy with a wide spectrum of severity. The approach to treatment is based on the fact that plasma levels of FXIII of >5% are sufficient to prevent bleeding and that the long in vivo half life of the factor XIII (11 to 14 d) makes it possible to infuse plasma cryoprecipitate or concentrates (all containing FXIII) at intervals of 1 month or longer.[2]
Spontaneous rupture of spleen can present with acute abdominal pain & clinical picture can vary from being stable to hemorrhagic shock. Clinically stable patient can be managed by non-operative conservative treatment. There has been a case report of hereditary factor XIII deficiency & spontaneous splenic rupture initially managed conservatively & at later age, patient developed recurrent splenic rupture which warranted emergency splenectomy in view of hemodynamic instability.

4. CONCLUSION

Factor XIII deficiency & spontaneous splenic rupture can be life threatening & requires high degree of clinical suspicion & low threshold for emergency laparotomy based on hemodynamic stability & radiological features.

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