Factor XIII Deficiency Presenting with Duodenal Hematoma in a 3-Year-Old Female: A Comprehensive Radiological Assessment

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Abstract

Key Clinical Message:

Factor XIII (FXIII) deficiency, though rare, should be considered in pediatric patients with unexplained abdominal pain, vomiting, anemia, and abnormal clotting. Prompt diagnosis enables non-surgical management with FXIII replacement, mitigating potential complications. Clinicians must include this condition in the differential diagnosis for effective patient care.

In the pediatric age group, FXIII deficiency can present as abnormal or delayed bleeding from the umbilical stump, soft tissue and subcutaneous bleeding, intracerebral hemorrhages, intra-oral bleeding, and poor wound healing. This case report describes an atypical presentation of FXIII deficiency in a 3-year-old female who presented to the emergency department with complaints of abdominal pain and vomiting. She was managed conservatively under the care of the pediatric surgery team.

[1] Introduction

Factor XIII (FXIII) deficiency is an autosomal recessive disorder characterized by a deficiency in FXIII, the final enzyme in the clotting pathway. FXIII is responsible for catalyzing the cross-linking between fibrin molecules, converting the fibrin polymer into a stable, organized, cross-linked structure that is relatively resistant to fibrinolysis [1-5].

In the pediatric age group, this deficiency can manifest as abnormal bleeding from the umbilical stump, soft tissue and subcutaneous bleeding, intracerebral hemorrhages, intraoral bleeding, and poor wound healing. This case report details a unique presentation of FXIII deficiency at our hospital, which was ultimately diagnosed as a large duodenal hematoma [2]. Duodenal hematomas are themselves rare, mostly caused by blunt abdominal trauma. Less frequently, they are associated with bleeding disorders, as in this case, or complications of anticoagulation therapy or endoscopy [6-9].

We present the case of a 3-year-old female who presented to the emergency room (ER) with abdominal pain and vomiting. This case report outlines the clinical presentation, radiological findings, and non-surgical management of the patient.

[2] Case History and Examination

A 3-year-old girl presented to our hospital with complaints of abdominal pain and vomiting. Upon examination, the patient was irritable yet active, and vitally stable. Systemic examination revealed no significant findings, with a soft and non-tender abdomen, absence of surgical scars, no visceromegaly, and normal bowel sounds. The child had no previous episodes similar to this.

[3] Methods (Differential Diagnoses, Investigations, and Treatment)

Laboratory investigations (**Table 1**) revealed decreased hemoglobin (Hb), leukocytosis, decreased mean corpuscular volume (MCV), low red blood cell (RBC) count, and platelets at the lower limit. The prothrombin time (PT) and activated partial thromboplastin time (APTT) were within the normal range.

The pediatric surgical team suspected intussusception initially and performed an ultrasound, which did not show signs of intussusception but revealed a rounded hypo to iso-echoic lobulated area adjacent to the porta hepatis and head of the pancreas, devoid of vascularity.

Further evaluation with cross-sectional imaging was recommended, leading to a computed tomography (CT) scan of the abdomen with contrast. The CT abdomen with contrast revealed a large intramural/submucosal duodenal hematoma along its entire length, causing luminal narrowing (**Figure 1**).

The radiology team suggested additional workup to rule out underlying hematological disorders or myeloproliferative disease. The coagulation profile showed a deficiency of FXIII (**Table 1**), which was determined to be the cause of the duodenal hematoma.

An esophagogram demonstrated a thin streak of contrast trickling into the proximal duodenum, suggestive of near-complete proximal duodenal obstruction (**Figure 2**). The patient was subsequently advised non-surgical management. Initially, she was kept nil per oral (NPO) and received intravenous hydration. A peripherally placed central venous catheter was placed under sedation after obtaining consent from the parents, and total parenteral nutrition (TPN) was initiated. The care plan was explained to the family, and the patient's hemodynamic parameters and output were closely monitored. Once stable, clear fluids were introduced, which the patient tolerated well.

[4] Conclusion and Results (Outcome and Follow-Up)

Upon request, the patient was discharged in a vitally stable condition with home care instructions. A follow-up appointment two weeks later showed no remarkable findings.

[5] Discussion

FXIII (also known as fibrin stabilizing factor) deficiency is one of the rarest factor deficiencies, occurring in approximately 1 in every 2 million people worldwide. Its function includes stabilizing blood clot formation,

aiding in tissue repair, depositing extracellular matrix, and contributing to the differentiation of osteoblasts [2]. The deficiency typically presents as excessive and prolonged bleeding from the umbilical stump, poor wound healing, and can lead to complications such as intracranial hemorrhage, either spontaneously or post-traumatically [10]. Additionally, superficial and deep hematomas are associated with the deficiency, although they are more commonly observed in older age groups [11].

Diagnosing FXIII deficiency can be challenging, as standard clotting tests such as PT, aPTT, and INR are usually normal. Key laboratory investigations for diagnosis include the clot solubility test, FXIII activity assay, FXIII antigen assay, inhibitor assay, and molecular diagnostics [2]. This case is unique because we believe it is the first reported instance of a duodenal hematoma due to FXIII deficiency. Duodenal hematomas are typically caused by crushing blunt force that ruptures intramural blood vessels or as a complication of anticoagulant therapy, endoscopic biopsy, vasculitis, pancreatitis, tumors, or bleeding disorders [6-9].

Early diagnosis of FXIII deficiency allows for conservative treatment, avoiding surgery [6, 8, 9, 12, 13]. Treatment options for FXIII deficiency include fresh frozen plasma (FFP) or cryoprecipitate. However, with advancements, FXIII concentrate and recombinant FXIII (rFXIII) are available for severe deficiencies to reduce bleeding events.

For duodenal hematomas, treatment can be conservative or invasive. Conservative management involves keeping the patient NPO for bowel rest and nasogastric (NG) decompression, placing a peripherally inserted central catheter (PICC), and initiating parenteral nutrition [14-17]. The duration of conservative management varies, and enteral nutrition can begin once gastric aspirates are no longer bilious.

Surgical treatment options include hematoma removal or bypass surgery [18]. In the past, when radiological advancements were not available, an exploratory laparotomy was performed after blunt abdominal trauma for evacuation, but this is now an option primarily in cases of jaundice and delayed bowel obstruction [19]. Invasive surgical procedures may still be necessary for complications such as uncontrolled bleeding or panperitonitis [20]. Recent advances include endoscopic drainage with fistula formation or mucosal puncture, which has shown success without major complications [21]. In cases of intramural duodenal hematomas (IMDH), percutaneous catheter insertion can be used for decompression [22].

[6] Conclusion

This case highlights the importance of swift clinical observation, thorough evaluation, and comprehensive radiological assessment in the management of duodenal hematoma, particularly in the pediatric age group. Medical practitioners should be vigilant of this rare condition and include it in their differential diagnosis for patients presenting with abdominal pain and vomiting, particularly those with a history of bleeding disorders. Early recognition and diagnosis can lead to timely and appropriate management, potentially avoiding complications and unnecessary surgical interventions.

Table 1: Results of the Laboratory Investigations

Investigation	Result	Normal Range
Hemoglobin (Hb)	7.3 g/dL	11.5 - 17.5 g/dL
Leukocyte count	Elevated	
Mean Corpuscular Volume (MCV)	72.4 fL	76 - 96 fL
Red Blood Cell (RBC) count	3.69 x 10^6/uL	4 - 6 x 10^6/uL
Platelets	150 x 10^9/L	150 - 400 x 10^9/L
Prothrombin Time (PT)	12.7 seconds	11 - 15 seconds
Activated Partial Thromboplastin Time (APTT)	27 seconds	25 - 40 seconds
Factor XIII	62%	70% - 140%

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