Abdominal Pain and Vomiting in an 11-Year-Old Boy with Factor VII Deficiency

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An 11-year-old Omani boy with congenital factor VII deficiency on demand factor VII replacement therapy. He was diagnosed with factor VII deficiencies since early life when he presented at the age of 14 months with a history of recurrent bruises and hemarthrosis, and he was found to have prolonged prothrombin time. Subsequently, the diagnosis was confirmed by the presence of a homozygous pathogenic variant in the F7 gene. He presented this time to the Emergency Department with abdominal pain for 4 days. The pain was centrally located with no radiation, colicky, and associated with bilious vomiting. He also reported constipation since the pain started without evidence of gastrointestinal bleeding. There was no history of fever, abdominal trauma, bleeding from any site, jaundice or skin pruritus. Upon examination, he was in pain, had mild pallor and hemodynamically stable. Cardiorespiratory and chest examinations were unremarkable. On abdominal examination there was tenderness in the epigastric and umbilical regions, with no rebound tenderness or organomegaly. Laboratory investigations revealed a hemoglobin level of 10.4 g/dL (reference range: 11.5-14), a platelet count of 506 x 10^9/L (reference range: 150-450), and a white blood cell count of 8.9 x 10^9/L (reference range: 4.5-12). The coagulation profile indicated a significantly prolonged prothrombin time (PT) (>180 seconds) (reference range: 10.3 - 12.1 seconds), elevated international normalized ratio (INR) (>20) (reference range: 0.9-1.2), normal activated partial thromboplastin time (APTT), and fibrinogen within the normal range. Additionally, C-reactive protein (CRP) was elevated at 18 mg/L (reference range: <5). Liver enzymes, amylase, and lipase levels were within the normal range.

Abdominal ultrasound done and reported as evidence of a hyperechoic mass in the region of the second part of the duodenum measuring approximately 6.4 x 2.7 cm. The mass projects into the lumen of the duodenum displacing the duodenal wall anteriorly with proximal gastric dilatation, rest of the scan was normal [Figure 1].

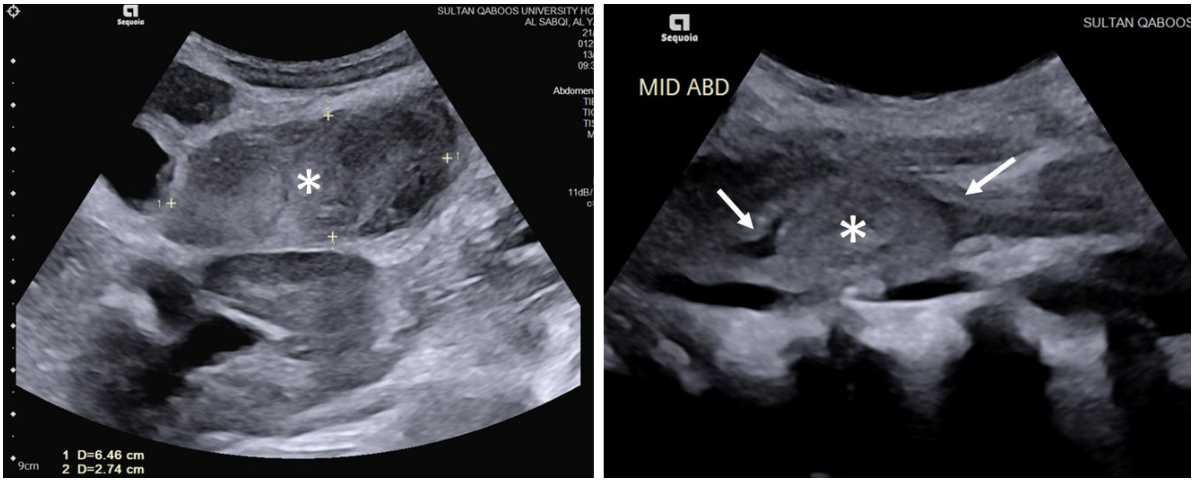

Figure 1: transverse and sagittal scan of upper abdomen showing hyperechoic mass (*) in the region of the second part of the duodenum. The mass projects in to the lumen of the duodenum displacing the duodenal wall anteriorly (white arrow).

Questions

1. What is the most likely diagnosis?

2. How would you confirm the diagnosis?

3. How would you manage this condition?

Answers

1. Intramural duodenal haematoma.

2. Computed tomography or magnetic resonant image of the abdomen.

3. To keep the patient nil per os with gastric decompression and consider total parental nutrition or naso-jejonal tube feeding.

Discussion

Congenital factor VII deficiency is a rare bleeding disorder characterized by decreased levels or activity of factor VII, a key component of the coagulation cascade. The clinical manifestations of this condition are heterogeneous, ranging from severe life-threatening haemorrhages, such as cerebral, gastrointestinal, and joint haemorrhages, to miscellaneous minor bleeding.[1] Patients with this condition are predisposed to bleeding episodes, but gastrointestinal involvement is not commonly reported. Here, we present a unique case of an adolescent with congenital factor VII deficiency exhibiting abdominal symptoms due to a duodenal mass.

The patient underwent a magnetic resonant image (MRI) of the abdomen which confirmed a lobulated intramural duodenal mass, extending from the second to the third part, causing significant luminal narrowing, consistent with a duodenal hematoma [Figure 2]. The patient was kept nil per os (NPO) and commenced on therapeutic intravenous dose of factor VII and total parenteral nutrition (TPN). The vomiting subsided and after 48hours, the patient was tried on fluid diet which he tolerated. Then the diet was upgraded gradually and TPN was ceased after few days.

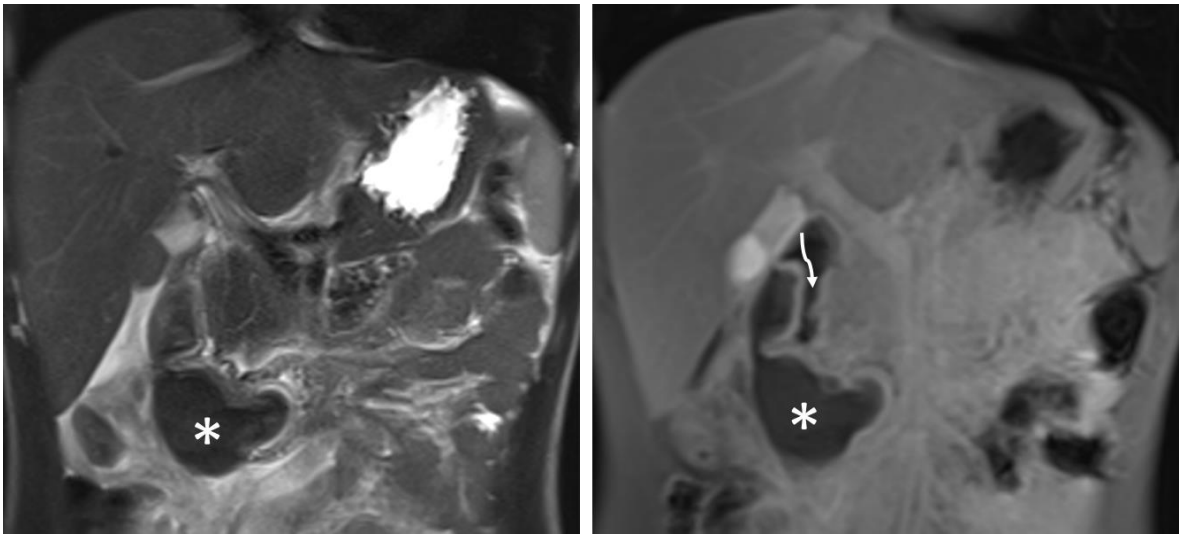

Figure 2: Coronal T2 weighted image with fat suppression and contrast enhanced coronal T1 weighted image with fat suppression show lobulated intramural duodenal mass (*) causing significant narrowing of the lumen (curved arrow). The mass has low T2 signal and shows no enhancement.

Intramural duodenal haematoma (IDH) is a rare condition in children. Blunt trauma to the abdomen is the most common aetiology, however, it has been reported to occur spontaneously in patients with coagulation disorders or those on anticoagulant treatment.[2] Iatrogenic causes like post upper gastrointestinal endoscopy and biopsy have also been reported especially in children post haemopiotic stem cell transplantation or with coagulopathy.[2,3] Although it is seen in patients with inherited coagulopathies, it is very uncommon.[4] Anatomically, the duodenum is divided into four parts, and it has no mesentery. The first part is located intraperitoneally, and the other parts are located retroperitoneally. Due to the retroperitoneal position of the duodenum, the lack of mesentery, and the close relation to the spine, in addition to the rich submucosal vascular supply, all these factors render the second and third parts of the duodenum vulnerable to intramural haematoma.[3] Which was the case with our patient.The clinical presentation of IDH may vary from mild, vague abdominal pain to complete gastroduodenal obstruction symptoms.[2] The patients usually present with abdominal pain, nausea, and vomiting. In children with bleeding diathesis and with little to tamponade the bleed, the IDH can reach a size large enough to cause partial or complete gastroduodenal obstruction.[4] These patients might also present with anaemia or acute obstructive pancreatitis secondary to the IDH.

Often the diagnosis is usually established after imaging and any modalities can be used to diagnose and monitor IDH. Computed tomography (CT) abdomen is considered the gold standard for the initial evaluation of the haematoma and related complications like intestinal obstruction, bowel perforation, and obstructive pancreatitis.[2,4,5] Ultrasound is increasingly used for follow-up. In this patient the ultrasound hinted to the diagnosis and was helpful in monitoring the haematoma, while the CT confirmed the diagnosis and excluded associated complications like pancreatic injury or hematoma.

IDH is managed conservatively with bowel rest and gastric decompression, pain management, and early initiation of TPN. Surgical intervention and percutaneous drainage are usually reserved for patients who fail conservative management and remain symptomatic for more than a week. However, these interventions can cause great trauma for the patient.[6] Ultrasound/ CT-guided drainage or endoscopic incision and drainage were reported to be successful.[,3,4,5] The endoscopic technique consists of an incision of the hematoma using a needle-knife or a biopsy forcep, which will result in rapid submucosal decompression. Other techniques involve using hot AXIOS (lumen-apposing stent).[6] Additionally, in patients with bleeding disorders, it is essential to manage the underlying coagulopathy.[4] This patient had factor VII deficiency, so he was treated with therapeutic dose of recombinant factor VII 30 mcg/kg/dose every 3 hours until hemostasis is achieved.

Although rare, spontaneous intramural duodenal haematomas can present as a gastroduodenal obstruction in patients with inherited coagulopathies. The diagnosis requires a high index of suspicions. Most cases can be managed conservatively with bowel rest, TPN and pain management. In addition to correction of the underlying coagulopathy.

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