Bedside Sonographic Diagnosis of Multiple Intussusceptions as a Primary Manifestation of Peutz-Jeghers Syndrome

Authors

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Abstract

Peutz-Jeghers syndrome (PJS) is a rare, autosomal- dominant hereditary disorder characterized by mucocutaneous pigmentation and gastrointestinal hamartomatous polyps. Our case report demonstrates how ultrasound aided in the timely diagnosis of multiple intussusceptions in a young female patient not previously known to have PJS. The importance of ultrasound in making an accurate early diagnosis at bedside, is emphasized.

Case presentation:

A 27-year-old female of Asian descent presented to the emergency department with a history of acute abdominal pain of 1-day duration. Her pain was colicky in nature, coming in bouts and localized mainly to the epigastric and central abdomen. The pain was associated with nausea but no vomiting. She had pain- free intervals between the attacks. There were no changes in her bowel and bladder habits. On examination, the patient was hemodynamically stable but looked tired and was in considerable pain. Systemic examination was remarkable for hyperpigmented spots over the lower lip & buccal mucosa. Abdomen was soft with significant tenderness in the epigastrium. No masses or organomegaly were appreciated.

Bedside ultrasound was performed by an emergency physician looking for free fluid and possible ectopic pregnancy. The hepatorenal and splenorenal spaces were scanned first and showed no free fluid. The suprapubic view revealed a bulky uterus with no intrauterine or extrauterine gestational sacs (image 4). After the urine pregnancy test was negative, a more focused bedside abdominal scan revealed multiple hyperechoic round donut-shaped lesions seen in the right iliac fossa, periumbilical and left hypochondriac areas (image 1-3).

There was an evident peristaltic movement seen in some of the masses, surrounded by large dilated loops of small bowel. These findings were suggestive of multiple intussusceptions.

The laboratory investigations showed mild leukocytosis. Other laboratory investigations were within normal limits including amylase and lactate. A contrast enhanced computed topography (CT) of the abdomen and pelvis was obtained which confirmed the diagnosis of three intussusceptions; proximal jejunum, distal jejunum, and the ileocecal junction (image 5,6). The CT also confirmed presence of adenomatous polyps and the absence of any free fluid (image 7).

The patient was admitted under the care of the General Surgery team where she remained under conservative treatment with no oral intake and was started on an intravenous drip of Hyoscine. Over the next 48 hours, her symptoms gradually subsided without any clinical or biochemical deterioration. She was advised about the need for definitive surgical intervention, which she opted to have in her home country.

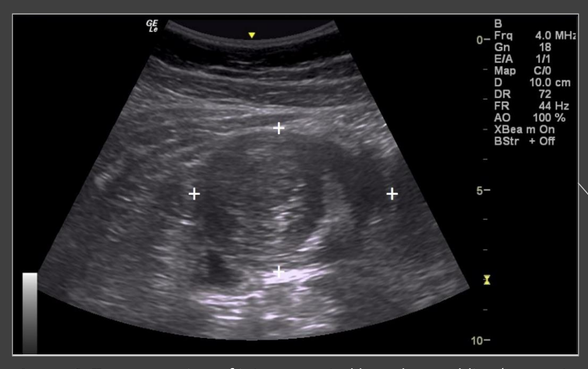

Image 1. Transverse view of intussuscepted bowel resembling 'crescent inside a donut" seen in the left upper quadrant of abdomen.

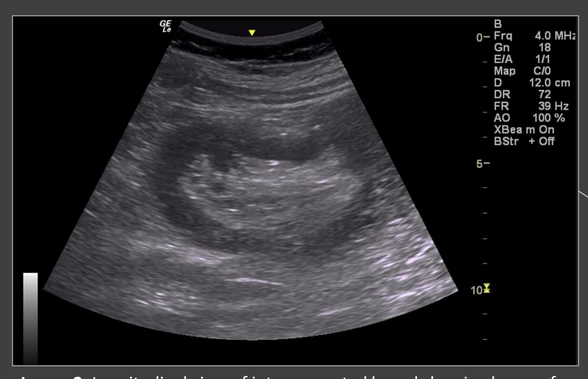

Image 2. Longitudinal view of intussuscepted bowel showing layers of thickened bowel walls. A peristaltic movement was seen in the intussucepted loop.

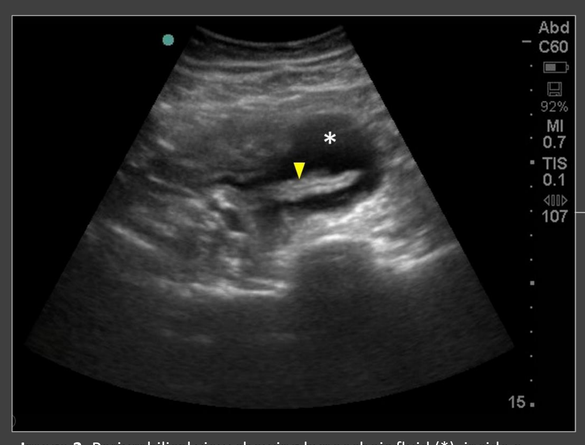

Image 3. Periumbilical view showing hypoechoic fluid (*) inside a bowel loop. This can easily be mistaken for free intraperitoneal fluid if the scan is not complete. The folded double bowel wall is seen in the middle (yellow indicator).

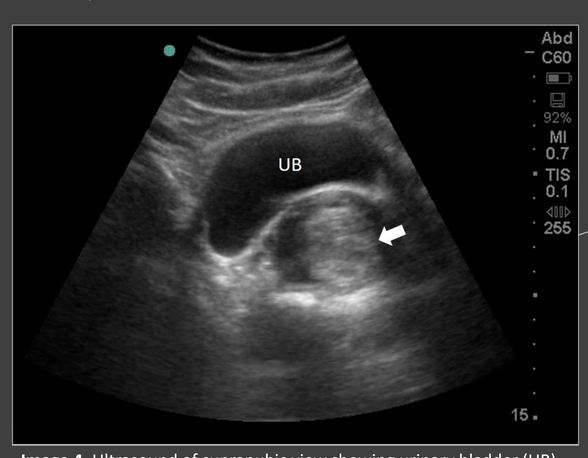

Image 4. Ultrasound of suprapubic view showing urinary bladder (UB) being pushed posteriorly by a round heterogeneous mass most likely to be a uterine leiomyoma (arrow).

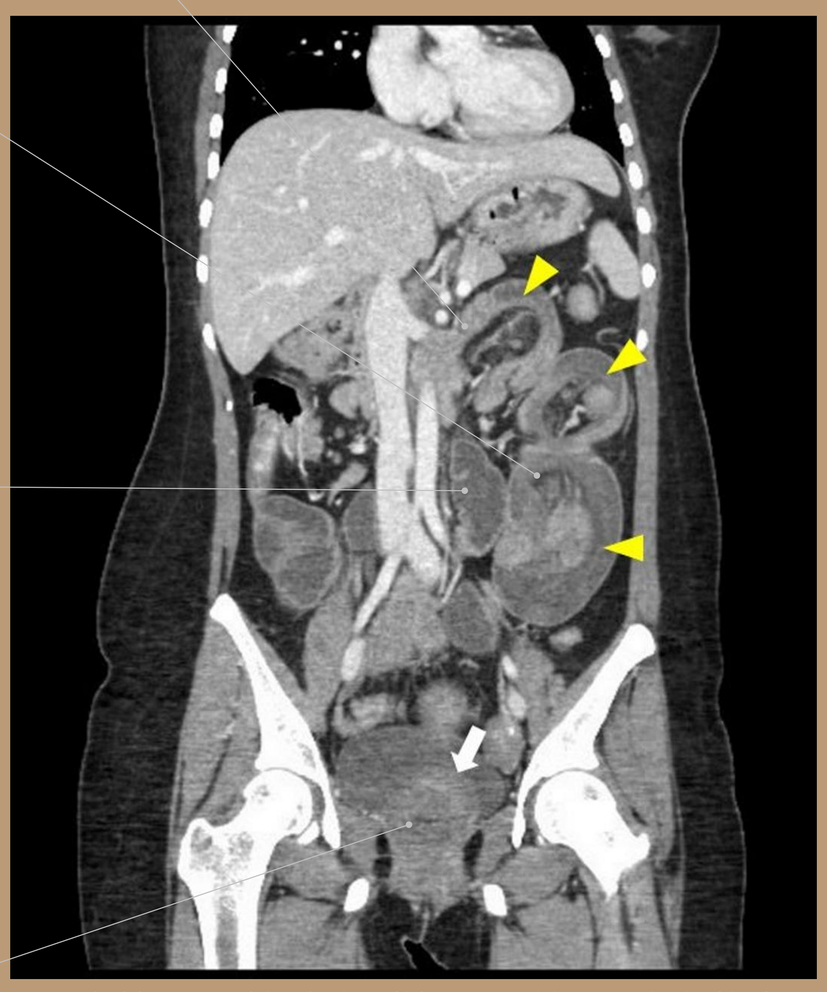

Image 5. CT abdomen, coronal view, showing multiple entero-enteric intussusceptions involving the proximal, mid and distal parts of jejunum telescoping in each other along with the mesentery as well as mesenteric fat and vessels (yellow indicators). The intussusceptions Bulky irregular uterus is seen (white arrow).

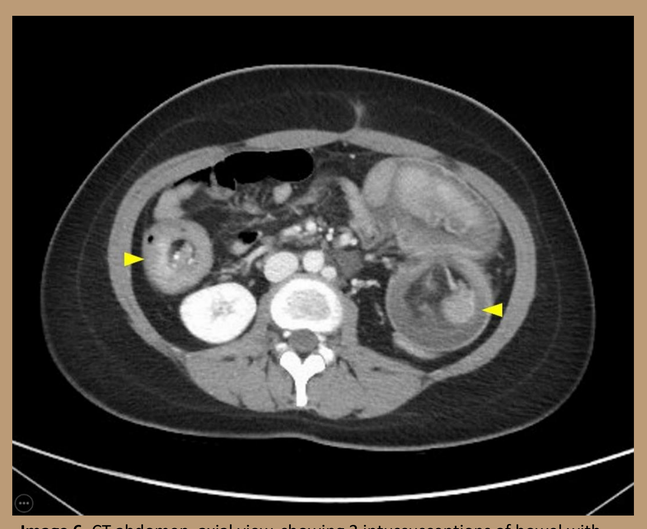

Image 6. CT abdomen, axial view, showing 2 intussusceptions of bowel with edematous walls (yellow indicators).

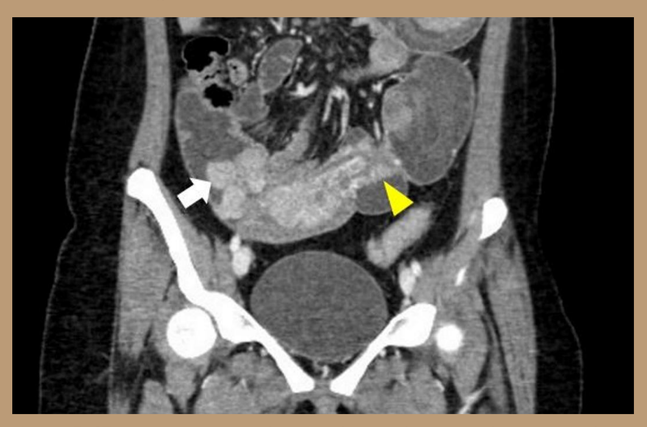

Image 7. CT abdomen, coronal view showing multiple polyps in the ascending colon (white arrow) and a segment of ileocecal intussusception with terminal ileal loops seen telescoping in the ascending colon (yellow indicator). The cecum is distended.

Discussion

Peutz-Jeghers syndrome (PJS) is an autosomal dominant hereditary disorder characterised by intestinal haemartomatous polyps in association with a distinct pattern of skin and mucosal macular melanin deposition (1). It is considered a rare disorder, with an incidence of 1 case per 60,000 to 300,000 people. The median time at which the first presentation of polyps occurs is at around 11-13 years of age. Around 50% of individuals have experienced symptoms by 20 years of age The overall incidence of intussusception in adults is 1 in 30,000 (2). Patients with PJS have a considerably higher incidence of intussusception owing to the presence of polyps which act as lead points. Lier et al reported an overall incidence of intussusception in PJS to be as high as 69% (3). Less than 1% of intussusceptions in PJS occur as multiple (4). Only a handful of cases of multiple intussusceptions in PJS have been reported. (5-9)

The role of ultrasound is well established in the evaluation of suspected intussusception, with published sensitivity and specificity rates of 98–100% and 88–100%, respectively (10). Bedside ultrasound performed by emergency physicians has similar sensitivity and specificity rates of 85% and 97%, respectively (11). A transverse view of the intussuscepted bowel appears as a swirled mass (usually > 5cm in size) of alternating echogenecities representing the echogenic mucosa, hypoechoic submucosa and echogenic muscularis layers of the intestinal wall. This finding has been named the 'target sign', 'crescent in a doughnut sign' and 'concentric ring sign' (image 1)(12, 13). On a longitudinal view, the loop-within-loop of bowel resembles a layered sandwich, described in the literature as a 'pseudokidney sign' (image 2)(13).

Additional findings of free intraperitoneal fluid or air may be seen if perforation has occurred. Bedside ultrasound plays an important role in ruling out free fluid in non-traumatic acute abdomen (23). Finally, bedside ultrasound is of particular value in excluding other differentials of abdominal pain such as ectopic pregnancy.

Abdominal computed tomography (CT) has a reported diagnostic accuracy of 58%-100% (14). The characteristic features of CT scan include an inhomogeneous 'target' or 'sausage'-shaped soft- tissue mass with multiple layers. Often, mesenteric vessels within the bowel lumen are visible. The main advantages of CT include the ability to identify the location as well as the underlying pathology of the intussusception. In addition to CT, intraoperative enteroscopy has been recommended, as it allows for identification of polyps that would have been missed on CT. Most of the intussusceptions in PJS require surgical intervention, however, laparoscopic-assisted enteroscopy may offer a less invasive approach for polyp removal.

Conclusion:

Ultrasound is a non-invasive diagnostic tool that can aid in rapidly making a bedside diagnosis of intussusception and ruling out other acute intra-abdominal pathologies. Patients with Peutz-Jeghers syndrome commonly suffer from intussusceptions however multiple intussusceptions are extremely rare.

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