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An Atypical Peutz-Jeghers Syndrome with Intussusception Presenting as an Acute Intestinal Obstruction in an Adolescent: A Case Report

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Abstract | **Case Report**

Peutz-Jeghers syndrome is a relatively rare inherited autosomal dominant hereditary cancer syndrome characterized by benign hamartomatous gastrointestinal polyps, mucocutaneous pigmentation and a high predisposition to many intestinal and extraintestinal cancers. It can be followed by variety of serious complications such as bleeding, obstruction, intussusception and malignant transformation. We report a case of a 16-year-old adolescent who presented with a 3-day history of abdominal pain and history of bleeding per rectum with no pertinent family history.

Keywords: Peutz-Jeghers syndrome, Intussusception, Hamartomatous polyps, Mucocutaneous pigmentation.

1. INTRODUCTION

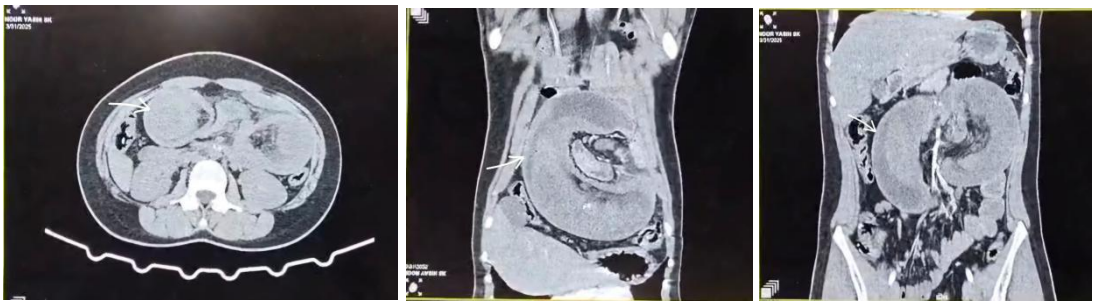
Peutz-Jeghers syndrome is a relatively rare inherited autosomal dominant hereditary cancer syndrome with mutation of the STK11/LKB1 gene located on chromosome 19p. It is characterized by benign hamartomatous gastrointestinal polyps, mucocutaneous pigmentation and a high predisposition to many intestinal and extraintestinal cancers. It can be followed by variety of serious complications such as bleeding, obstruction, intussusception and malignant transformation. We report the case of a 16-year-old male who presented with acute onset of abdominal pain, history of bleeding per rectum and no pertinent family history with evidence of jejuno-jejunal intussusception on computed tomography (CT) imaging and confirmation of Peutz-Jeghers hamartomatous polys on tissue specimen which was the lead point for intussusception. This case report has been reported according to the revised SCARE guidelines, 2023[18].

2. CASE PRESENTATION

A 16-year-old male presented to the emergency department of our hospital with acute onset abdominal pain of 3 days duration, pain is generalized and associated with nausea and vomiting with loss of appetite. He had a history of recurrent abdominal pain and two episodes of bleeding per rectum with no previous medical or surgical history. He was otherwise healthy and fit. The patient was hemodynamically stable. On examination patient was dehydrated with tenderness present throughout the abdomen and abdominal lump was palpable in the umbilical region all around. Bowel sounds were exaggerated. Per rectal examination revealed empty rectal vault. Blood investigations showed high normal white blood cell (WBC) count and other investigations were within normal range. Ultrasound of abdomen (USG) demonstrated features of intestinal obstruction. A CT scan was done on emergency basis and it showed dilated proximal jejunum with bowel-within-bowel configuration suggesting Jejuno-jejunal intussusception and intestinal obstruction (Figure 1). Patient was examined for mucocutaneous pigmentations where none can be seen.

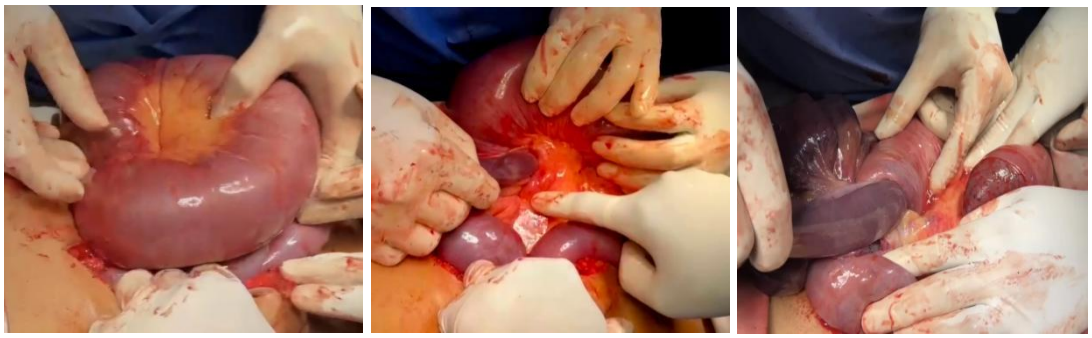
On Emergency exploratory laparotomy for intestinal obstruction after adequate resuscitation. It revealed a large Jejunojejunal intussusception 20cm distal to Duodenojejunal junction with proximal jejunum being engulfed into distal part. Complete manual reduction of intussusception was done revealing about 120 cm of congested bowel which was part of intussusception. The intussuscepted bowel was deemed nonviable and resection of the entire congested bowel with end-to-end anastomosis of the healthy bowel was done (Figure 3). During resection at the proximal end

there were masses palpable intraluminally, on laying it open there were three pedunculated polyps which were the lead point for intussusception (Figure 3). Rest of the mucosa was normal. The entire specimen was sent for histopathological examination.



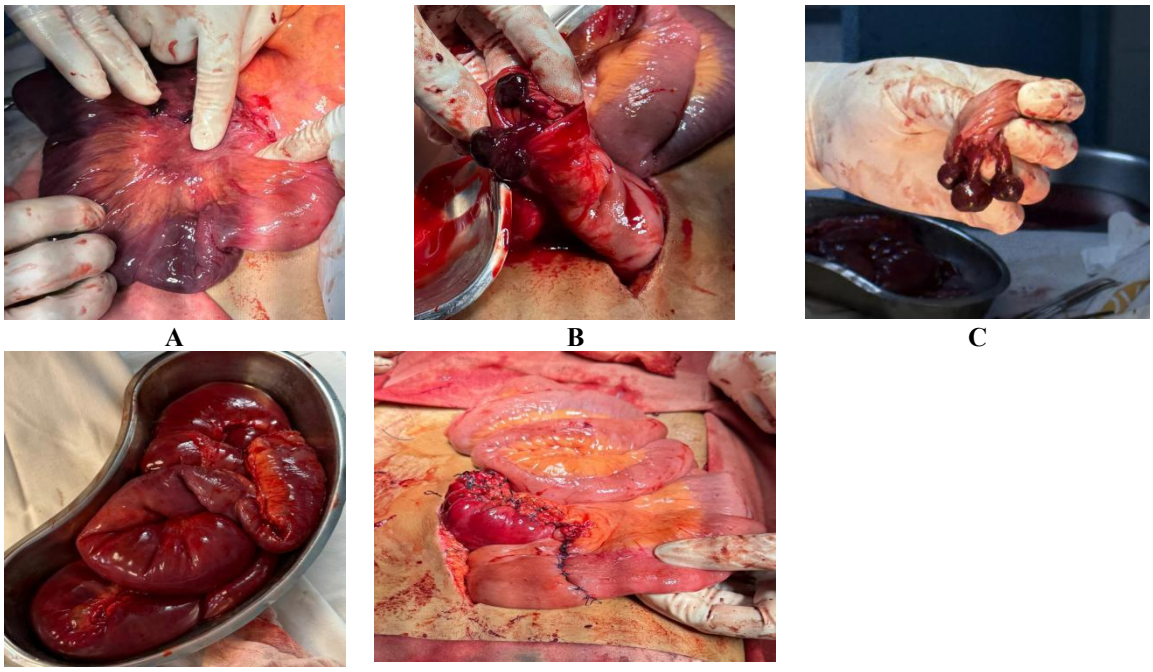
(A) (B) (C)

Figure 1: CT scan of abdomen: (A)-axial view, (B), (C)-coronal view, all showing dilated jejunum(arrows) on both sides of abdomen with bowel within bowel configuration [Target sign - in (A) and Pseudokidney sign in (C)] and interbowel fluid



(a) (b) (c)

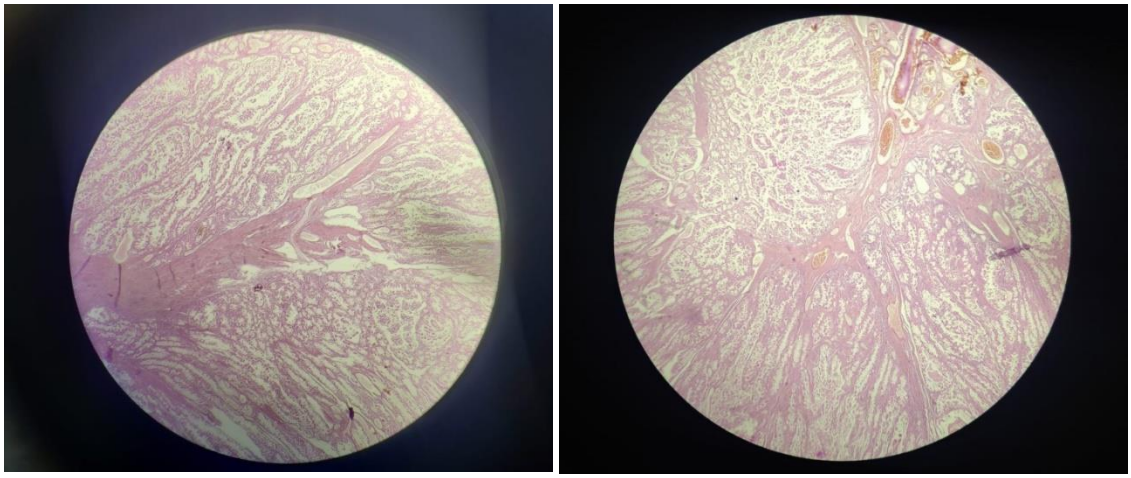
Figure 2: (a)- Intraoperative picture of large jejunojejunal intussusception 20cm from duodenojejunal junction. (b), (c)- Manual reduction of intussusception



A B C

D E

Figure 3: (a) - Transition area between congested gangrenous bowel and normal bowel (b), (c) - Three pedunculated polyps at the proximal end of congested bowel largest measuring 2.5*1.0*0.5cm. (d)- Resected congested gangrenous bowel (about 120cm in length). (e)- End to end Anastomosis of the healthy bowel.



(a) **(b)**

Figure 4: Histopathological examination demonstrated arborizing smooth muscle bundles extending from the muscularis mucosa into the polyp core, producing the characteristic tree-like architecture of Peutz–Jeghers hamartomatous polyps.

Post operatively the patient was hospitalized for 1 week during which he had episodes of passing black colored stools which eventually reduced. Other post op events were uneventful and the patient was discharged on seventh postop day. Patient was on regular follow up with gastroenterology clinic. After one month postoperatively patient was advised upper gastrointestinal endoscopy and colonoscopy. These showed no evidence of any polys in stomach, duodenum, colon and distal small bowel. Patient was kept on six monthly follow up. Patient family members were also advised regarding cancer risks in the family and were advised for followup.

3. DISCUSSION

Peutz-Jeghers syndrome was first described in literature in 1895 by Dr. Connor,a British physician who reported identical twin sister's with oral pigmentation. One sister died due to intestinal obstruction age 20 years while the other died due to breast cancer aged 59 years. Later the primary description of PJS was published by Peutz in 1921 in one Dutch family as a gastrointestinal familial polyposis with pigmentations. Jeghers specified the relations between pigmented lesions, gastrointestinal polyposis and increased risk of carcinoma in 1949[1]. In 1954, Dr. Bruwer of the Mayo Clinic named the syndrome as “Peutz-Jeghers syndrome” [2].

Peutz-Jeghers syndrome (PJS) is a relatively rare inherited autosomal dominant hereditary cancer syndrome with mutation in the STK11/LKB1 gene located on chromosome 19p13.3[4]. As many as 50% of affected individuals are new cases and have no family history of the disease as a result of spontaneous mutation. It is seen equally in men and women and diagnosed mostly during childhood to early adulthood. It has a reported an estimated incidence of 1:50,000 to 1:2,00,000[3]. It is characterized by benign hamartomatous gastrointestinal polyps (90%), mucocutaneous pigmentation (95%) and a high predisposition to many intestinal and extra intestinal cancers (90%). Abdominal discomfort and distention are the most common presenting symptoms. One-third of PJS patients become symptomatic during the first decade of life and nearly half experience symptoms that require an operation for bowel obstruction by the age of 20 years. It can be followed by variety of serious complications such as bleeding, obstruction, intussusception and malignant transformation. It is part of the hamartomatous polyp syndromes that also include juvenile polyposis syndrome (JPS), and *PTEN* hamartoma tumor syndrome (PHTS), which includes Cowden syndrome (CS) and Bannayan-Riley-Ruvalcaba syndrome (BRRS).

PJS is a clinical diagnosis based on any one of the following World Health Organization criteria (WH0):

1. three or more histologically confirmed Peutz-Jeghers polyps
2. Any number of PJ polyps with a family history of PJS
3. characteristic, prominent, mucocutaneous pigmentation with a family history of PJS
4. any number of Peutz-Jeghers polyps and characteristic prominent, mucocutaneous pigmentation [6].

The polyps can arise anywhere in the GI tract, but the small intestine (75%) is the most common site (jejunum-90%, ileum-80%) [3] followed by colon (42%), stomach (38%), and rectum (28%). Extra intestinal polyps, though rare, have also been reported in gallbladder, ureter, tracheobronchial tree and tonsils [5]. Peutz-Jeghers polyps typically have an abnormal muscularis mucosa branching into the lamina propria, giving it the appearance of a Christmas tree [7] and do not have dilated cystic-filled spaces pathognomonic for juvenile polyps.

The other characteristic finding in PJS is the mucocutaneous pigmentations seen in over 95% of

patients are due to melanin deposition. They are seen over lips and perioral region (94%), hands (74%), feet (62%), perianal and genital region [5]. The macules increase in intensity from infancy and early childhood but often fade in adult life and completely disappear in some cases. Pigmented spots on the buccal mucosa are present in over two-thirds of individuals, but unlike the skin macules, the buccal lesions usually persist through life.

Bleeding, anemia, intussusception, and intestinal obstruction are common complications associated with hamartomatous polyps [8]. The usual age of presentation following intussusception-related symptoms is in late childhood to early adulthood. Intussusception is a process that results from invagination of a portion of proximal bowel (intussusceptum) into another more distal bowel(intussuscipiens). Clinical presentations vary according to the site of intussusception, the timing of the presentation, and the possibility of self-reduction. Some common symptoms are colicky abdominal pain, red currant jelly stool, features of acute bowel obstruction, or occult gastrointestinal bleeding [9]. A cohort study by MGF van Lier *et al.,* concluded that the probability of intussusception was at 44 % by age 10 and 50 % by age 20. 69 % had at least one intussusception with the median age at first incidence being 16. Also, the risk of intussusception rises when the polyp size is 15 mm or larger [10]. If left untreated this may lead to bowel ischemia, necrosis, perforation, shock and can progress to Malignancy. So conservative management of reduction by hydrostatic reduction is not implicated if we suspect a pathologic lead point. Definitive surgery is mandated because the presence of lead point may cause recurrence. In surgical approach careful manual reduction of intussusception is done and bowel is assessed for viability if bowel is not viable and gangrenous resection of the gangrenous bowel and end to end anastomosis of the healthy bowel is done (11).

PJS is associated with a 15-fold increased relative risk of malignancies compared to the general population [12]. There is a substantial risk of both GI and non-GI cancers with a combined occurrence of any sort of cancer up to 93% by age 65 years. The intestinal cancer risks included colon (39%), pancreas (36%), stomach (29%), small bowel (13%), and esophagus (0.5%) [2]. Non-digestive organs at risk for cancer include breast (54%), ovary (21%), lung (15%), and uterus (9%). Cancers of the biliary tree and gallbladder have also been reported. Unusual genital tract tumors can also occur [13]. So the patients with PJS should be closely monitored for these malignancies by colonoscopy, sigmoidoscopy and upper GI endoscopy at 3-year interval from the age of 18 according to the American college of Gastroenterology (ACG)[14].

Histopathological examination is necessary in the diagnosis of this syndrome [15]. Here in this case patients age of 16 years with jejunojejunal intussusception and histopathological examination of 3 pedunculated polys were all suggestive of PJS.

However, lack of family history and mucocutaneous pigmentation makes the diagnosis difficult. And also the diagnosis of Intussusception is challenging preoperatively due to its non-specific presentation. So it is important to have well established approach to confirm the diagnosis [15]. Most patients present with features of typical small intestinal obstruction.

Conventional contrast studies previously used are not used now a days. Now a days non-invasive studies like USG and CT scan are being done. Plain x-ray erect abdomen shows air-fluid levels dilated bowel loops and rules out pneumoperitoneum. USG abdomen may show thickened bowel walls and abnormal peristalsis suggesting intestinal obstruction. It shows target or doughnut sign in intussusception with almost 100% diagnostic accuracy in children but only 60% in adults. A CT scan is now the preferred diagnostic tool in adult patients with suspected intussusception, which may detect the site and type of intussusception and lead point and identify complications [15]. It also shows Target or doughnut sign and Pseudokidney sign. A multicenter study by Barussaud *et al.,* [16] reported that CT associated or not with barium enema may be the most accurate modality for diagnosis of intussusception in adults. However, the overall diagnostic approach still depends on the patient's clinical presentation. In patients with suspected PJS or with a strong family history, capsule endoscopy or magnetic resonance enterography is recommended for screening the gastrointestinal tract [14].

Asymptomatic polyps encountered during screening can be effectively treated by polypectomy to avoid further future urgent operations and small bowel syndrome. It can be done by double balloon enteroscopy or during laparotomy by intraoperative endoscopy. This intraoperative endoscopy minimise the need for repeated surgery by visualising entire small bowel and removing all polyps. This “clean sweep” approach is recommended for the management of small intestinal polyps in patients with PJS [17]. However, in cases where polyps are rapidly growing or associated with intussusception, surgical resection remains the recommended treatment [10].

4. CONCLUSION

Diagnosing a case of Peutz-Jeghers syndrome is challenging especially if there is no family history or absence of any mucocutaneous pigmentation as in this case. USG and CT of abdomen are essential to diagnose intussusception presenting as an intestinal obstruction. PJS should be considered in differential diagnosis in patients with intussusception with suggestive histopathological examination of hamartomatous polys

in the intestines showing abnormal muscularis mucosa branching into lamina propria giving it an arborising (christmas tree) pattern even in the absence of family history and mucocutaneous pigmentation which are typical features of PJS, as in this case. This will help in timely diagnosis and management of PJS accordingly. There is also high potential for high gastrointestinal complications and high cancer risk in a case of PJS. So, early diagnosis by clinical examination and surveillance play a key role in reducing these risks. The clean sweep procedure is one of the modern approaches to managing this condition.

5. LIMITATION

Genetic testing for STK11/LKB1 mutation could not be performed because of limited institutional resources and financial constraints. Nevertheless, the diagnosis was established based on characteristic histopathological findings fulfilling WHO clinical criteria for PJS.

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