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Ileoileal Intussusception Caused by A Solitary Polyp in A Young Woman with Peutz-Jeghers Syndrome: A Case Report

Dr. Yogesh[1], Dr. Sanjeev Sharma[2], Dr. Pankaj Dugg[3], Dr. Harsimrat Singh[4]

[1]Junior Resident, Dept of Gen Surgery GMC AMRITSAR
[2]Professor and Head, Dept of Gen Surgery GMC AMRITSAR
[3,4]Assistant Professor, Dept of Gen Surgery GMC AMRITSAR

ABSTRACT

Background: Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant hereditary disorder characterized by hamartomatous polyps in the gastrointestinal tract and distinctive mucocutaneous pigmentation. Intussusception, a condition where one segment of the bowel telescopes into another, is a well-documented complication of PJS, although its presentation in adults is rare.

Case Presentation: We present the case of a 19-year-old woman from Punjab who presented with acute abdominal pain and vomiting. Clinical examination revealed tenderness and guarding in the right upper abdomen, with mucocutaneous pigmentation on her lips and oral mucosa, consistent with PJS. Imaging studies, including ultrasonography and contrast-enhanced computed tomography (CECT), demonstrated small bowel obstruction and ileo-ileal intussusception. Exploratory laparotomy revealed intussusception with a lead-point polyp, which was resected, followed by ileo-ileal anastomosis. Histopathological examination confirmed the presence of a hamartomatous polyp typical of PJS. The patient recovered well postoperatively and remained asymptomatic during six months of follow-up.

Discussion: Intussusception in PJS often results from polyps in the small intestine, most commonly in the jejunum or ileum. Diagnostic challenges arise due to the nonspecific and recurrent nature of symptoms. Imaging modalities such as CT and endoscopy are essential for diagnosis, while therapeutic approaches include endoscopic polypectomy or surgical intervention, depending on the polyp size and location.

Conclusion: Ileo-ileal intussusception in PJS is rare and often misdiagnosed due to its atypical presentation. Prompt diagnosis using a combination of history, physical examination, imaging, and endoscopy is critical to avoid complications. Surgical management remains effective in cases where endoscopic techniques are unavailable or unsuccessful.

Keywords: Peutz-Jeghers syndrome, intussusception, hamartomatous polyps, ileo-ileal intussusception, case report

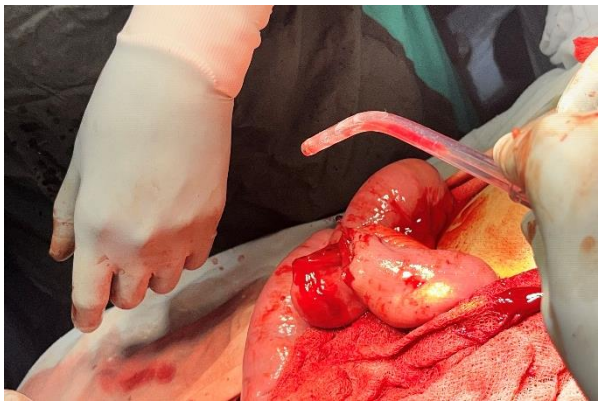

Fig 1 :Intraop picture post intususception reduction

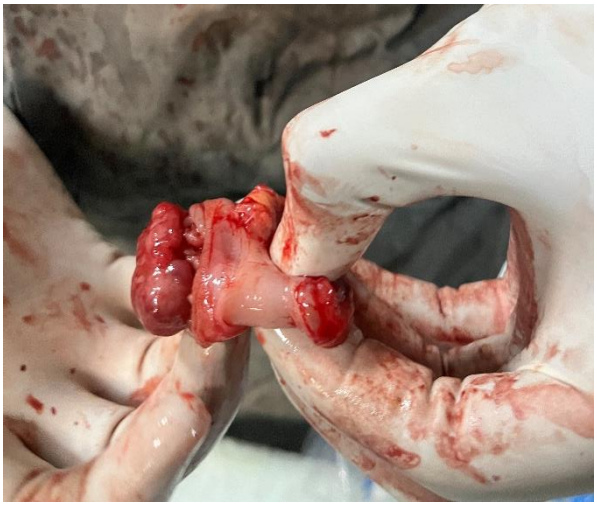

Fig 2 : Excised Ileal leadpoint band with solitary Polyp

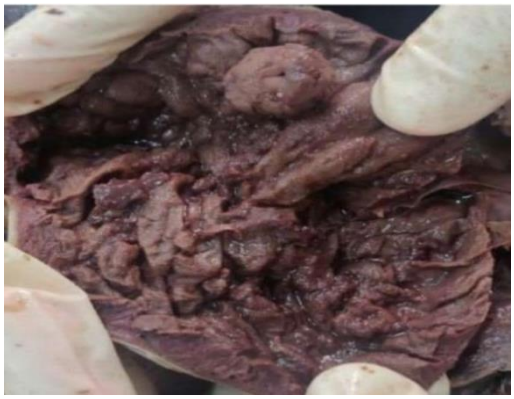

Fig3 : Histopath gross cut section with polyp

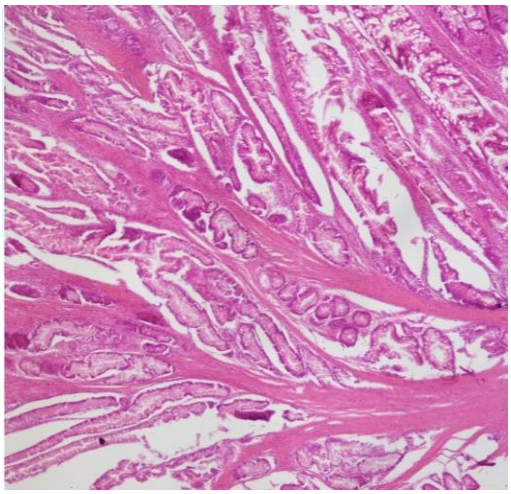

Fig 4 : Microscopic picture

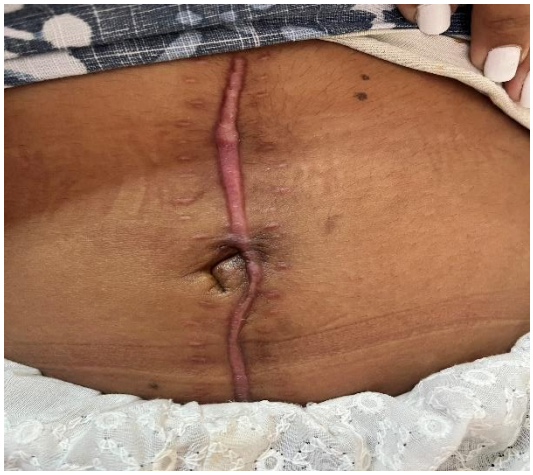

Fig 5 : Post op follow up of patient

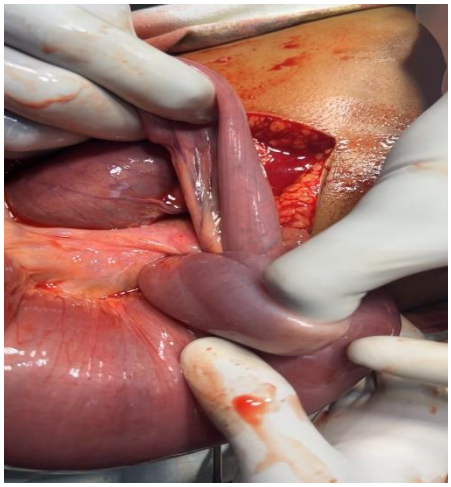

Fig 6 : Intraop Ileo-ileal intususception

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is a rare autosomal dom inant disorder characterized by hamartomatous polyps throughout the gastrointestinal tract and characteristic mucocutaneous pigmentation, primarily of

the lips and oral and gingival mucosae. Polyps are found throughout the gastrointestinal tract but most are confined to the small bowel (60% to 90%) and the colon (50% to 64%). These polyps may also be found at extra-intestinal sites such as the gallbladder, bronchi, bladder and ureter [1].

The hamartomatous polyps of PJS have been reported to cause gastrointestinal bleeding, leading to iron deficiency anemia, in 14% of patients and recurrent intestinal obstruc tions due to the size of polyps in 43% of patients. However, adult intussusception is relatively rare; only 5% to 16% of cases are in adults, and they contribute to only 1% of all causes of intestinal obstruction [2].

CASE PRESENTATION

A 19-year-old woman from Punjab was admitted to our surgical emer gency service with complaints of abdominal pain and vomiting that had intensified over two days.

This colicky abdominal pain was located in her right upper abdomen and was found to be non-radiating in nature. The pain became stronger after eating or drinking, and vomiting was found to relieve the pain for a while.

Her upper abdomen was tender upon palpation, and guarding was significant in her right upper abdomen. Rebound tenderness or rigidity were not detected in our patient. bowel sounds were significantly increased at this location. Mucocutaneous pigmentation was noted on her lips and oral mucosa .

Her vital signs were normal except for mild tachycardia (110 beats/min). Her leukocyte count was 13.1×109 cells/L, and her total and direct bilirubin counts were 4.0mg/dL and 2.9mg/dL, respectively. Her blood amylase level was slightly increased (230U/L). Except for the la boratory values noted, all other laboratory tests were deemed normal.

Our patient had not experienced any prior chronic abdominal pain and had no history of prior hospi talizations or surgery.

Imaging findings: Ultrasonography – Case of dilated gut loops showing whirlpool configuration in the right iliac fossa . ? Volvulus

CECT- Small bowel obstruction with transition points and hypoenchancing edematous wall of mid and distal jejunal loops with surrounding mesenteric edema and stranding , likely closed loop obstruction .

Intra-operative findings: Exploratory laparotomy under general anaesthesia – After painting with providone iodine and draping with sterile sheets , Abdominal midline incision is given and abdomen opened in layers , gut is thoroughly explored and an Ileo-Ileal intussusception is found with dilated proximal and collapsed distal gut , it is reduced manually and the ileal lead point with the band is resected and sent for HPE, ileo-ileal anastomosis done and hemostasis secured . Abdomen washed thoroughly with normal saline and drain placed in the pelvis . Sponge and instrument count confirmed and abdomen closed in layers .

Gross pathologic findings : revealed an 3cm × 3cm × 3cm polypoid tissue mass ,globular in shape and greyish white in color.

Histopathological examin ation: H&E stained sections prepared from the soft tissue piece labelled as the lead point of ileo-ileal intussusception show arborizing network of connective tissue , smooth muscle and lamina propria separating the glands of intestinal type.Immunohistochemical examination showed branching smooth muscle bands in the stroma of the polyp as typ ically seen in hamartomatous polyps of PJS (Figure).

Our patient had an uncomplicated postoperative course and was discharged home on postoperative day six. During the 6 month follow-up period, our patient did not have any further episodes of intussusception.

DISCUSSION:

Peutz-Jeghers syndrome (PJS) is a rare hereditary polyposis disorder associated with autosomal dominant inheritance. It is characterized by the presence of hamartomatous polyps in the gastrointestinal tract and distinctive mucocutaneous pigmentation. This condition arises due to a germline mutation in the STK11 (LKB1) gene, with an estimated prevalence of 1 in 120,000 births. Less than 5% of individuals with PJS do not exhibit mucocutaneous melanotic pigmentation, and fewer than 5% of those with this pigmentation do not develop PJ polyps.[3]

A clinical diagnosis of PJS may be made when any one of the following conditions is present in a single individ ual:

1. Two or more histologically confirmed PJ polyps;
2. Any number of PJ polyps detected in one individual who has a family history of PJS in a close relative
3. Characteristic mucocutaneous pigmentation in an individual who has a family history of PJS in a close relative.
4. Any number of PJ polyps in an individual who also has characteristic mucocutaneous pigmentation [1].

Intussusception occurs when one segment of the bowel (intussusceptum) telescopes into an adjacent segment (intussuscipiens). This phenomenon has been observed in 47% to 69% of adult patients with Peutz-Jeghers syndrome (PJS), with most cases caused by polyps in the small intestine. The majority of reported PJS-related intussusceptions involve the ileum or jejunum, while colo-colonic intussusception is rare.

Diagnosing intussusception in patients with PJS relies on their medical history and physical examination. Common findings include abdominal distention and localized tenderness, though a palpable abdominal mass is uncommon, noted in only 12.5% of cases. Abdominal CT scans are considered the most effective imaging tool for identifying the precise location of polyps and distinguishing between lead-point and non-lead-point intussusception. This distinction is crucial for determining appropriate treatment and reducing unnecessary surgeries.

Endoscopy plays an important role in both diagnosing and treating intussusception. Therapeutic options include endoscopic polypectomy and double-balloon enteroscopy, even for patients with a history of extensive abdominal surgeries. Double-balloon enteroscopy can reduce the need for laparotomy in PJS patients, potentially preventing short bowel syndrome caused by multiple intussusceptions. Additionally, this approach may improve overall health and life expectancy. However, it has limitations in addressing large solitary polyps and requires specialized expertise.

Endoscopic removal is the ideal method of treating a pedunculated polyp; however, when this is not possible, laparoscopy or laparotomy can be a safe and effective alternative for reduction of the intussusception and bowel resection [4,5].

In our case the necessary equipment for endoscopy and double-balloon enteroscopy was unavailable.

In our patient, the polyp causing the intussusception was located in the mid ileal segment with massive dilatation of the proximal segment and an ileo-ileal intussusception. Therefore, we per formed a laparotomy to reduce the intussusception and resection anastomosis.

CONCLUSION:

Ileo-ileal intussusception is a difficult situation because of its infrequency and nonspecific clinic presentation. Diagnosis is usually missed or delayed because of the patient's nonspecific, chronic and

recurrent symptoms. A thorough review of the patient's history, physical exam ination, and radiological and endoscopic findings are critical in these cases, which is a rare form of an uncommon presentation.
Consent Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

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