Patient with Peutz-Jeghers Syndrome
Recurrent Intestinal Obstruction with Double-site Small Bowel Intussusception in Patient with Peutz-Jeghers Syndrome

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Abstract
Background: Peutz-Jeghers Syndrome (PJS) is one of the intestinal polyposis syndromes that can be associated with multiple polyps throughout the gastrointestinal tract in addition to characteristic circumoral mucocutaneous pigmentation.

Case presentation: A 28-year old male patient with known history of PJS presented to the emergency department with acute small bowel obstruction. The patient’s history included two previous laparotomies for similar condition. Investigations revealed elevated leucocyte count, multiple air-fluid levels in plain abdominal x-ray, and dilated small bowel loops with pathognomonic target sign in abdominal ultrasonography. Midline exploratory laparotomy was performed and a double-site small bowel intussusception was found and treated with reduction of the proximal intussusception and resection of one meter of the small intestine including both lesions with re-anastomosis of the bowel ends. The patient showed a smooth, uneventful recovery and was discharged with instructions on regular follow-up.

Conclusion: Patients with PJS who have history of complicated intestinal polyps are amenable for recurrent episodes of complications which may warrant surgical resection of the affected bowel. Small bowel intussusception should be included in the list of differential diagnoses of intestinal obstruction in patients with PJS and the possibility of more than one lesion should be always considered.

Keywords
Peutz-Jeghers syndrome; Small bowel; Obstruction; Intussusception; Double-site

Introduction
Peutz-Jeghers Syndrome (PJS) is an autosomal dominant disorder caused by a germline mutation of the serine/threonine kinase 11 (STK11) tumor suppressor gene in up to 94% of cases [1]. The pathognomonic features of PJS include several hamartomatous polyps throughout the gastrointestinal tract, mainly in the small bowel and colon, and characteristic circumoral pigmentation [2]. The reported frequency of PJS in the United States is about one tenth that of familial adenomatous polyposis. PJS commonly presents clinically in adolescence and early adulthood with approximately half of the patients are symptomatic by the age of 20 years [3].

The numerous polyps of PJS usually manifest with gastrointestinal bleeding and/or recurrent intestinal obstruction. Intestinal obstruction is usually attributed to the size of the intestinal polyps rather than intussusception which is relatively rare; occurring in less than 16% of patients [4]. The intussusception associated with PJS is usually jejunojejunal, jejunoileal, or ileocolic. Nonetheless, duodenal intussusception has also been reported in the literature [5].

Case presentation
A 28-year old male with a known history of PJS who presented to the emergency department with acute small bowel obstruction, the chief complaint of the patient was colicky abdominal pain that was diffuse throughout the abdomen, associated with non-bilious repeated vomiting and absolute constipation. The patient disclosed a relevant history of recurrent episodes of intestinal obstruction secondary to small bowel intussusception for which he underwent two previous laparotomies with resection of the affected bowel loops. He also reported history of frequent blood transfusion for iron-deficiency anemia caused by bleeding intestinal polyps.

Clinical examination revealed a low-grade fever of 37.8°C, tachycardia, normal blood pressure and respiratory rate. Labial and oral pigmentation (Figure 1) were clinically evident on examination. There was no apparent jaundice, cyanosis, or pallor. Abdominal
examination showed a diffusely distended abdomen with upper abdominal tenderness and rigidity. The intestinal sounds were exaggerated.

The routine laboratory tests suggested a significant increase in the leucocyte count\( (16,000/\text{mm}^3) \). Hemoglobin level, platelet count, and serum creatinine were within normal range. Arterial blood gases revealed metabolic acidosis \( (\text{pH}=7.28) \) with normal serum bicarbonate level. Abdominal X-ray revealed multiple air fluid levels and evidence of jejunal obstruction. Abdominal ultrasonography showed thickened edematous intestinal loops with target sign diagnostic of small bowel intussusception. Abdominal computed-tomography (CT) scan was not done, since the diagnosis was already established based on the relevant history, clinical findings, abdominal ultrasound and the patient had intestinal obstruction.

After proper fluid resuscitation and optimization of the patient’s condition, midline laparotomy was conducted at the site of previous scar. Dilated jejunal loops were found with an evident jejuno-jejunal intussusception (Figure 2) 60-cm distal to the dudeno-jejunal flexure. On further exploration of the abdomen, a second ileo-ileo intussusception was found around one meter distal to the first intussusception (Figure 3). The entire small and large bowel was full of variable-size polyps.

After reduction of the proximal intussusception, resection of around one meter of the small bowel involving both the proximal and the distal lesions was performed with re-anastomosis of the bowel ends. On opening of the resected bowel segment, numerous sessile, variable-sized polyps were found. An intra peritoneal drain was inserted and the abdomen was closed with non-absorbable poly prolene sutures.

The patient resumed oral feeding 48 hours postoperatively, and the abdominal drain was removed 72 hours after the procedure. Histopathologic examination of the resected bowel segment revealed numerous hamartomatous mucosal polyps with central core of branching smooth muscle characteristic of Peutz-Jeghers type of polyps. An informed written consent about the publication of this case report was obtained from the patient.

Discussion

The patient in the present report met the WHO diagnostic criteria for PJS [6] which comprise of any number of hamartomatous polyps throughout the entire gastrointestinal tract associated with characteristic, prominent, mucocutaneous pigmentation. Despite the rare incidence of PJS (1 in 50 000 to 1 in 200 000 live births) [7], it is considered a clinically diverse disease entity with various presentations and a high lifetime risk of developing complications or malignancy.

Double-site intussusception is a very rare condition, and to the best of our knowledge it has been reported in the literature sparingly [8,9]. Only a few conditions can affect the gastrointestinal tract at multiple sites and includes polyposis syndromes such as familial adenomatous polyposis (FAP), juvenile polyposis syndrome and PJS. Nevertheless, lymphoma in younger patients as well as intestinal tuberculosis should be also included. The presented case might be one of the rare case scenarios by not only presenting with double-site intussusception but also recurrent acute intestinal obstruction after two previous laparotomies.

Although due to the relevant history of PJS and previous attacks of intussusception the etiology of intestinal intussusception in the present case was easily predictable, other causes of adult intussusception should be considered in the differential diagnosis. These other causes include: malignant tumors which account for 66% of large bowel intussusception and 30% of small bowel intussusception, adenomatous polyps, lipomas, fibroma, hamartomas, lymphoid hyperplasia, and inflammatory bowel diseases. It should be noted that around 16% of small intestinal intussusception are idiopathic with no organic lesions identified [10].

Surgical resection might not be the first choice for treatment as endoscopic resection or combined modalities have shown better outcome in terms of quality of life [11]. However, considering the patient’s history, surgical resection can be deemed appropriate for patients with troublesome polyp’s complicated with bleeding, intestinal obstruction and intussusceptions.

Owing to the high risk of complications and malignancy, patients with PJS should be directed to a surveillance program. There is no evidence that suggests when to start the surveillance process; however, the general consensus is from the onset of symptoms. Our
patient was scheduled for regular follow-up visits and according to available guidelines and recommendations [2], complete blood count, liver function testing, and clinical examination will be performed annually as well as video capsule endoscopy or upper gastrointestinal endoscopy every three years.

**Conclusion**

Patients with PJS who have history of complicated intestinal polyps are amenable for recurrent episodes of complications which may warrant surgical resection of the affected bowel. Small bowel intussusception due to PJS should be included to the list of differential diagnoses of intestinal obstruction and the possibility of more than one lesion should be always considered. Surveillance of patients with complicated intestinal polyps is imperative in order to prevent further complications in the future.

**References**