Case Report

Unveiling an unusual presentation in an unaccustomed syndrome: Intussusception and Peutz-Jeghers - A case report

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ABSTRACT

Peutz-Jeghers syndrome (PJS) is an uncommon hereditary gastrointestinal (GI) polyposis syndrome that is associated with pathognomonic mucocutaneous hyperpigmentation. Here, we present a case of ileoileal intussusception causing acute small bowel obstruction in a young female with PJS. In our patient, we considered PJS due to the findings of intestinal polyposis with mucocutaneous hyperpigmentation in an otherwise young female with a positive family history of similar hyperpigmentation who presented with clinical features suggestive of acute small bowel obstruction and on imaging was confirmed to be secondary to intussusception. She underwent an exploratory laparotomy and bowel resection, followed by endoscopic evaluation of the entire GI tract.

Key words: Bowel obstruction, Cancer screening, Mucosal hyperpigmentation, Peutz-Jeghers syndrome

Peutz-Jeghers syndrome (PJS) is an uncommon hereditary gastrointestinal (GI) polyposis syndrome that is associated with pathognomonic mucocutaneous hyperpigmentation. It has a variable penetrance and is inherited in an autosomal dominant fashion. The estimated incidence is 1 in 8,300–1 in 200,000 live births and a prevalence rate of 1 in 50,000–1 in 200,000 people [1]. Patients have varied presentations, including small bowel obstruction, anemia, or features of malignancy in the late stages. The most common complication is the development of intussusception in adults, which warrants a surgical exploration. Most of the currently available guidelines on the screening and management of PJS are based on expert opinion rather than evidence-based. The disease process significantly affects the quality of life, and efforts need to be taken to improve it by scientific advances and research [2].

Here, we present a case of Ileo-ileal intussusception causing acute small bowel obstruction in a young female with PJS.

CASE PRESENTATION

A 29-year-old female, resident of Uttar Pradesh, India, and homemaker by occupation, presented to our general surgery emergency department with chief complaints of abdominal pain for 8 days and bilious vomiting for 5 days. The abdominal pain was perceived throughout the abdomen, more in the lower quadrants. It was acute in onset, colicky in nature, and non-radiating. It was aggravated by food intake and not relieved by taking medications. The abdominal pain was associated with non-passage of flatus and feces and bilious emesis for 5 days before presentation. The patient denied any history of blood in stools, melena, jaundice, fatigue, or similar acute episodes of pain in the past. No history of loss of appetite and/or loss of weight, menstrual irregularities, or any prolonged hospitalization. She, however, complained of on-and-off constipation. She had no known medical comorbidities and had a cesarean section done 4 years back for her first daughter. A reminder of the history was non-contributory.

On examination, the patient was alert, oriented, with a Glasgow Coma scale score of 15/15. On presentation, she had a pulse rate of 82/min; blood pressure of 116/74 mmHg; respiratory rate of 17/min, and was afebrile. She was thin-built and malnourished. She was noted to have hyperpigmented macules in the lips, inner mucosa of the cheeks, and palms (Fig. 1). On retrospective questioning, it was found that the patient's mother also had similar perioral pigmentation. She was not evaluated for any polyps. Per abdominal examination, showed a distended abdomen with raised bowel sounds. No signs of guarding or rigidity, or rebound tenderness were noted. A digital rectal examination showed a collapsed rectum with no obvious palpable mass or blood staining. An initial working diagnosis of small bowel obstruction was made, and the patient was further evaluated.

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Figure 1: Hyperpigmented macules in lips and inner cheek

Routine blood investigations showed complete blood count, renal function, and liver function tests being within normal limits. X-ray abdomen showed multiple air fluid levels. Ultrasonography of the abdomen gave an impression of ileoileal intussusception with proximal bowel dilatation. Contrast-enhanced computed tomography of the abdomen confirmed the distal ileoileal intussusception causing bowel obstruction. It also revealed multiple homogeneously enhancing intraluminal polypoidal mucosal lesions in the jejunum and proximal ileum. A provisional diagnosis of PJS was considered, taking into account the intestinal polyposis with mucocutaneous hyperpigmentation in a young female with a positive family history presenting as acute small bowel obstruction secondary to intussusception.

She underwent an exploratory laparotomy, and an ileoileal intussusception 25 cm proximal to the ileocecal junction was noted. The length of the bowel segment telescoping was 20 cm and was congested and edematous with concealed perforation at approximately 30 cm proximal to the ileocecal junction. Attempts at manual reduction failed, and so the decision was made to do resection with the creation of a double-barreled ileostomy (Fig. 2). Multiple polypoidal masses were palpable throughout the jejunum and proximal ileum.

The patient tolerated the surgery well and had an uneventful post-operative course, and was discharged home on post-operative day six. On routine follow-up visits, she had an uneventful recovery from the initial surgery. Due to the unavailability of endoscopic facilities at our hospital, she was referred to a nearby higher center for the same. An esophagogastroduodenoscopy, enteroscopy through the proximal ileostomy and colonoscopy was done 4 weeks after the surgery. The endoscopies reported the presence of multiple hamartomatous polyps throughout the stomach, duodenum, colon, and rectum. PJS-type polyps were confirmed by histopathological analysis of the polyps that were biopsied. Histopathological examination of sections from gastric and rectal polyps showed arborizing smooth muscle bundles extending till the surface, associated with the mucosa of the native site of origin. No evidence of dysplasia and/or malignancy was noted. Focal active colitis was additionally noted in the sigmoid colon biopsy submitted.

She had a cervical pap smear that was normal. She then underwent a distal loopogram that showed a normal patent distal loop, and so had stomal reversal done after 3 months from her index surgery. The patient had an uneventful post-operative course and was discharged post-operative day 5 with instructions for follow-up.

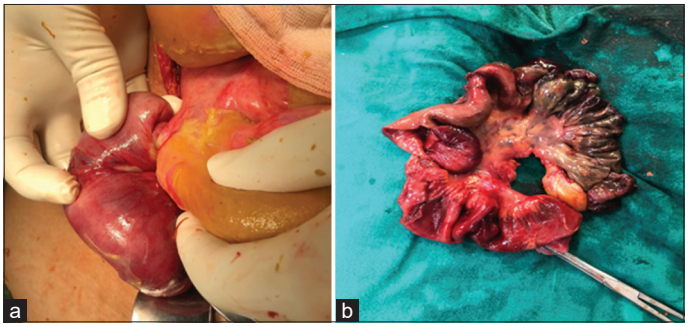

Figure 2: (a) Intraoperative picture of ileoileal intussusception; (b) Resected bowel segment with the telescoping bowel segment reduced

Genetic testing and magnetic resonance cholangiopancreatography were not done due to a lack of resources at our facility.

DISCUSSION

Jan Peutz was the first to describe the association of polyps and mucocutaneous papules in 1921 [3]. In 1949, it was formulated as "Peutz-Jeghers syndrome" by Harold Jeghers [4]. Even earlier, the "Hutchinson twins" with oral and labial pigmentation were described by Conor in 1895 and were later illustrated by Hutchinson in 1896 [5,6]. However, the association of polyps and cancer with mucocutaneous pigmentation was not made at that time.

PJS is one of the hereditary polyposis syndromes. It is inherited in an autosomal dominant fashion. Germline mutation in STK11 (*LKB1*) genes in chromosome 19p13.3 has been reported to be responsible for PJS [7]. It is a tumor suppressor gene that plays a key role in cell cycle regulation. Loss of function mutations will lead to the development of unregulated proliferation of cells, resulting in hamartomas. STK11 mutation testing analysis should be done only in patients with phenotypic features of PJS. Inhibition of COX-2 will help prevent the progression of hamartomatous polyps [8]. The most common malignancies associated with PJS are of colorectal, breast, small bowel, gastric, pancreatic, and genital origin. PJS patients had a 39% risk of developing colorectal malignancies compared to 5% in the general population [9]. It was followed by breast (32–54%), stomach (29%), ovary (21%) and small bowel (13%) malignancies. The cumulative lifetime cancer risk in PJS patients rise up to 93% [10]. The incidence of PJS is noted to be around 1 in 8,300–200,000 live births [11,12]. It is equally distributed among males and females. There is no racial or ethnic predilection to the development of PJS.

The clinical diagnosis is based on having one of the following clinical features [13]: Two or more PJS-type hamartomatous polyps confirmed histologically; any number of PJS-type polyps with a family history of PJS in at least one close relative; any number of PJS-type polyps who also have characteristic mucocutaneous pigmentation; and characteristic mucocutaneous pigmentation

with a family history of PJS in at least one close relative. The patients who exhibit only mucocutaneous pigmentation are defined as having incomplete PJS.

The presenting complaints are abdominal pain, blood in stools, and features of obstipation. Classical presentation is bowel obstruction due to intussusception from the hamartomatous polyps in the GI tract [14]. Intussusception accounts for 1–5% of the etiology of bowel obstruction overall, and in adults, it is more often than not associated with a lead point pathology [15]. Peutz-Jeghers polyps have papillary villous architecture with tree-like arborization of smooth muscle bundles with a relatively normal overlying epithelium. Dysplasia is rare in these polyps. Epithelial components are usually arranged in a lobular configuration with secondary changes of erosion and ulceration [16].

Management is of two-fold. It consists of screening for prevention of the manifestations of the disease condition and treatment of the complications that arise from it. Patients with confirmed PJS as well as their family members should be on close surveillance and periodic screening evaluations to improve their clinical outcomes in terms of prevention of avoidable surgical emergencies [17].

Dutch surveillance recommendations and European experts' guidelines for screening in Peutz-Jeghers syndrome patients are put forward to enhance the screening of patients with PJS [18,19]. Routine surveillance of uterine, ovarian, thyroid, lung, or liver malignancies is not advocated unless patients show symptoms pertaining to that malignancy.

There are no definitive consensus guidelines for PJS polyps' management with each patient requiring an individualized approach depending on the manner and time of presentation. Management of hamartomatous polyps involves upper GI endoscopy and colonoscopy for polypectomy, with open surgical methods reserved for dealing with complications, such as bowel obstruction or hemorrhage. Recent advances allow a combined approach of surgical and intraoperative endoscopic procedures for the management of multiple hamartomatous polyps in PJS patients [20].

Other hamartomatous polyposis syndromes, such as Juvenile polyposis syndrome, hereditary mixed polyposis syndrome, Familial adenomatous polyposis, Bannayan-Riley Ruvalcaba syndrome, and Cowden syndrome, and others are to be considered as possible differentials. Other syndromes with mucocutaneous hyperpigmentation, such as Laugier-Hunziker syndrome and isolated melanotic mucocutaneous pigmentation, should also be considered. Understanding the importance of early diagnosis and close follow-up will help patients lead a better quality of life, failure of which would result in multiple hospital visits and surgeries as described in this report [21].

CONCLUSION

Given the uncommon prevalence of PJS and the extremely high risk of incidence of cancer in these patients, identification of patients early with screening of their family members becomes vital. Close surveillance will improve the possibility of early diagnosis of associated malignancies and also prevent any surgical complications that would otherwise warrant surgical management. Prophylaxis also offers the possibility of endoscopic polypectomy, thereby preventing short gut syndrome from less economical bowel resections if warranted.

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