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CASE REPORT

Recurrent Intussusception Secondary to Peutz-Jeghers Syndrome – A Case Report

Zakir Jamal,[1] Faryal Ali,[2] Haifa Khan,[3] Mohibah Khaliq,[4] Kanza Farrukh,[5] Fahad Akhtar,[6] Sajid Ali Shah,[7] S H Waqar[8]

[1,4,5,6]Postgraduate Resident, Department of General Surgery, PIMS, Islamabad
[2,3]House Officer, Department of General Surgery, PIMS, Islamabad
[7]Associate Professor, Department of General Surgery, PIMS, Islamabad
[8]Professor & HOD General Surgery, PIMS, Islamabad

ABSTRACT

Peutz-Jeghers syndrome is a rare genetic condition characterized by the development of benign, hamartomatous polyps along with characteristic mucocutaneous lesions. It is an inherited autosomal dominant disorder due to mutations in the STK-11 gene. This syndrome is associated with short bowel syndrome due to recurrent intussusception and an increased risk of malignancies later in life.

Reported here is a case of a 15-year-old female suffering from Peutz-Jeghers Syndrome. She had repeated episodes of intussusception for which she has undergone exploratory laparotomy thrice.

Key Words: Peutz-Jeghers syndrome, Intussusception, Benign hamartomatous polyps.

Correspondence:
Dr Zakir Jamal
Postgraduate Resident,
Department of General Surgery,
PIMS, Islamabad
Email: zakirjamal89@gmail.com

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Introduction

Peutz-Jeghers syndrome (PJS) is a rare genetic condition characterized by benign, hamartomatous polyps throughout the small and large bowel, along with distinctive pigmented mucocutaneous lesions. The incidence of Peutz-Jeghers syndrome is found to be 1 in 100,000, around the globe;[1] however, limited data is available, especially among the Asian population. The pathogenic gene identified as STK11 is present in about 70% of patients with clinical symptoms resembling PJS.[2] A strong family history is a key factor in diagnosing such cases.[3] There is a rare, sporadic occurrence, too. Solitary PJS must be considered when there is an absence of characteristic mucocutaneous pigmentation, family history of PJS, or additional polyps within the GI tract, but pathological analysis suggests the disease.[4] There is a higher incidence of developing malignancies later in life, such as Colorectal cancer, Breast cancer, Pancreatic cancer, Ovarian cancer, Gastric cancer, Lung cancer, small bowel cancer, Cervical and Uterine cancer.[5] Patients have typical symptoms, including recurring bouts of abdominal pain, diarrhea, intussusception, and gastrointestinal bleeding. The presence of a pathognomonic sign of pigmented mucocutaneous lesions can help formulate the diagnosis.[6]

Here we are going to discuss a case of PJS who presented to us with repeated attacks of intestinal obstruction due to intussusception.

Case Report

A 15-year-old female, born to consanguineous parents, presented to the emergency department with a history of abdominal pain and constipation for 4 days, vomiting, and abdominal distension for 3 days. The abdominal pain was gradual in onset, colicky in nature, and did not have any aggravating or relieving factors. It was accompanied by multiple episodes of bilious, non-

projectile vomiting. Her history includes an exploratory laparotomy at 8 years of age for intussusception when resection and anastomosis were done, but no previous record was available. She had a family history of her maternal aunt with similar complaints, for which she had undergone laparotomy too, but no proper documentation was available.

On examination, the patient was anemic and tachycardic, with visible brownish pigmented spots on her buccal mucosa and lower lip (Figure 1). Her abdomen was generalized, tender, and distended, along with sluggish bowel sounds. The right iliac fossa was empty with a mass in the right hypochondrium. Mass was firm, non-fluctuant, with indistinct margins. The remainder of the physical examination was unremarkable. Digital rectal examination revealed an empty rectum, normal anal tone, with no fecal staining. Laboratory investigations were mostly unremarkable, except for hypochromic, microcytic anemia with a hemoglobin level of 10.2 g/dL.

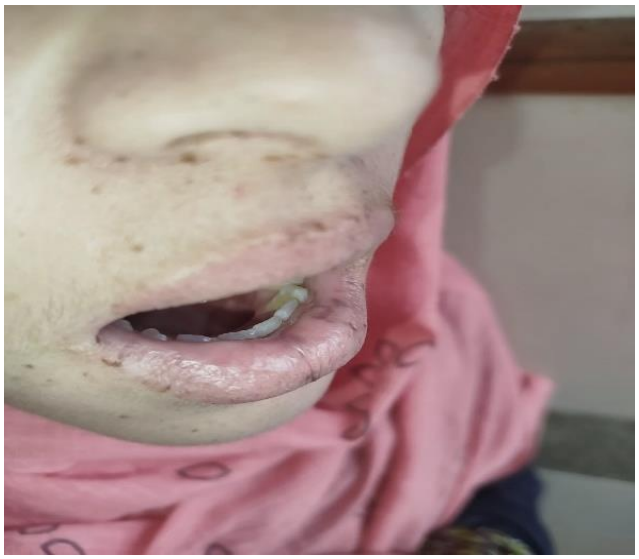

Figure 1. Multiple Pigmented Spots.

Abdominal X-rays showed multiple air-fluid levels and dilated small bowel. Abdominal ultrasound showed a doughnut sign, which is pathognomonic for intussusception (Figure 2). The patient subsequently underwent another exploratory laparotomy, ileoileal intussusception was found, which was released with a polyp as the leading point. The necrotic segment was resected. Multiple intraluminal polyps were identified at 2, 4, 5, and 7 feet from the duodenojejunal junction (DJ), with the largest polyp located 1.5 feet from the ileocecal junction (ICJ). Segmental resection, polypectomy were performed, and a double-barreled ileostomy was fashioned.

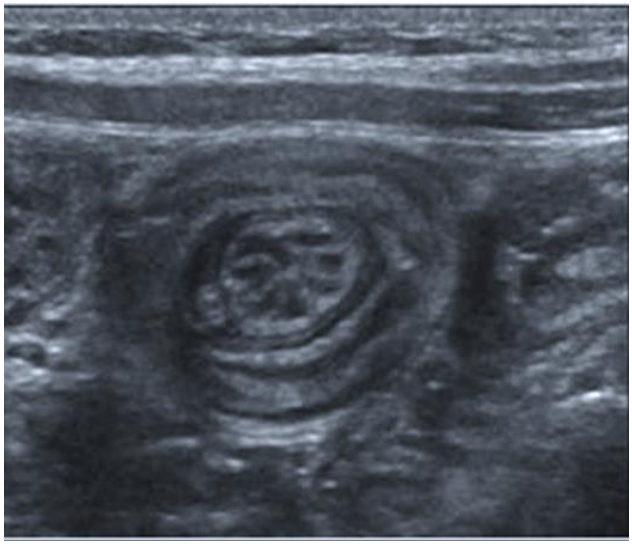

Figure 2. Ultrasound Showing Intussusception.

Histopathological report of the resected segment revealed colonic mucosa with one fragment showing a retention-type polyp, thickened muscle, negative for dysplasia or malignancy, and significant features suggestive of Peutz-Jeghers syndrome. The patient was advised to follow up closely.

After six months, ileostomy reversal was planned. Meanwhile, she presented to the emergency department with symptoms of abdominal pain, vomiting, and constipation. Her abdomen was tense and tender with a mass at the paraumbilical region. Chest X-ray showed pneumoperitoneum. She underwent another exploratory laparotomy, which revealed recurrent ileoileal intussusception 1.5 feet from ICJ. There was a perforation in the necrotic segment 2.5 feet from the ileocecal junction (ICJ), with 1000 mL fecal contamination. Resection of the necrotic segment and ileostomy was refashioned. Later, a colonoscopy through the stoma revealed multiple polyps extending from the rectum to the ascending colon, with the largest polyp located near the splenic flexure. Polypectomy was done, and multiple biopsies were subsequently taken, which were all negative for dysplasia or malignancy.

She developed stomal prolapse after 2 months, but otherwise, there were no complaints (Figure 3).

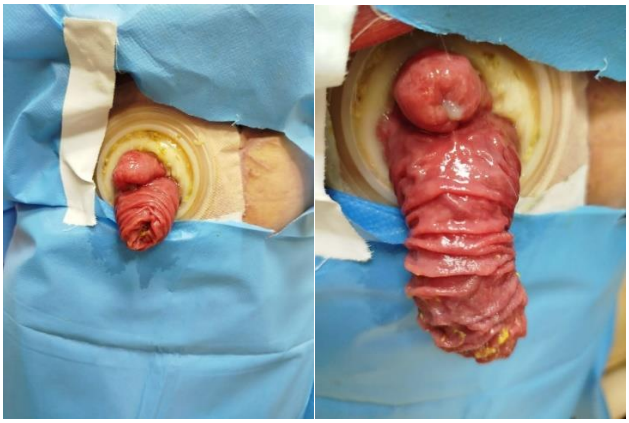

Figure 3. Stoma Prolapse

After another 3 months, again, ileostomy reversal was planned. Per operatively, few polyps were identified, and polypectomy was done along with reversal. The specimen was sent for histopathology. She had an uneventful recovery and was discharged with advice to follow up.

Discussion

Peutz-Jeghers is a rare syndrome. It is characterised by Hamartomatous polyps, two or more in number, at least one first-degree relative, and mucocutaneous pigmentation.[7] Our patient had all of these features.

Its occurrence is equal in both males and females. The clinical manifestation of PJS is asymptomatic, which can present at any time as abdominal pain and intestinal obstruction due to intussusception or prolapse of polyps through the rectum, occult per-rectal bleeding, and rarely as malignancy.[8] An early diagnosis, along with close follow up is important for early detection and overall survival. Initial management is just like any other case of acute abdomen. Ultrasound and computed tomographic findings are characteristic of the doughnut and target sign, respectively. Barium study reveals claw sign.[9] There are recent advances like video capsule endoscopy (VCE), CT enterography, or magnetic resonance enterography (MRE) that can help in early detection in suspected cases.[10]

Polypectomy is recommended for polyps in the stomach or colon that are more than 1 cm in size, found during endoscopic surveillance. Surgery is recommended for symptomatic or rapidly growing small intestinal polyps or asymptomatic polyps more than 1–1.5 cm in size. Surveillance in the form of endoscopy with polypectomy reduces the frequency of emergency laparotomy and bowel loss resulting from recurrent intussusception.[8] Balloon-assisted enteroscopy allows polypectomy of small-bowel polyps. Intraoperative enteroscopy and enterotomy are other techniques by which large distal small-bowel polyps can be removed.[9] If there is a family history, then treatments like prophylactic mastectomy can be offered to patients. Similarly, for gynecological carcinomas, however, no prospective data are available to support it.[9] Help must be taken from newer techniques, as explained in managing such patients.

Conclusion

Short bowel syndrome is a matter of great concern in such cases. To prevent intra-abdominal adhesions due to repeated laparotomies, management should be combined with recent techniques, along with close surveillance and counselling.

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