CASE REPORT

A rare cause of mechanical intestinal obstruction due to small bowel intussusception: "A solitary Peutz-Jeghers type hamartomatous polyp"

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ABSTRACT

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant disorder which is characterized by hyperpigmentation in mucocutaneous membranes and hamartomatous polyps in the gastrointestinal tract (GIT). Common complications reported in patients with PSJ are bleeding and mechanical intestinal obstruction due to the hamartomatous polyps. There is also an increased risk of gastrointestinal and extra-intestinal malignancies in patients with PJS. A 28-year-old female patient was admitted to the emergency service with complaints of abdominal pain and vomiting. In addition to distention and tenderness on abdominal examination, revealed hyperpigmented lesions on her lips. An abdominal examination did not reveal any scar from the previous abdominal operation. The patient with suspected mechanical intestinal obstruction at pre-diagnosis demonstrated intussusception in the distal jejunal loops on abdominal tomography. In the diagnostic laparoscopy observed intussusception in jejunal loops. After a minimal suprapubic incision, small intestine loops were checked through alexis with bidigital palpation and no other intraluminal mass were detected. Laparoscopy-assisted jejunojejunal resection and anastomosis was performed for the intussusception segment, where the polyb is located. It has been recommended that endoscopic polyps removal should be performed to avoid multiple surgical resections, which lead to short bowel syndrome. It has been recommended that endoscopic polyps removal should be performed to avoid multiple surgical resections, which lead to short bowel syndrome. By the nature of the disease, there may be multiple polyps simultaneously in the GIT and the associated risk of recurrent intussusception attacks in patients with PJS. To prevent short bowel syndrome and intra-abdominal adhesions due to repeated, laparotomies treatment with combined endoscopy and laparoscopic/laparoscopy-assisted surgery should be preferred in patients with PJS.

Keywords: Hamartomatous polyp; intussusception; laparoscopic surgery; Peutz-Jeghers syndrome.

INTRODUCTION

Intussusception cases are generally reported in childhood and only 5% of the total cases are observed in adults. Intussusception constitutes 1–5% of the causes of mechanical ileal obstruction in adults. Approximately 90% of the adult intussusception cases require definitive therapy and this is mostly ensured with surgical resection.[1]

Peutz-Jeghers syndrome (PJS) was first identified in 1921 by Peutz, and the clinical features of the syndrome were defined in 1949 by Jeghers.[2] PJS is a disease with an autosomal dominant inheritance pattern caused by serine/threonine kinase 11 (STK11/LKB1) tumor suppressor gene mutations on the short arm of chromosome 19 (19p13.3).[3] The prevalence is reported to be 1/100,000. The prevalence in females and males is equal.[4] It is characterized by hyperpigmentation in mucocutaneous membranes and hamartomatous polyps commonly reported in the gastrointestinal tract (GIT) but also outside the GIT. The polyps in the GIT are most commonly localized

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in the small intestine.[5] Common complications reported in patients with PSJ are bleeding due to hamartomatous polyps, abdominal pain, and mechanical intestinal obstruction clinic secondary to intussusception. Approximately 50% of the patients experience intussusception complications to due polyps localized in their intestines during their lifetime.[6]

This article presents an adult case diagnosed with PJS who visited us due to mechanical intestinal obstruction clinic caused by jejunal intussusception.

CASE REPORT

A 28-year-old female patient was admitted to the emergency service with complaints of abdominal pain for 2 past days and vomiting that started during the day. It was learned from her medical history that she used iron preparations due to iron deficiency anemia and that she had no other disease. It was reported that the patient was unable to defecate for the past 3 days. The direct abdominal radiography of the standing patient showed wide-base small intestine type air-fluid levels (Fig. 1), and distention was detected during the abdominal examination.

Since the patient with mechanical intestinal obstruction clinic is in her third decade and she has no history of past operations, and abdominal cross-sectional imaging was requested to reveal the pathology causing the obstruction. In the abdominal tomography of the patient, mechanical intestinal obstruction due to intussusception in the distal jejunal loops was detected (Fig. 2). After taking a more detailed medical history, it was learned that the patient had been hospitalized at an external center twice in the past 3 years with similar clinical findings, and her symptoms regressed after medical treatment. The physical examination of the patient revealed hyperpigmented lesions on the lips. It was learned that these lesions existed since birth.

The routine surgical blood tests were requested. The complete blood count of the patient revealed a hemoglobin value of 8.4 g/dL and a leukocyte value of 21,000/mm^3. Her blood biochemistry was within normal limits except for hypopotassemia and increased C-reactive protein.

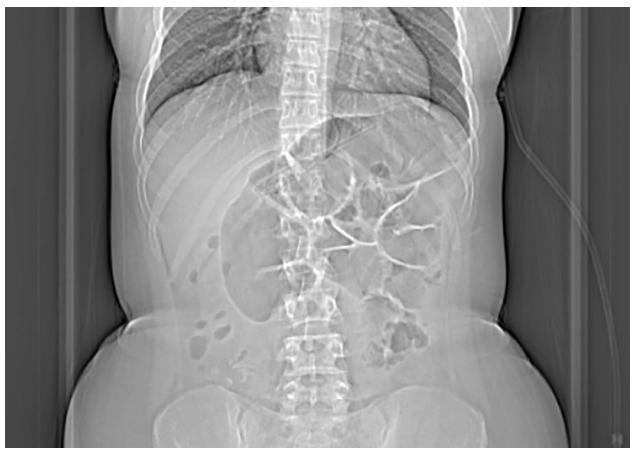

Figure 1. Wide-base small intestine type air-fluid levels on standing direct abdominal radiography.

Oral intake of the patient, who had a nasogastric catheter, was discontinued. She was started on appropriate fluid and antibiotic treatment. The patient was admitted to the general surgery service and received medical treatment for 2 days; however, since the patient was still unable to defecate and the nasogastric drainage continued to be bilious, diagnostic laparoscopy was performed.

In the laparoscopy, it was observed that the jejunal loop of approximately 20 cm from 180 cm of the jejunal loops distal to the treitz ligament was proximally intussuscepted. This area was excised from a 5 cm suprapubic incision using Alexis mass lesion within the jejunal loop which was palpated (Fig. 3). Partial resection and jejunojejunal anastomosis were performed on the 20 cm jejunal segment that was intussuscepted to in-

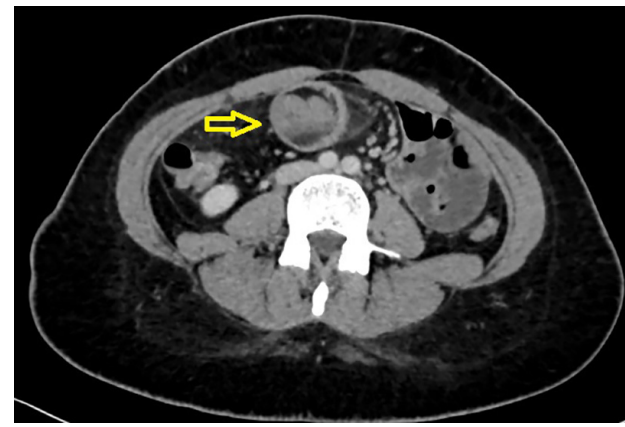

Figure 2. Intussusception in the distal jejunal loops on abdominal tomography.

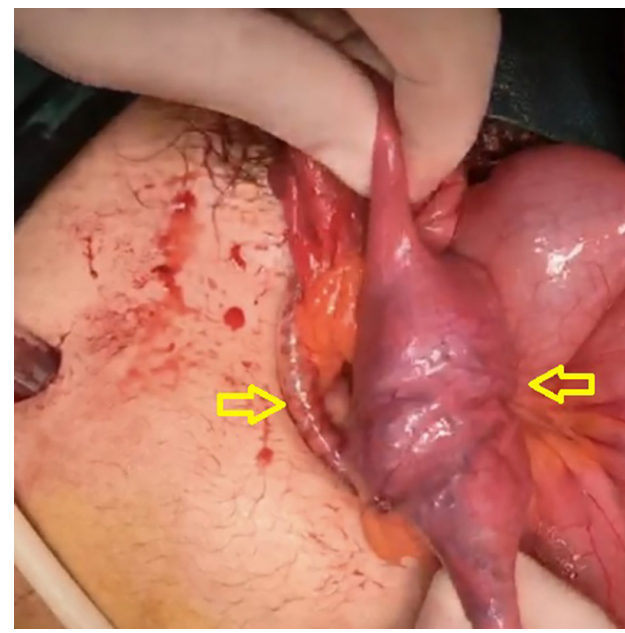

Figure 3. Alexis and mass lesion within the jejunal loop.

clude the area of the intraluminal mass. Following the anastomosis, the small intestine loops were checked through Alexis with bidigital palpation and no other lesions were detected.

In the post-operative follow-up, the general condition of the patient was good and she was able to defecate and started oral intake; therefore, she was discharged with full recovery. The patient, who was followed up regularly after the operation and who had a hamartomatous polyp specific to PJS according to the pathology results, was referred to the medical genetics department of our hospital for genetic screening. The STK11/LKB1 gene mutation was detected in the genetic screening.

Family members of the patient diagnosed with PJS were referred for genetic screening. The patient was informed about PJS and was followed up regularly in our clinic.

DISCUSSION

PJS was first identified in 1921 in a case report published by a Dutch medical doctor named Jan Peutz, who revealed that there was a link between mucocutaneous macules and gastrointestinal polyps in the members of a family.[1,7] Harold Jeghers, an American medical doctor confirmed that the disease was a syndrome in his studies in 1949.[7,8]

PJS is a disease with an autosomal dominant inheritance characterized by hamartomatous polyps and hyperpigmentation in mucocutaneous membranes in the GIT.[5,9] The prevalence of PJS, which is a rare disease, has been reported to be 1/100,000. It has no racial superiority and its prevalence in females and males is equal.[4,9]

Although the etiology of PJS is not clear, it has been proven that 70–80% of the patients have serine/threonine kinase 11 (STK11/LKB1) tumor suppressor gene line mutations on the short arm of chromosome 19 (19p13.3).[3,10] This mutation leading to loss of STK11 protein kinase activity has also been associated with the occurrence of certain malignant diseases. It has been reported that the incidence of malignancy in patients with PSJ is increased due to the presence of this mutation compared to the normal population. The most common malignancies associated with PJS are colorectal cancers; and it has been reported that breast cancer, small intestinal cancers, gastric, and pancreatic cancers are more common in patients with PJS compared to the normal population.[11]

The World Health Organization (WHO) established diagnostic criteria for PJS. Having any of the four criteria is sufficient for diagnosis.

- Three or more histopathologically confirmed Peutz-Jeghers type polyps
- A positive family history of PJS and any number of Peutz-Jeghers type polyps
- A positive family history of PJS and characteristic prominent mucocutaneous pigmentation
- Any number of histologically confirmed PJS polyps and characteristic prominent mucocutaneous pigmentation.[10,12]

The patient in our case presentation met the diagnostic criteria of the WHO with the presence of hyperpigmented lesions on the lips and the presence of jejunal Peutz-Jeghers type hamartomatous polyp.

Pigmentations usually occur in the 1st year of life, but they may fade and, in some cases, even disappear in adulthood.[6] When PJS is suspected during the pre-diagnosis, positive family history due to the autosomal dominant inheritance of the disease, and the presence of mucocutaneous pigmentation, which is common from birth, allows the diagnosis in childhood. It has been reported that the disease is frequently diagnosed within the first two decades with a detailed medical history and a careful physical examination.[9]

In PJS, polyps usually show symptoms in adolescence and early adulthood. About 90% of people with PJS who reach adulthood have been reported to have small intestine polyps.[6,9] Although polyps are most common in the small intestine, which is the primary location, they have also been reported in the colon, stomach, and rectum, in descending order of incidence. The literature search reveals that polyps are often of the small intestine origin, as in our case presentation. Polyps located outside the small intestine have also been reported in the literature. In the case presentation of Hammouda et al.,[12] the polyp originated from the ascending colon, and in the case presentation of Shah et al.,[7] the polyp originated from the colon. Gülten et al.[13] presented the presence of PJS type rectal polyp in their case presentation. Polyps are most commonly located in the jejunum in the small intestine and are also located in the ileum and duodenum, respectively. Other than GIT, extraintestinal polyps have been reported in the gallbladder, ureter, respiratory tract, and tonsils.[6]

Patients with PJS usually present with polyps-related symptoms such as abdominal pain, gastrointestinal bleeding, anemia associated with this, and intestinal obstruction due to intussusception.[14] Symptoms often occur in the first two decades. Intussusception is a significant condition that can cause mortality in patients with PJS.[9,14]

Intussusception cases are generally reported in childhood and only 5% of the total cases are observed in adults. Intussusception constitutes 1–5% of the causes of mechanical ileal obstruction in adults.[1] This condition, which is generally primary and benign in children, responds to medical treatment in 80% of the cases. On the other hand, approximately 90% of intussusception cases reported in adults occur secondary to a pathological condition. The presence of the primary pathology causing intussusception such as polyps, carcinomas,

Meckel's diverticulum, colon diverticulum, strictures, and benign neoplasms should be considered in the differential diagnosis in adult intussusception cases.[1,15] Surgical treatment planned to eliminate mechanical obstruction are required in 70 –90% of these pathologies.[1]

The primary pathology that causes intussusception in patients with PJS is hamartomatous polyps. The pathology that most commonly requires surgery in this patient group is intussusception, and it may not always possible to diagnose preoperatively.

In the case presentation of Nasri et al.,[9] the intussusception due to jejunal polyps was detected intraoperatively, and it is thought that intussusception may be asymptomatic and possible intestinal polyps are inevitable for this complication. In the cohort study of Wang et al.,[16] intestinal obstruction due to intussusception was diagnosed intraoperatively only in one case, and all other cases were diagnosed preoperatively. In our case presentation, the presence of intussusception due to jejunal polyp was detected by pre-operative examinations. In the light of the current literature, we believe that the preoperative or intraoperative diagnosis of intussusception secondary to jejunal polyp depends on the size of the polyp, the length of the invaginated loop, and the effectiveness of pre-operative imaging examinations.

As in our case presentation, the purpose in patients undergoing emergency operations secondary to mechanical intestinal obstruction is to eliminate mechanical obstruction in the GIT. However, it should be noted that hamartomatous polyps may involve the entire GIT and may be in large numbers in this patient group. Focusing only on the segment with major intussusception may lead to incomplete surgery in some patients. In the case presentation of Emile et al.,[17] they reported intussusceptions in two different regions simultaneously in the small intestine of a patient with PJS. This shows that we need to check the entire GIT with bidigital palpation during the exploration to reveal any possible asymptomatic intussusceptions, to find other polyps, if any, and to excise them with enterotomy.

As seen in the case report of Çakmak et al.,[18] many patients with PJS have multiple operations histories due to recurrent intussusceptions. Repetitive operations and segmental resections in the GIT have been reported to cause short bowel syndrome in patients with PJS.[19] Laparoscopic surgery advantageously shortens post-operative recovery time in addition to leading to a very low rate of complications due to recurrent laparotomies such as intra-abdominal adhesions and incisional hernia.[20,21] Even if the patient is not diagnosed with PJS preoperatively, if an intussusception secondary to polyp in the GIT is confirmed during the operation, an enterotomy and polypectomy can be performed and a frozen study can be performed as reported by Çakmak et al.[18] If the result is consistent with PJ type polyp, the approach to the case will change. Combined endoscopy and surgery are recommended in these patients to prevent repetitive explorations and to reduce the risk of possible short bowel syndrome.[9] In our case presentation, a frozen study and pre-operative endoscopy could not be performed due to insufficient medical means. Only bidigital palpation could be performed to detect any other possible polyps.

Patients with PJS are reported to have an increased risk of malignancy compared to the normal population.[11,12] There is no evidence that neoplastic lesions in these patients are caused by the transformation of hamartomatous polyps.[12] The early diagnosis of PJS and performing a detailed screening program is recommended to prevent gastrointestinal complications and possible malignancies.[9,12]

In addition to annual general internal medicine, chest diseases, gynecology, or urology clinical examinations, the patients diagnosed with PJS are recommended to have complete blood count tests, liver function tests, specific blood tests, and chest radiography. It has been reported that capsule endoscopy should be performed every 3 years in these patients in addition to gastroscopy and colonoscopy.[12,14]

Conclusion

PJS should be included among the pathologies to be suspected in pre-diagnosis, and detailed medical history and clinical examination should be evaluated accordingly, especially in adult intussusception cases. If medical means are available, the patient determined to have a polyp in GIT should be studied frozen preoperatively. Considering that, there may be multiple polyps simultaneously and the associated risk of recurrent intussusception attacks in these patients, treatment with combined endoscopy and, if possible, laparoscopic surgery should be preferred.

The advantages of laparoscopic surgery are preventing intra-abdominal adhesions caused by repetitive abdominal operations and shortening the post-operative recovery time. Patients with PJS may need repetitive abdominal operations throughout their lives due to obstructive pathologies secondary to hamartomatous polyps. Considering this, we believe that laparoscopic surgery will decrease post-operative complications such as incisional hernia, short bowel syndrome, and intra-abdominal adhesion formation secondary to laparotomy.

Informed Consent: Written informed consent was obtained from the patient for the publication of the case report and the accompanying images.

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OLGU SUNUMU - *ÖZ*

İnce bağırsak intusepsiyonuna bağlı nadir bir mekanik bağırsak obtrüksiyon nedeni: "Peutz-Jeghers tipi soliter hamartomatöz polip"

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Peutz-Jeghers Sendromu (PJS), mukokutanöz membranlarda hiperpigmentasyon ve gastrointestinal sistemde hamartomatöz polipler ile kendini gösteren otozomal dominant geçişli, nadir bir hastalıktır. Hamartomatöz poliplere bağlı kanama ve mekanik intestinal obstrüksiyon bu hastalar için bildirilen başlıca komplikasyonlardır. Ayrıca PJS'li hastalarda artmış gastrointestinal ve gastrointestinal dışı malignite riski mevcuttur. Yirmi sekiz yaşında kadın hasta karın ağrısı ve kusma şikayetleri ile acil servise başvurdu. Karın muayenesinde distansion ve hassasiyet olan hastanın dudaklarında hiperpigmente lezyonlar görüldü. Karın muayenesinde geçirilmiş karın operasyonuna ait skar gözlenmedi. Ön tanıda mekanik intestinal obstrüksiyon düşünülen hastanın, karın tomografisinde distal jejunal anslarda intusepsiyon varlığı tespit edldi. Tanısal laparoskopide jejunal anslarda intusepsiyon gözlendi. Suprapubik yapılan minimal insizyondan sonra, ince bağırsak ansları aleksis içerisinden bidigital palpasyon ile kontrol edildi ve başka bir lümen içi kitle tespit edilmedi. Polibin bulunduğu intususepsiyon segmenti içine alan laparoskopik yardımlı jejunojejunal rezeksiyon ve anastomoz uygulandı. PJS'li hastalarda, hastalığın doğası gereği gastrointestinal sistemde aynı anda birden fazla polip olabilir ve bununla ilişkili olarak tekrarlayan intusepsiyon atakları riski vardır. Kısa bağırsak sendromu ve tekrarlayan laparotomilere bağlı karın içi yapışıklıkları önlemek için PJS'li hastalarda kombine endoskopi ve laparoskopik/laparoskopik yardımlı cerrahi tedavi tercih edilmelidir.

Anahtar sözcükler: Hamartomatöz polip; intusepsiyon; laparoskopik cerrahi; Peutz-Jeghers sendromu.

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