

Acute intussusception and polyp with malignant transformation in Peutz-Jeghers syndrome: A case report

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Abstract. Intussusception is one of the most frequent complications of Peutz-Jeghers syndrome, and has been well described in previous studies. More attention has been paid to malignancy, which is another complication of Peutz-Jeghers syndrome and which leads to increased mortality. Few cases of intussusception combined with malignant polyps in Peutz-Jeghers syndrome have been reported to date. In the present study, we report a case of intussusception and malignant polyps occurring in various parts of the small intestine in a 43-year-old male. In addition to repair of the intussusception and partial resection of the small intestine with malignant polyps, we also simultaneously performed polypectomy of as many polyps as possible without resection of the small intestine. Our aim is to make clinicians aware of intussusception and malignant polyps coexisting in Peutz-Jeghers syndrome when performing emergency surgery. Prophylaxis and polypectomy of the entire small bowel is an effective way to reduce the frequency of laparotomies in patients with this disease.

Introduction

Peutz-Jeghers syndrome is a rare autosomal dominant inherited disease characterized by mucocutaneous pigmentation and gastrointestinal polyps (1). It was first described by Peutz (2) in 1921 and Jeghers *et al* (3) in 1949. The incidence has been estimated to be between 1 in 8,300 and 1 in 200,000 live births (4). The polyps are located predominantly in the small intestine, and may cause various complications, including abdominal pain, anemia and acute intussusceptions (5). Patients with Peutz-Jeghers syndrome have an increased risk of developing cancer or transformation to malignant polyps in the gastrointestinal tract and

other organs (6). The coexistence of adult intussusception caused by Peutz-Jeghers syndrome and malignant polyps is rarely reported. Here, we report a case of intussusception and malignant polyps occurring in the small intestine in a 43-year-old male. The patient provided written informed consent to the publication of this case.

Case report

Patient presentation. A previously healthy 43-year-old male presented to the emergency room at Tianjin First Central Hospital, China, with a 3-day history of nausea, vomiting and increasing crampy abdominal pain. The patient had previously undergone resection of the small intestine for intussusception. He had no recent weight loss and no family history of bowel disease.

Upon examination, the patient had a tender distended abdomen with no peritoneal signs, fever or adenopathy. The patient's skin, including the perioral area, appeared normal, as did the oral mucosa. Clear evidence of intestinal peristalsis in the left upper abdomen was observed when the patient experienced abdominal pain, and an increase in bowel sounds was noted. Laboratory tests indicated normal hemoglobin levels, blood count and liver and renal function. An erect abdominal radiograph of the patient revealed air-fluid levels. Subsequent computed tomography (CT) imaging of the abdomen revealed features typical of intussusception (Fig. 1).

Surgery and diagnosis. At surgery, part of the proximal jejunum, 20 cm away from the Treitz ligament, was observed to be invaginated into the proximal jejunum, which determined the diagnosis of intussusception (Fig. 2A). A mass of ~3 cm could be felt at the lead point of the intussusception. Following the removal of the intussusception, this polyp was completely resected (Fig. 2B). There were numerous larger and smaller polyps identified by palpation and intermittent opening of the bowel which were also completely resected (Fig. 2C). The malignancy in the small intestine, 15 cm away from the ileocecal valve, involved the intestinal wall and appeared white and cauliflower-like. Partial resection of the small intestine with ileo-ileo anastomosis was performed (Fig. 3A). Through histopathological examination carried out following surgery, the mass was identified as a solitary Peutz-Jeghers polyp with malignant features (Fig. 3B).

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Figure 1. Computed tomography scan revealing the intussusception.

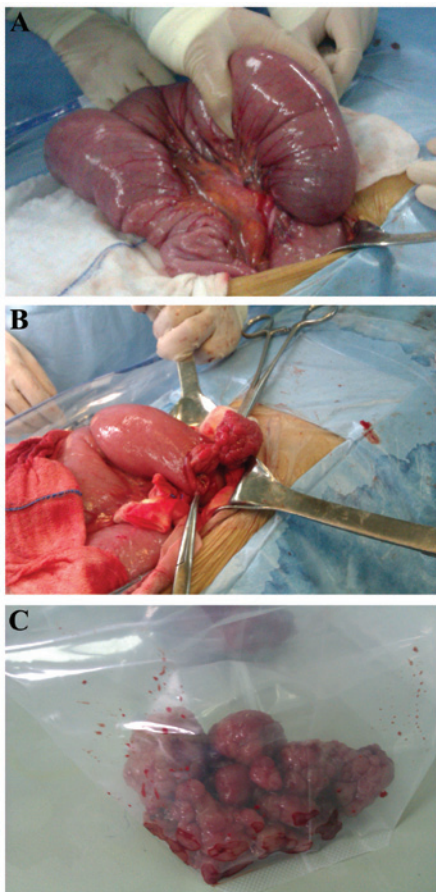


Figure 2. Surgical images. (A) Proximal jejunum intussusception. (B) Polyp that acted as a lead point for the intussusception. (C) Larger and smaller completely resected polyps.

Follow-up. The patient recovered with no further events and was discharged 10 days after surgery. He remained well 20 months later, and follow-up is ongoing.

Discussion

Peutz-Jeghers syndrome is a rare, autosomal dominant inherited disorder, which is characterized by mucocutaneous pigmentations, gastrointestinal polyposis and an increased risk of cancer (1). The sites most commonly affected

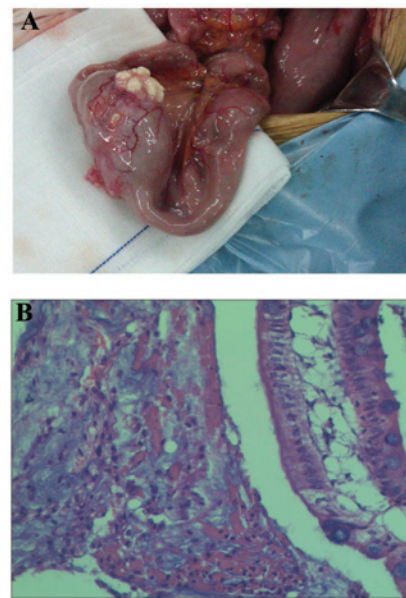


Figure 3. (A) The malignancy invaded the intestinal wall and was white and cauliflower-like. (B) Histopathological examination revealed polyps with malignant transformation invading the deep muscular layer outside the serosa in the small intestine (hematoxylin and eosin staining; original magnification, x200).

by Peutz-Jeghers polyps in the gastrointestinal tract are the jejunum, colorectal region, duodenum and stomach, in decreasing order of frequency. The most common histological type of polyps is hamartoma, which exhibits epithelial elements in the submucosa, muscularis propria and subserosa. The polyps often surround mucin-filled spaces. These hamartomas may induce significant complications including intussusception or gastrointestinal bleeding, requiring a number of laparotomies and bowel resections. During surgery for intestinal intussusception, it is preferable to simultaneously resect as many polyps as possible to prevent recurrence of the complications they cause (7).

Peutz-Jeghers syndrome patients are at a higher risk of cancer, and hence have an increased mortality rate. The incidence of tumors in patients with Peutz-Jeghers syndrome patients is 15 times higher than that observed in normal individuals, and the incidence of malignant tumors may be as high as 20% (8). Malignancy in Peutz-Jeghers syndrome is considered to arise in adulthood, and occurs only rarely in children (9). In addition to the hamartomas, malignant tumors of other organs outside the gastrointestinal tract also evolve into adenomas and carcinomas in a process occurring via *de novo* pathways (10). STK11/LKB1 (hereafter referred to as STK11) germline mutations account for the majority of cases of Peutz-Jeghers syndrome, whereas large deletions account for ~30% of cases (11). In addition, malignant polyps may be associated with interferon-induced transmembrane glycoprotein-1 (12), which may be an effective marker to test for polyps in the process of malignant transformation in Peutz-Jeghers syndrome. The main therapeutic treatment used for Peutz-Jeghers syndrome is non-steroidal anti-inflammatory drugs. Wei *et al* (13) also demonstrated that rapamycin inhibits the enzyme encoded by STK11, which inhibited the growth of hamartoma.

Indications for surgery include polyps with abdominal cramps, intestinal bleeding, intussusception, intestinal obstruction and malignant polyps confirmed by endoscopic biopsy. As a result of the risk of complications related to the hamartomatous polyps and the increased risk of cancer, prophylaxis and polypectomy of the entire small bowel are effective ways of reducing the frequency of laparotomies in patients with Peutz-Jeghers syndrome (14). The combination of surgical and endoscopic polypectomy is an efficient and practical choice in the management of polyps in patients with the disease (7). Radical surgery is required only if the polyps are confirmed to be malignant.

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