

CLINICAL NOTE

## Recurrent small intestine intussusception in a patient with Peutz-Jeghers syndrome

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### ABSTRACT

Peutz-Jeghers syndrome is a rare hereditary autosomal dominant disease caused by a mutation of the tumor suppressor gene serine/threonine kinase 11 located in chromosome 19p13.3. It is characterized by the presence of extensive mucocutaneous pigmentation, especially of the lips and the occurrence of hamartomatous polyps throughout the gastrointestinal tract. Gastrointestinal hamartomas occur predominantly in the small intestine and can become symptomatic leading usually to intestinal obstruction and abdominal pain. We present a case of recurrent intestinal obstruction caused by small bowel intussusception treated by reduction, enterotomy and polypectomy and followed by intraoperative enteroscopy and endoscopic polypectomy.

**Key words:** Enterotomy. Polypectomy. Hamartomatous polyps.

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### INTRODUCTION

Peutz-Jeghers syndrome is a rare hereditary autosomal dominant disease (1) with a reported incidence of 1 per 120,000 to 1 per 200,000 live births (1,2). It is characterized by the presence of extensive mucocutaneous pigmentation, especially of the lips in 94% of the patients and also buccal mucosa (66%), hands (74%) and feet (62%) (3) and the occurrence of hamartomatous polyps throughout the digestive

tract (1-3). Gastrointestinal hamartomas occur predominantly in the small intestine (78%), especially the jejunum, but also frequently in the colon (42%), stomach (38%) and rectum (28%) (1). These polyps develop during the first decade of life and can become symptomatic leading to intestinal obstruction (43%), abdominal pain (23%), rectal bleeding (14%) and anal extrusion of polyp (7%) (3,4). We present a case of recurrent intestinal obstruction caused by small bowel intussusception treated by reduction, enterotomy and polypectomy and followed by intraoperative enteroscopy and endoscopic polypectomy.

### CASE REPORT

A 19 year old male presented to the emergency department of our hospital with clinical symptoms and signs of intestinal obstruction. He had a 24 hour history of abdominal pain, nausea, vomiting and constipation. At the age of 9 he had developed intussusception for which he underwent small bowel resection and had then been diagnosed with Peutz-Jeghers syndrome. However he didn't follow the surveillance recommendations and had never had an upper endoscopy or a small bowel series. Clinical examination revealed an abdominal tenderness while laboratory examination showed microcytic, hypochromic anemia (Ht 30.3%, Hb 8.6 g/dL) while all other exams was within the normal limits. The abdominal X-ray revealed multiple air fluid levels, indicative of small intestine obstruction and the abdominal CT demonstrated the small intestine intussusception.

An exploratory laparotomy was performed which confirmed the small bowel intussusception about 170 cm from Treitz ligament (Fig. 1). The intussusception was reduced and an enterotomy was carried out which revealed a large polyp measuring 8 x 5 x 3 cm which was resected (Fig. 2). Through the enterotomy, an intraoperative enteroscopy was performed which allowed the visualization of the small intestine until the ileocecal valve and the polypectomy of 6 additional pedunculated polyps, measuring from 0.5 cm to

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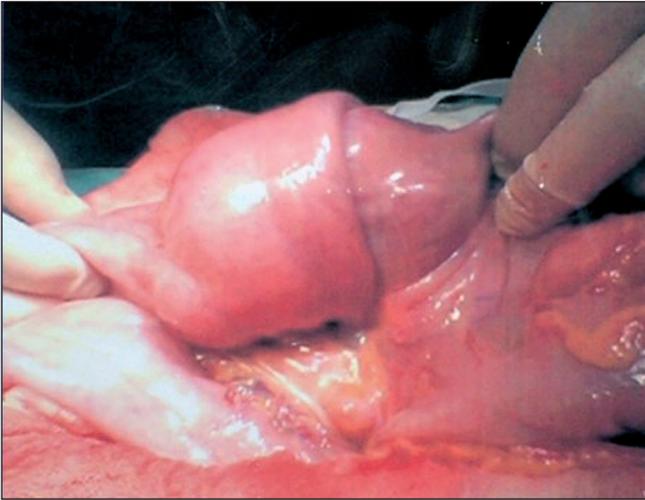


Fig. 1. Intussusception of the small intestine.

1.5 cm (Fig. 3). Histopathological examination showed that the polyps consisted of branching thick smooth muscle bundles arising from the muscularis mucosa and were covered by intestinal epithelial cells and glands. Histopathological findings were consistent with hamartomatous polyps (Fig. 4).

The postoperative course was uneventful and the patient was discharged on day six.

### DISCUSSION

PJS is caused by a mutation of the tumor suppressor gene serine/threonine kinase 11 (STK11 also known as LKB1), located in chromosome 19p13.3 (1,3). The average age of diagnosis is 25.2 years (1). The diagnosis is based on the presence of two of the following criteria: characteristic melanin pigmentation, family history of the syndrome and small bowel polyposis along with the presence of hamartomatous polyp

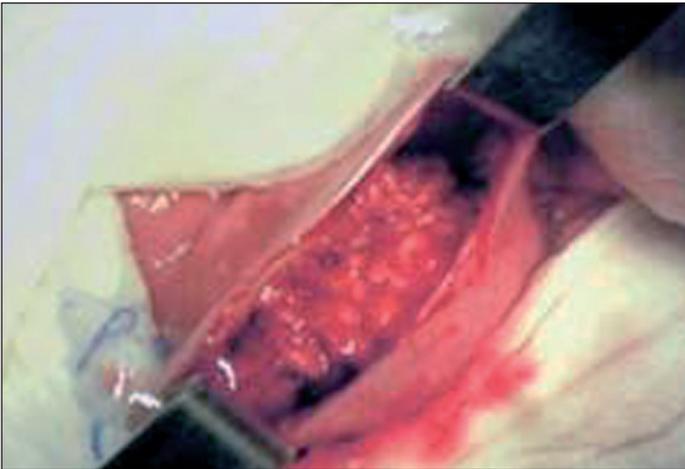


Fig. 2. Enterotomy of the small intestine revealing the large polyp.



Fig. 3. Intraoperative enteroscopy of the small intestine.

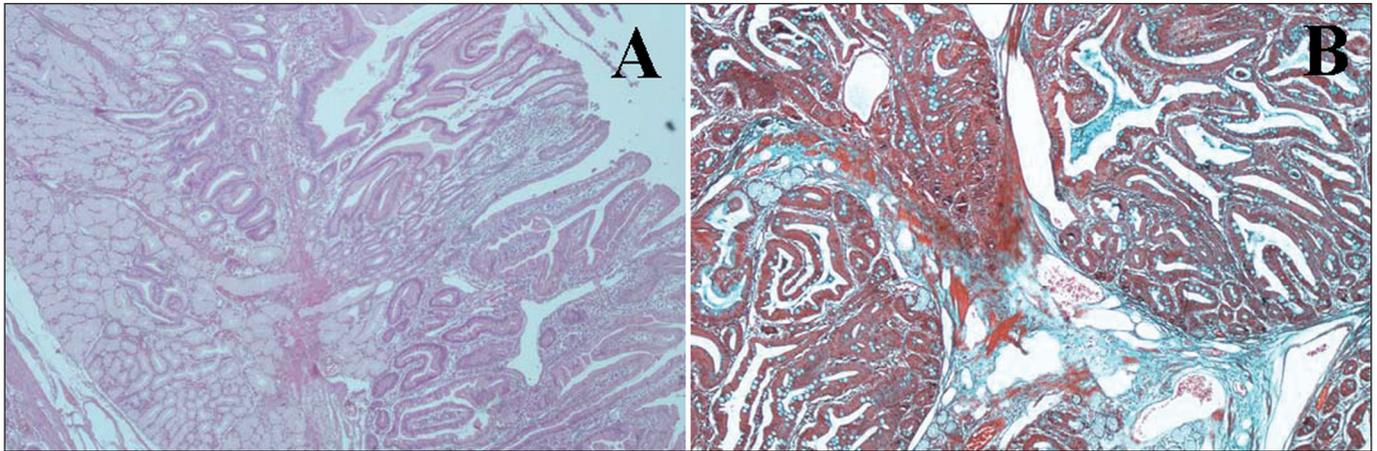


Fig. 4. A. The hamartomatous polyp consisted of branching thick smooth muscle bundles covered by normal intestinal epithelial cells and glands (H&E X20). B. Branching framework of smooth muscles (Masson's trichrome X20).

or polyps (3). The melanotic pigmented lesions are usually less than 5 mm in diameter, dark brown or blue brown and are present in more than 95% of the patients (2,3). The hamartomatous polyps in PJS have unique histopathologic characteristics. They are characterized by hypertrophied smooth muscle bands with an intermingling epithelial and glandular component, a frond-like structure and appropriate epithelium for each part of the digestive tract (2,3). They consist of a branching framework of smooth muscle cells that extend into the epithelial layer (2,3). The polyps vary in size from 0.1 to 5 cm in diameter and usually have a long thick stalk (3,5), but in the present case the polyp was measuring 8 cm in the maximum diameter, a rather large polyp.

In PJS the polyps grow in a very young age and the majority of patients become symptomatic during the first three decades of life (2,3). The most common clinical presentation is obstruction and occult gastrointestinal bleeding (2). Intussusception is the most frequent complication (47%) and in 95% of the cases it occurs in the small intestine (3). Up to one third of the patients will require surgery within the first decade of life (2) and recurrence of intussusception occurs in at least 10% of cases (1, 4), much like the patient in the present case report who also suffered from anemia. The risk for intussusceptions is not influenced by the STK11 mutation status (6).

Intussusception can be managed successfully with invasive approaches. Intussusception should be reduced and followed by polypectomy (1,2). Because of the recurrence rate small bowel resection should be avoided unless absolutely necessary (2,4) in order to avoid short bowel syndrome and malabsorption (5). Also during laparotomy, the rest of the intestine should be examined and the polyps should be removed either by enterotomies or preferably by intraoperative endoscopic polypectomy (2), even those measuring less than 1 cm. This attempt to clear the small intestine of polyps, named "clean sweep", appears to decrease the need for recurrent small bowel surgery (3). Laparoscopic intussusception reduction and laparoscopic as-

sisted enteroscopy has been also used successfully and offers a less invasive approach (1).

PJS is associated with a 2-10% increased risk of gastrointestinal cancer (1) and there seems to be a small but real risk of malignant transformation of hamartomas (2,4). Besides this risk, polypectomy is recommended for polyps in the stomach and colon greater than 1 cm as they can become symptomatic (1-3). Also surgery has been recommended for small bowel polyps greater than 1-1.5 cm and symptomatic or rapidly growing polyps (3). Surveillance includes an upper endoscopy and barium study every 2 years and colonoscopy every 3 years. Barium studies may be replaced by wireless capsule endoscopy and double balloon endoscopy (5).

Except for the increased risk of intestinal malignancies, patients with PJS have also an increased risk of extraintestinal malignancies including mainly the breast, ovaries, uterus, cervix, testicles and pancreas (5) and less commonly the fallopian tubes, thyroid, lungs, gallbladder and bile ducts (1). Periodical screening for these malignancies is recommended (1-5).

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