

# Peutz-Jeghers Syndrome Complicated by Intestinal Intussusception: A Case Report

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**Abstract:** Peutz-Jeghers Syndrome (PJS) is a rare genetic condition characterized by the presence of hamartomatous polyps in the gastrointestinal tract and mucocutaneous hyperpigmentation. Intussusception is a relatively common complication in these patients, although progression to extensive intestinal necrosis is uncommon and potentially fatal. This report describes a 21-year-old woman with PJS diagnosed in childhood, admitted with acute abdominal pain, hyporexia, and vomiting. Computed tomography revealed extensive jejuno-ileal intussusception, and emergency laparotomy identified 90 cm of invaginated and necrotic small bowel caused by a hamartomatous polyp in the mid-jejunum. Segmental enterectomy with primary side-to-side anastomosis was performed, and the patient had a satisfactory postoperative recovery, being discharged on the fourth day. She remains under outpatient follow-up without signs of short bowel syndrome. This case highlights the importance of structured endoscopic surveillance and early surgical intervention in PJS, especially in settings where advanced enteroscopy techniques are not available.

**Keywords:** Peutz-Jeghers Syndrome; Polyps; Intussusception; Laparotomy.



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## 1. Introduction

Intestinal intussusception consists of the invagination of a proximal segment of the intestine into a distal one, resulting in luminal obstruction, vascular compromise, and potentially necrosis or perforation of the intestinal wall [1]. Although it is a common cause of acute abdomen in children, it is a rare condition in adults and is generally associated with a secondary etiology such as tumors, polyps, or structural abnormalities of the intestinal wall [1,2]. Among the causes of intussusception in adults, Peutz-Jeghers Syndrome (PJS) stands out. PJS is a rare autosomal dominant genetic disorder caused by mutations in the *STK11/LKB1* gene, which encodes a protein with tumor-suppressor function [1,2]. The syndrome is characterized by the presence of multiple hamartomatous polyps distributed throughout the gastrointestinal tract and by distinctive mucocutaneous hyperpigmentation, especially on the lips and oral mucosa [3-5].

Hamartomatous polyps act as lead points for recurrent episodes of intestinal intussusception, which is the most frequent complication and one of the main causes of morbidity in these patients [2,6]. Early recognition of this manifestation is essential, as intussusception may progress to ischemia and intestinal necrosis, potentially requiring multiple resections throughout life [9-11]. This report describes the case of a patient with PJS who developed intestinal intussusception complicated by extensive jejunal necrosis, highlighting not only the potential severity of this complication but also the direct impact of

incomplete small-bowel surveillance. The case reinforces the need for early recognition, access to appropriate diagnostic methods, and timely surgical intervention to prevent severe outcomes in patients with PJS. The aim of this report is to describe a severe presentation of intestinal intussusception in a young patient with PJS, discussing diagnostic and therapeutic challenges in settings where advanced endoscopic methods are unavailable.

## 2. Case Report

The patient is a 21-year-old woman diagnosed with Peutz-Jeghers Syndrome (PJS) at 1 year of age. She has a positive family history, with both her father and sister affected by the same syndrome. She was admitted to the Referral Unit of the Clinical Hospital of the Federal University of Paraná (HC-UFPR) presenting with upper abdominal and left flank pain of continuous character, associated with hyporexia and episodes of vomiting, without relieving or aggravating factors, and no changes in bowel habits.

She had no prior surgical history but had a known history of gastric and intestinal hamartomatous polyposis, involving the duodenum, jejunum, descending colon, and sigmoid colon, documented through previous Magnetic Resonance Imaging, Upper Digestive Endoscopy (UDE), and colonoscopy, all with histopathological confirmation. These intestinal polyps had been resected one year prior to the current presentation, except for one polyp measuring approximately 12 mm located in the jejunum, which remained due to inaccessibility with the endoscopic methods available at the institution. Advanced techniques such as device-assisted enteroscopy (double-balloon or single-balloon enteroscopy) were not available, preventing prophylactic removal as recommended by international guidelines.

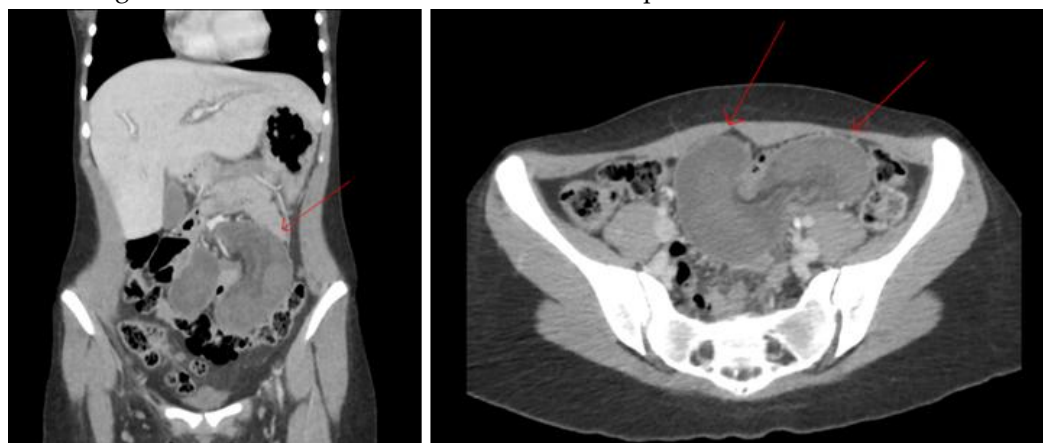
On physical examination, the patient was in good general condition, hemodynamically stable, and breathing comfortably on room air, without signs of respiratory distress. Melanotic macules characteristic of PJS were observed on the lips and perioral region (Figure 1). The abdomen was flat, with present bowel sounds, normal tympany, and tenderness on deep palpation in the epigastrium, left hypochondrium, and left flank, without palpable masses or visceromegaly and with no signs of peritonitis.

**Figure 1.** Melanotic macules in the labial and perioral region, a typical finding in Peutz-Jeghers Syndrome. The lesions are flat, brownish-gray in color, and well circumscribed, representing a classic cutaneous marker of the syndrome.



A contrast-enhanced abdominal computed tomography scan (Figure 2) was performed and demonstrated segmental distension of a jeuno-ileal small-bowel loop in the left flank, measuring up to 37 mm in diameter and approximately 96 cm in length, with mesenteric invagination compatible with intussusception. There was no evidence of pneumatosis intestinalis or pneumoperitoneum, and only a minimal amount of free fluid was observed in the pelvis, associated with mesenteric fat stranding. Emergency laparotomy was indicated due to the extent of the affected segment and the suspicion of intestinal compromise.

**Figure 2.** Contrast-enhanced abdominal computed tomography demonstrating mesenteric invagination consistent with intestinal intussusception.

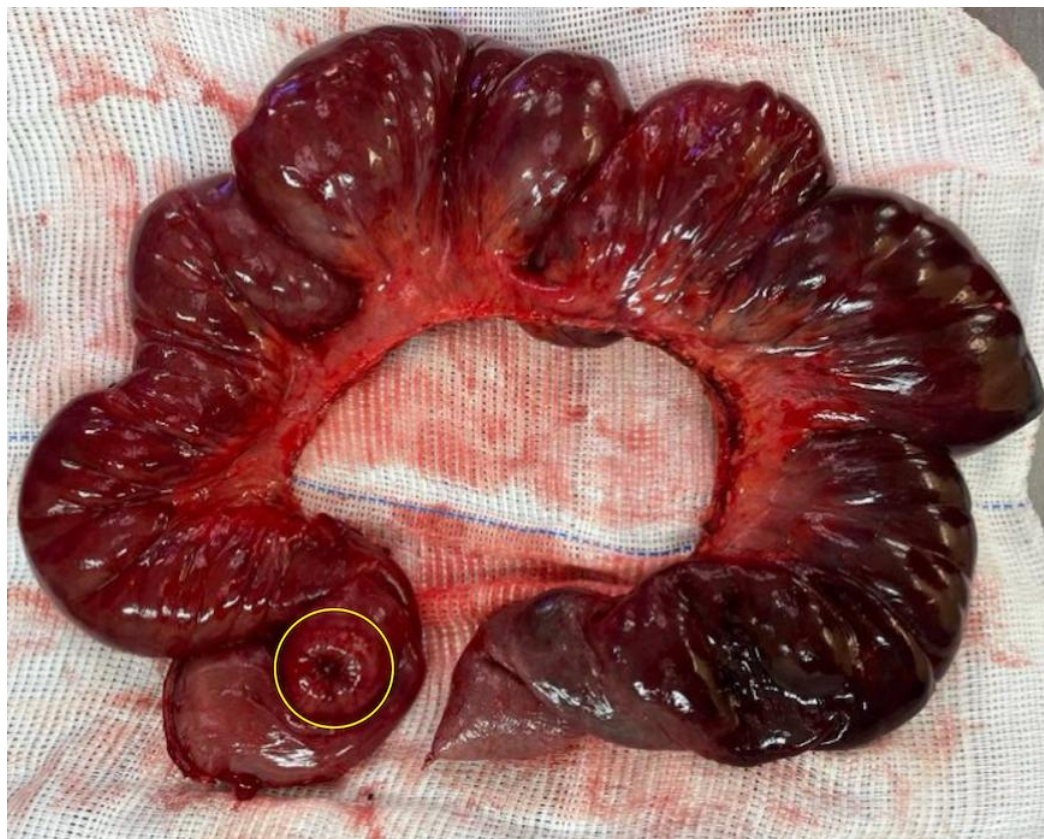


During the intraoperative procedure, under general anesthesia and through a transumbilical incision, an exploration of the abdominal cavity was performed. A segment of ischemic small bowel was identified approximately 100 cm from the ligament of Treitz, along with a small amount of free fluid in the cavity and no apparent perforations. Careful traction of the ischemic loop revealed an intussusception involving approximately 90 cm of small bowel, with its lead point corresponding to a polyp located in the mid-jejunum (Figure 3). Given the absence of tissue viability, cautious ligation of the mesenteric vessels supplying the necrotic segment was performed, followed by enterectomy of the devitalized bowel using a linear stapler (Figure 4).

**Figure 3.** Intraoperative appearance of the small-bowel loops. Intussusception in the mid-jejunum is observed, with a polyp serving as the lead point.



**Figure 4.** Surgical enterectomy specimen showing a segment of small intestine with transmural necrosis and a hamartomatous polyp (yellow circle) in the mid-jejunum, measuring approximately 12 mm in diameter.



A primary side-to-side isoperistaltic enteroenterostomy was then constructed using a linear stapler, followed by continuous oversewing to reinforce the staple line and complete the anastomosis with 3-0 Vicryl. This technique was chosen because it provides a larger functional lumen and a lower risk of stenosis. Subsequently, the mesenteric defect was closed, and the small-bowel loops were inspected, with no additional lesions identified. The final measurement of the small intestine was approximately 210 cm, with 110 cm from the ligament of Treitz to the anastomosis and 110 cm distal to the anastomosis.

The patient was transferred to the ward in the immediate postoperative period, where she evolved without significant clinical complications. She reported incisional pain consistent with the postoperative course, which was adequately controlled with opioids. Gradual progression of diet was implemented according to tolerance, along with early ambulation and motor and respiratory physiotherapy. She was discharged on the fourth postoperative day in good general condition. Histopathological analysis of the enterectomy specimen revealed transmural intestinal infarction and an infarcted hamartomatous polyp with features consistent with intestinal intussusception. Despite the extensive resection, the patient remains without symptoms of malabsorption or short bowel syndrome. She currently undergoes annual endoscopic surveillance and magnetic resonance enterography, with no recurrence of symptoms or complications, although new hamartomatous polyps requiring endoscopic resection have been identified.

### 3. Discussion

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant genetic condition predominantly caused by mutations in the *STK11/LKB1* gene, a tumor suppressor that plays an essential role in the p53-dependent apoptosis pathway [7,8]. Clinically, it is characterized by the presence of hamartomatous polyps distributed throughout the gastrointestinal

tract, except the esophagus, and by distinctive mucocutaneous melanotic macules. In addition, these patients have a significantly increased risk of both gastrointestinal and extraintestinal malignancies [3-5].

The clinical diagnosis can be established when any of the following criteria are present: two or more typical hamartomatous polyps; characteristic mucocutaneous hyperpigmentation in an individual with a positive family history; a Peutz-Jeghers polyp in a patient with an affected first-degree relative; or the combination of polyps and typical pigmentation in the same individual [3,4]. Mucocutaneous pigmented macules, present in more than 95% of patients, result from the accumulation of pigmented macrophages in the dermis. These lesions are typically flat, brown to blue-gray in color, and measure between 1 and 5 mm in diameter. The most frequently affected regions include the lips and perioral area (94%), palms (74%), oral mucosa (66%), and soles of the feet (62%). Although they serve as an important clinical marker, they have no malignant potential [3,4].

Regarding hamartomatous polyps, the most affected site is the small intestine (60–90%), particularly the jejunum [1]. However, they may occur throughout the entire gastrointestinal tract. Although half of patients with PJS are asymptomatic at diagnosis, the main causes of morbidity and mortality usually occur in the second decade of life and may be related to the presence of polyps, such as obstruction caused by intussusception or luminal occlusion, abdominal pain due to infarction, hematochezia, anemia from acute or chronic bleeding, colonic polyp prolapse, and malignancy [2,9].

Approximately 70% of patients experience intussusception at some point, most frequently in the small intestine [6,10]. Episodes may be transient and manifest as recurrent colicky abdominal pain. Acute complications, such as intussusception with extensive necrosis, are uncommon and represent a significant diagnostic and therapeutic challenge [11-13]. The present case is notable for illustrating a severe and rare presentation of this syndrome in a young patient, underscoring the importance of early diagnosis and timely surgical intervention.

Initial symptoms of intussusception, abdominal pain, vomiting, and distension, are nonspecific, which often delays diagnosis. This is particularly true in patients with PJS, who commonly experience intermittent abdominal pain secondary to subocclusive obstructions that frequently resolve spontaneously [14]. International guidelines recommend that every episode of acute abdominal pain in patients with PJS be urgently evaluated by a surgical team due to the high probability of intussusception. Contrast-enhanced computed tomography is the diagnostic method of choice, as it allows confirmation of intussusception, identification of signs of intestinal ischemia, and appropriate therapeutic planning [15,16].

In this case report, the presence of clinical and radiological signs of vascular compromise justified the need for immediate laparotomy. Radiologic or endoscopic reduction of intussusception is not indicated when intestinal obstruction is secondary to a symptomatic hamartomatous polyp. Furthermore, PJS is associated with an increased risk of both gastrointestinal and extraintestinal malignancies. The most common sites of cancer include the colon, small intestine, stomach, pancreas, and breast [18,19]. For this reason, surveillance of the stomach, small intestine, and colon is strongly recommended. Periodic endoscopic surveillance is essential to reduce morbidity and mortality. American and European guidelines recommend upper gastrointestinal endoscopy (UGIE) and complete colonoscopy starting at 8 years of age, with repeat examinations every 1–3 years if polyps are present. In the absence of lesions, the next evaluation should be performed at 18 years of age, and after 50 years, surveillance should occur at 1–2-year intervals due to the markedly increased risk of malignancy [12, 14, 20, 21].

For the small intestine, surveillance methods include small-bowel capsule endoscopy (SBCE), balloon-assisted enteroscopy (BAE), magnetic resonance enterography (MRE), and computed tomography enterography (CTE). The first examination should be performed at 8 years of age or earlier if symptoms are present, with follow-up every 1–3 years when polyps are identified [2, 12, 14, 20].

Management of PJS focuses on treating complications related to polyps. Interventions include endoscopic polypectomy, segmental enterectomy, or oncologic resection when malignancy is present. Aggressive procedures should be avoided due to the diffuse nature of the disease [19, 20]. According to the US Multi-Society Task Force on Colorectal Cancer and the National Comprehensive Cancer Network (NCCN), endoscopic resection of polyps is recommended whenever possible. Endoscopic polypectomy is indicated for polyps  $\geq 5$  mm identified on UGIE and colonoscopy, whereas small-bowel polyps  $\geq 10$  mm or symptomatic lesions should also be removed to prevent complications. Surgery is reserved for cases in which endoscopic control is unfeasible (large, multiple, or neoplastic polyps) or in the presence of obstruction or intestinal intussusception. When laparotomy is required, intraoperative enteroscopy is recommended to identify and remove residual polyps [12, 19, 20].

In this case, the failure to achieve endoscopic removal of a polyp accessible by device-assisted enteroscopy represents a potentially avoidable gap in care, highlighting disparities in access to advanced techniques within the Brazilian public health system. This report reinforces the need for a high index of suspicion for intussusception in patients with PJS, even when symptoms present atypically or with low specificity. It also underscores that early surgical intervention is essential to prevent potentially fatal outcomes, including extensive necrosis, short bowel syndrome, sepsis, and death—particularly in settings where small-bowel endoscopic surveillance was limited by the unavailability of advanced techniques.

#### 4. Conclusion

Intestinal intussusception in patients with Peutz–Jeghers syndrome represents a potentially severe complication, whose unfavorable progression can be mitigated by early clinical recognition, timely radiological diagnosis, and prompt surgical intervention. The present case demonstrates that the inability to perform prior resection of a jejunal polyp, due to the unavailability of advanced enteroscopic techniques, contributed to the occurrence of the acute event.

Thus, the importance of comprehensive and technically adequate endoscopic surveillance programs, as well as prophylactic resection of larger polyps, is emphasized to reduce the risk of obstructive or ischemic complications. Structured follow-up measures are essential to optimize prognosis and minimize the need for emergency interventions throughout these patients' lives. In this context, the present case illustrates how the lack of access to advanced enteroscopy can result in severe surgical complication, representing an important marker of healthcare inequity in rare diseases.

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**Conflicts of Interest:** All other authors declare no conflicts of interest.

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