Radiology Case Reports 20 (2025) 2445–2449

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Peutz-Jeghers syndrome revealed by CT finding of acute small bowel intussusception: A case report ☆

Paola Milillo, MD, PhD, Gianluca Prencipe, MD, Stefano Piscone, MD, Sara Saccone, MD, Luca Cappucci, MD, Maria Grazia Scarale, MD, Roberta Vinci, MD, Luca Macarini, MD, Luca Pio Stoppino, MD, PhD*

Department of Medical and Surgical Sciences, Section of Diagnostic Imaging, University of Foggia, Viale Luigi Pinto n. 1, 71122 Foggia, Italy

ARTICLE INFO

Article history:
Received 16 January 2025
Revised 9 February 2025
Accepted 10 February 2025

Keywords:
Peutz-Jeghers Syndrome
Intussusception
Small bowel disease
Computed tomography
Emergency radiology
Case report

ABSTRACT

Peutz-Jeghers syndrome (PJS) is a rare genetic disorder characterized by multiple gastrointestinal hamartomatous polyps and distinctive mucocutaneous pigmentations. Intussusception is a significant complication associated with Peutz-Jeghers syndrome and it can lead to potential bowel obstruction and ischemia. We present a case report in which PJS was first suspected by a radiologist after performing abdomen Computed Tomography (CT) at the emergency department. We will discuss the clinical presentation, radiological findings and surgical management in a case of acute small bowel intussusception.

Introduction

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by the development of multiple hamartomatous polyps in the gastrointestinal tract and distinctive mucocutaneous pigmentation [1].

One of the most severe and common complications associated with PJS is intussusception, where a segment of the intestine invaginates into an adjacent segment, leading to potential bowel obstruction and ischemia [2].

Computed Tomography (CT) finding of multiple intestinal polyps, with or without associated intussusception, should prompt consideration of hereditary gastrointestinal polyposis syndrome as a possible diagnosis [3].

This article aims to discuss the clinical presentation, radiological findings, and management of intussusception in a young patient with unknown PJS, focusing on CT imaging.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

* Corresponding author.
E-mail address: stefano.piscone@unifg.it (S. Piscone).

https://doi.org/10.1016/j.radcr.2025.02.041

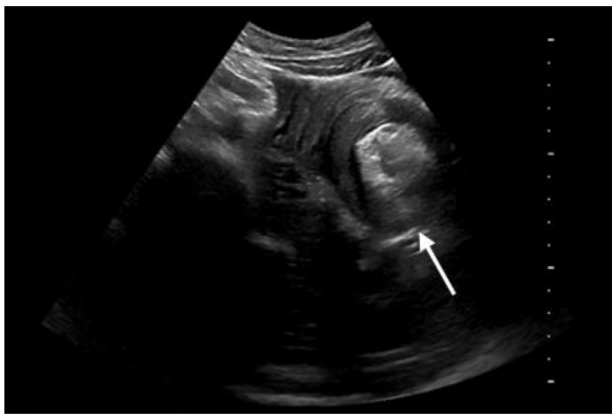


Fig. 1 – Abdominal ultrasound image revealed the "target sign": a characteristic concentric ring appearance where alternating hypoechoic and hyperechoic layers resemble a bullseye or target (long arrow).

Case presentation

A 19-year-old male with no significant past medical history presented to the emergency department with an acute onset of severe abdominal pain. The patient reported that the pain began suddenly and described it as colicky, progressively worsening over the preceding 24 hours. Associated symptoms included persistent nausea, several episodes of vomiting, and a noticeable decrease in appetite.

On physical examination, the abdomen was diffusely tender to palpation, with notable rebound tenderness in the mid-lower left quadrants. Abdominal distension was also observed. The patient's vital signs were normal, though he exhibited a slightly elevated heart rate and a mild fever.

Initial laboratory tests revealed only a mild leukocytosis, while other routine blood tests, including electrolytes and liver function tests, were normal.

An urgent abdominal ultrasound was performed, which showed no significant abnormalities in the major abdominal organs. However, in the left middle quadrant, a suspicious finding was noted: the characteristic "target sign" (or "bull's-eye" sign) was visible. This finding is highly suggestive of intussusception, where concentric alternating echogenic and hypoechoic rings represent the bowel walls telescoping into each other (Fig. 1).

Given the limitations of ultrasound, particularly in evaluating small bowel pathology, the patient underwent a CT scan of the abdomen and pelvis with intravenous contrast.

The CT imaging revealed multiple well-defined polyps scattered throughout the stomach and the small intestine, ranging in size from 0.5 to 4 cm in diameter. The lead point of the intussusception was identified as a large polyp, approximately 4 cm in diameter, at the junction of the involved bowel segments (Fig. 2).

The affected bowel segments displayed a characteristic "double-layer" or "double-wall" sign, further confirming the diagnosis of bowel wall invagination (Fig. 3).

Further radiological features included the "mesenteric vessels convergence sign", where mesenteric vessels were drawn into the intussuscepted bowel, forming a swirling pattern (Fig. 3).

Contrast enhancement of the polyp's soft tissue component was also observed, emphasizing its vascularity, a typical feature of hamartomatous polyps that are often hypervascular (Figs. 2-3).

Additionally, the CT scan demonstrated moderate free fluid accumulation in the pelvic cavity, likely indicative of reactive peritoneal fluid secondary to bowel obstruction (Fig. 3).

Physical examination revealed characteristic perioral pigmentation, adding to the suspicion of PJS. The combination of imaging findings and clinical features strongly suggest this diagnosis.

The patient underwent urgent laparotomy due to acute intussusception. Intraoperatively, the involved intestinal loop was identified, and a segmental resection of the affected bowel was performed. The resected segment contained the large polyp, confirming its role as the lead point for the intussusception, thus corroborating the preoperative diagnosis.

A thorough manual examination of the remaining small intestine was performed to check for additional polyps. This exploration revealed multiple small to medium-sized polyps, consistent with the preoperative CT findings. These polyps were not causing obstruction or intussusception at the time of surgery, and thus no further bowel resections were deemed necessary.

The surgical team opted for conservative management, focusing on resolving the acute pathology while preserving as much healthy bowel segment as possible. The decision to avoid further resections was based on the size and distribution of the polyps, as well as the potential risk of short bowel syndrome from extensive resections.

Histopathological analysis of the resected tissue confirmed the presence of hamartomatous polyps, consistent with PJS.

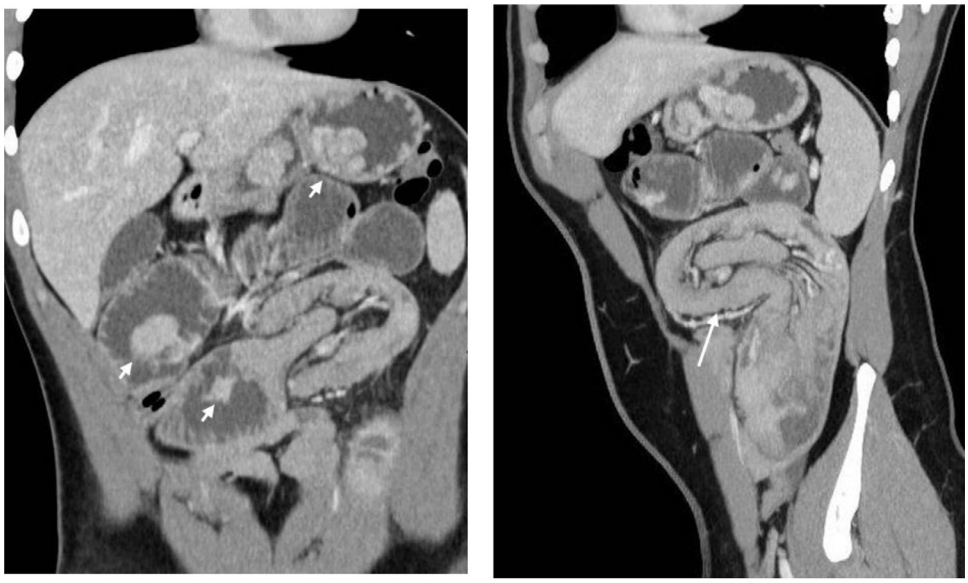
Postoperatively, the patient was closely monitored and recovered without complications. His bowel function gradually returned, and he was able to resume oral intake within a few days.

The patient and his family were counseled on the implications of PJS, including the need for genetic counseling, regular endoscopic surveillance for polyp development, and active monitoring for the increased risk of malignancy associated with the syndrome.

Discussion

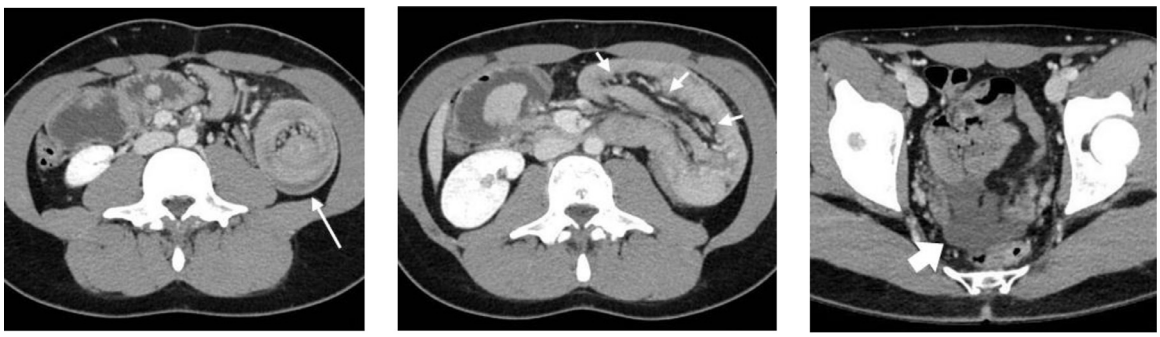
PJS is an extremely rare autosomal dominant disorder with an incidence of 1 in 12–30,000 live births [4]. This condition was first described by Peutz in 1921 and Jeghers in 1944 and it caused by a mutation in the tumor suppressor gene STK11 on chromosome 19p13, which encodes a serine–threonine kinase involved in several cellular processes, including the regulation of the mTOR pathway and chromatin remodeling [1,5].

Despite its rarity, PJS has become well known for its distinctive development of hamartomatous polyps throughout the gastrointestinal tract, predominantly in the small intestine (96%), colorectum (25%-50%), and stomach (25%). Addi-



(A) (B)

Fig. 2 – (A) Coronal CT image demonstrates multiple intestinal polyps throughout the stomach and small intestine; CT image shows also a segment of jejunum invaginating into an adjacent segment; image demonstrates multiple intestinal polyps throughout the stomach and small intestine (short arrows). (B) Sagittal CT reformatted image demonstrates the "sausage-shaped" or "reniform" mass (long arrow).



(A) (B) (C)

Fig. 3 – (A) The "target sign" on axial CT image with a typical "double-wall" appearance (long arrow); (B) The characteristic central low-attenuation fat and mesenteric vessels of intussusception (short arrows); (C) Free fluid in the pelvic cavity (fat arrow).

tionally, individuals with PJS often exhibit characteristic mucocutaneous pigmentation in the perioral region [1].

Beggs et al. proposed the following diagnostic criteria for PJS:

- Two or more histologically confirmed PJS polyps.
- Any number of PJS polyps in an individual with a family history of PJS.
- Characteristic mucocutaneous pigmentation in an individual with a family history of PJS.
- Any number of PJS polyps and a characteristic mucocutaneous pigmentation in the same individual [6].

The diagnosis of PJS in our patient was made according to the fourth Beggs' criterion, as he exhibited multiple gastrointestinal PJS polyps associated with pigmented macules in the perioral region.

PJS polyps possess distinctive histological features: they are hamartomatous polyps with a microscopic appearance of a tree-like branching smooth muscle fibers originating from the muscularis mucosa and extending into the polyp, which is covered by normal mucosal tissue. The size of these polyps can vary widely, generally ranging from 0.1 to 5 cm in diameter [7,8].

The 2 most significant complications of PJS are small bowel intussusception, which can present as an urgent and potentially life-threatening issue, particularly in children, and the elevated risk of cancer development in adulthood [4].

Individuals with PJS frequently present with colicky abdominal pain, abdominal discomfort, or sudden and severe

abdominal pain that can be either localized or diffuse. This is often accompanied by additional symptoms such as nausea, vomiting and lower gastrointestinal bleeding. These symptoms are typically due to large small-bowel polyps, which can lead to intussusception with severe consequences, including small bowel obstruction, ischemia, perforation, ulceration, and bleeding [6].

Our patient presented to the emergency department with acute small bowel intussusception, a manifestation that often leads to the discovery of PJS in children [9].

In PJS small bowel intussusception is a frequent occurrence, with a cumulative risk of 15% by age 10% and 50% by age 20, as reported in a multicenter study by Van Lier et al. [2]. The preferred site for intussusception is the jejunum, as in our case, followed by the ileum and the duodenum [10].

Imaging plays a pivotal role in these patients. Investigations such as esophagogastroduodenoscopy, colonoscopy, CT, MRI enterography, or video capsule endoscopy should be regularly employed to detect large PJS polyps, which is crucial for preventing complications and improving outcomes [7,10].

The European Society of Gastrointestinal Endoscopy (ESGE) recommends gastrointestinal surveillance every 3 years, beginning at age 8 for asymptomatic PJS patients, or earlier if symptoms are present [11].

In emergency settings, X-ray, Ultrasound (US) and Computed Tomography (CT) are common techniques used to identify small bowel intussusception and more severe complications related to PJS. Plain abdominal X-ray has limitations in detecting obstruction or pneumoperitoneum [10].

Ultrasonography is noted for its accuracy, especially in symptomatic PJS cases. On US examination, polyps appear as iso- to hyper-echoic intraluminal masses, with characteristic findings such as the "target" or "doughnut" appearance in transverse views and the "pseudo-kidney" sign in longitudinal views [5]. However, challenges such as bowel distension, obesity, or the need for skilled radiologists often necessitate abdominal CT, which is widely available and considered a routine imaging technique for determining the etiology of abdominal pain.

On CT scans, intussusception is identified by a "bowel-within-bowel" configuration, which appears as a complex soft-tissue mass consisting of an inner intussusceptum and an outer intussuscipiens. This mass is characterized by a crescent-shaped attenuation of mesenteric fat, which delineates the intussusception and contributes to the distinctive 3-layered appearance; enhancing vessels within the mesenteric fat are often visible [5,12,13]. Characteristic radiological signs of intussusception on CT include the "doughnut", “target”, or “bulls-eye” sign, seen on images where the plane is perpendicular to the longitudinal axis of the intussusception. This appearance represents concentric rings of bowel and mesentery. Another typical finding is the "sausage-shaped" or “reniform” mass, visible on images where the plane is parallel to the longitudinal axis of the intussusception, which indicates the telescoping of one bowel segment into another [5,10,14,15]. CT imaging may also reveal multiple hamartomatous polyps throughout the gastrointestinal tract, which serve as lead points for intussusception. These polyps typically appear as well-defined, homogeneous soft-tissue masses of varying sizes and may show enhancement following contrast administration, aiding in their identification [5,7]. The lead-point polyp may be observed at the apex of a small bowel intussusception, with the loss of the classic 3-layered structure [10,12,13]. CT is also useful in detecting potential complications of intussusception such as bowel ischemia, obstruction, or perforation. In cases of bowel ischemia, CT may show signs of bowel wall thickening, submucosal edema, and reduced enhancement, indicating compromised blood flow. In bowel obstruction, proximal bowel loops may be dilated with air-fluid levels, and the presence of free intraperitoneal air suggest bowel perforation [13].

Magnetic Resonance Imaging (MRI) provides similar diagnostic results compared to CT, but it is less commonly used in emergency settings and is primarily reserved for routine monitoring of PJS polyps or asymptomatic intussusceptions. On MRI, polyps appear as T2-isointense intraluminal masses, with homogeneous enhancement postcontrast administration [7,10,14].

M.G.F. van Lier et al. [2] analyzed 128 intussusceptions in 76 patients: among them, only 8.5% (11 of 128) were diagnosed via radiological imaging report (including X-ray, US and CT), while the remaining diagnosis of intussusception were primarily made intraoperatively (107 of 128). Nowadays, CT is considered the most accurate preoperative diagnostic tool, with an accuracy of approximately 83% [3].

Intussusception in PJS requires a multidisciplinary approach, beginning with patient stabilization, fluid resuscitation, and nasogastric decompression. Nonoperative reduction may be attempted, though surgery is frequently necessary, particularly in the presence of complications such as bowel ischemia, perforation, or unsuccessful nonoperative reduction [10].

In a large prospective cohort study, M.G.F. van Lier et al. [2] reported that up to 92.5% of patients with PJS had undergone emergency laparotomy for one of the major complications of intussusception. Surgical treatment typically involves resection of the affected bowel segment and removal of polyps to prevent recurrence. Given the recurrent nature of intussusception in PJS, continuous long-term follow-up with endoscopic and radiological surveillance is crucial to detect new polyps, prevent complications, and reduce future emergency surgeries [3,4]. Traditional laparotomy with bowel resection for symptomatic polyps poses a risk of short bowel syndrome. A recent technique called the "clean sweep", which combines both endoscopic and surgical approaches, is now recommended for managing small bowel polyps in PJS [3,16].

Conclusions

PJS is a rare condition with multiple gastrointestinal manifestations, including the risk of intussusception.

This case highlights the need to consider PJS in younger patients presenting with acute abdominal pain and signs of bowel obstruction, particularly when accompanied by distinctive mucocutaneous pigmentation.

CT imaging is very important in diagnosing intussusception, providing detailed visualization of the involved bowel segments and identifying severe complications such as is-

chemia or bowel obstruction. Early diagnosis and timely management are crucial to prevent further complications and optimize patient outcomes.

In this case, successful surgical intervention alleviated the acute symptoms, and the patient is educated to continue regular surveillance for gastrointestinal complications associated with PJS.

Patient consent

Informed consent was obtained by the patient for diagnostic examination. The patient understood that their anonymity would be preserved, and no identifiable information would be disclosed.

REFERENCES

[1] Katabathina VS, Menias CO, Khanna L, Murphy L, Dasyam AK, Lubner MG, et al. Hereditary gastrointestinal cancer syndromes: role of imaging in screening, diagnosis, and management. Radiographics 2019;39(5):1280–301. doi:10.1148/rg.2019180185.

[2] van Lier MG, Mathus-Vliegen EM, Wagner A, van Leerdam ME, Kuipers EJ. High cumulative risk of intussusception in patients with Peutz-Jeghers syndrome: time to update surveillance guidelines? Am J Gastroenterol 2011;106(5):940–5. doi:10.1038/ajg.2010.473.

[3] Bellamlih H, El Farouki A, Oulahyane R, Moatassim Billah N, Nassar I. Unusual case of small bowel intussusception in adult revealing a Peutz-Jeghers syndrome. BJR Case Rep 2021;8(2):20210082. doi:10.1259/bjrcr.20210082.

[4] Rahma C, Najoua BK, Mohamed Z, Manel M, Rim D, Dhaou Mahdi B, et al. Peutz-Jeghers syndrome revealed by recurrent small bowel intussusceptions in children: a case report. Clin Case Rep 2022;10(9):e6354. doi:10.1002/ccr3.6354.

[5] Rufener SL, Koujok K, McKenna BJ, Walsh M. Small bowel intussusception secondary to Peutz-Jeghers polyp. Radiographics 2008;28(1):284–8. doi:10.1148/rg.281075092.

[6] Beggs AD, Latchford AR, Vasen HFA, Moslein G, Alonso A, Aretz S, et al. Peutz-Jeghers syndrome: a systematic review and recommendations for management. Gut 2010;59:975–86. doi:10.1136/gut.2009.198499.

[7] Tomas C, Soyer P, Dohan A, Dray X, Boudiaf M, Hoeffel C. Update on imaging of Peutz-Jeghers syndrome. World J Gastroenterol 2014;20(31):10864–75. doi:10.3748/wjg.v20.i31.10864.

[8] Sharma H, Kaushik D. Intussusception in Peutz-Jeghers Syndrome: management of unusual acute abdominal presentation. Indian J Surg Oncol 2022;13(2):262–6. doi:10.1007/s13193-021-01448-9.

[9] Shah J, Sunkara T, Xiao P, Gaduputi V, Reddy M, Razia S. Peutz-Jeghers syndrome presenting as colonic intussusception: a rare entity. Gastroenterology Res 2018;11(2):150–3. doi:10.14740/gr964e.

[10] Pham HD, Nguyen THT, Tran TQ, Bui VG. Multidetector computed tomography findings of acute abdominal intussusception due to Peutz-Jeghers syndrome. Cureus 2021;13(7):e16401. doi:10.7759/cureus.16401.

[11] Wagner A, Aretz S, Auranen A, Bruno MJ, Cavestro GM, Crosbie EJ, et al. The management of Peutz-Jeghers syndrome: European Hereditary Tumour Group (EHTG) guideline. J Clin Med 2021;10(3):473. doi:10.3390/jcm10030473.

[12] Kim YH, Blake MA, Harisinghani MG, Archer-Arroyo K, Hahn PF, Pitman MB, et al. Adult intestinal intussusception: CT appearances and identification of a causative lead point. Radiographics 2006;26(3):733–44. doi:10.1148/rg.263055100.

[13] Valentini V, Buquicchio GL, Galluzzo M, Ianniello S, Di Grezia G, Ambrosio R, et al. Intussusception in adults: the role of MDCT in the identification of the site and cause of obstruction. Gastroenterol Res Pract 2016;2016:5623718. doi:10.1155/2016/5623718.

[14] Marsicovetere P, Ivatury SJ, White B, Holubar SD. Intestinal intussusception: etiology, diagnosis, and treatment. Clin Colon Rectal Surg 2017;30(1):30–9. doi:10.1055/s-0036-1593429.

[15] Somma F, Faggian A, Serra N, Gatta G, Iacobellis F, Berritto D, et al. Bowel intussusceptions in adults: the role of imaging. Radiol Med 2015;120(1):105–17. doi:10.1007/s11547-014-0454-4.

[16] Nasri S, Kellil T, Chaouech MA, Zouari K. Intestinal intussusception in Peutz Jeghers syndrome: a case report. Ann Med Surg (Lond) 2020;54:106–8. doi:10.1016/j.amsu.2020.04.013.