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Peutz-Jeghers Syndrome in a 7-Year-Old Male Presenting with Jejunojejunal Intussusception and a Giant Incidental Meckel's Diverticulum: A Case Report

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Abstract

Background: Peutz-Jeghers syndrome (PJS) is an autosomal dominant disorder characterized by gastrointestinal hamartomatous polyposis, most commonly the small intestine. Although intussusception is a recognaized complication of PJS, primary intussusception is uncommon in children older than three years of age, necessitating a high index of suspicion for underlying pathological lead points.

Case Presentation: We report a case of type B1 PJS in a seven-year-old male who initially presented with an acute abdomen. Subsequent laparotomy revealed a jejunojejunal intussusception along with an incidentally discovered large Meckel's diverticulum. Notably, the patient lacked the classic mucocutaneous hyperpigmentation typically associated with PJS.

Conclusion: Approaching an acute abdomen in a pediatric patient requires a careful diagnostic evaluation. In children older than three years presenting with intussusception, secondary causes such as PJS must be considered, even in the absence of pathognomonic mucocutaneous pigmentation.

Key Words: Intussusception; Meckel Diverticulum; Peutz-Jeghers Syndrome.

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1- INTRODUCTION

Peutz-Jeghers syndrome (PJS, OMIM: 175200) is an autosomal dominant polyposis syndrome that has been recognized as a distinct clinical entity for over a century. It is characterized by gastrointestinal (GI) hamartomatous polyposis, typically affecting the small intestine, which predisposes patients to an increased risk of malignancies both within and outside the GI tract. A major clinical concern regarding this syndrome is its age-dependent cumulative cancer risk, which reaches 75%–89% by the age of 70 years (1, 2). PJS has an estimated prevalence of 1 in 100,000 (3). Diagnosis is primarily based on clinical criteria, although genetic testing plays a definitive confirmatory role.

While instances of PJS presenting with intussusception have been documented in the medical literature, we report a unique case of a seven-year-old male with PJS who initially presented with intussusception and, interestingly, an incidentally discovered giant Meckel's diverticulum during laparotomy.

2- CASE PRESENTATION

A seven-year-old boy presented to our pediatric surgery center with a two-day history of abdominal pain. Initially, the pain, occurred intermittently every 15 minutes but gradually became continuous. Although the patient reported normal bowel movements at the onset of symptoms, he progressively lost the ability to pass stool and flatus. Concurrently, the child became acutely ill, febrile, and nauseated, with episodes of vomiting.

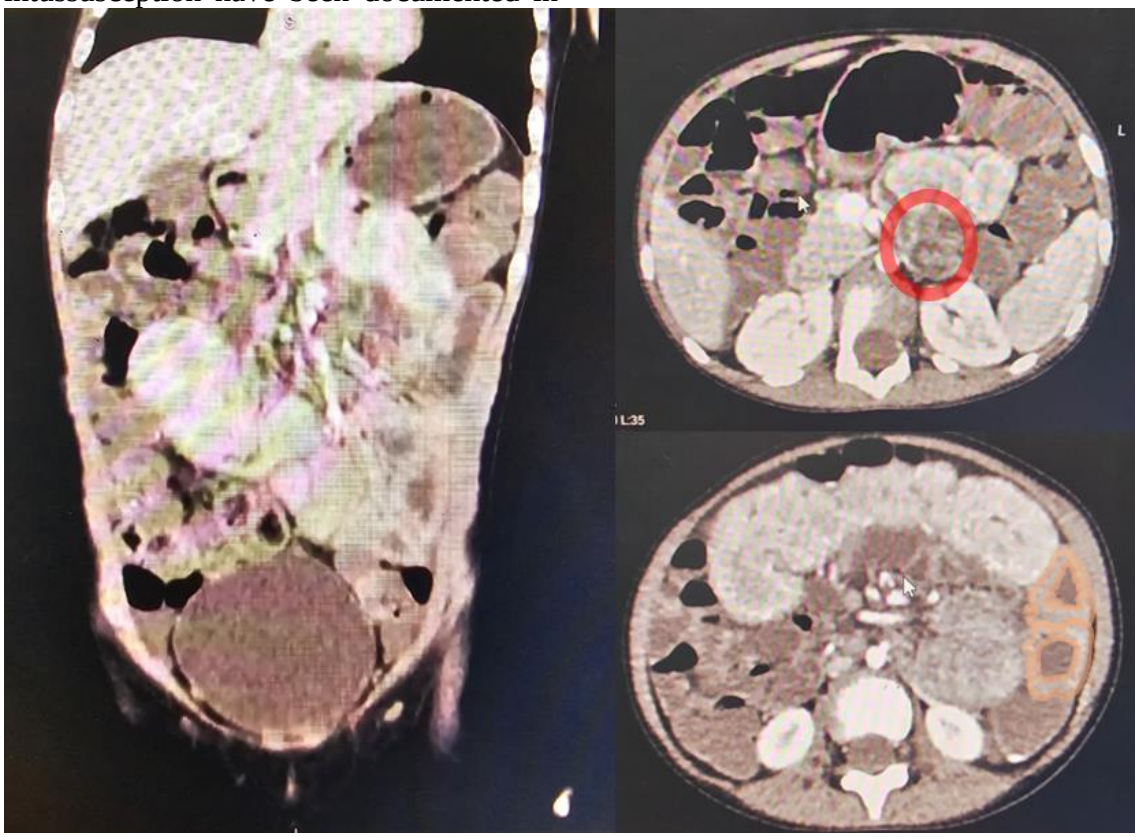

Figure-1: Preoperative coronal (left) and axial (right) computed tomography (CT) scans revealing a jejunojejunal intussusception. A target sign measuring 34×44mm is visible in the left lower quadrant at the proximal jejunum (red circle), accompanied by marked thickening of the jejunum and proximal ileum (orange circles).

Upon initial physical examination, the patient was tachycardic and tachypneic, with mild abdominal distension. The abdomen was symmetrical, but palpation elicited periumbilical tenderness, predominantly in the epigastric region. Abdominopelvic ultrasonography revealed a jejunojejunal intussusception. Computed tomography (CT) confirmed the ultrasonographic findings, demonstrating a "target sign" suggestive of intussusception in the left lower quadrant, measuring 34×44 mm, located in the proximal jejunum. There was also significant wall thickening and marked enhancement in the jejunum and proximal ileum (Figure 1). The radiological differential diagnosis included inflammatory bowel disease, celiac disease, eosinophilic enteritis, and, less likely, infiltrative neoplastic lesions. The invaginated segment demonstrated reduced enhancement compared to the adjacent jejunum, suggestive of ischemia.

With a preoperative diagnosis of incarcerated intussusception, the patient underwent an emergency midline laparotomy. Initial exploration revealed a 30-cm segment of thickened intestine, starting 10 cm distal to the ligament of Treitz. Palpable intraluminal polyps were identified within this segment (Figure 2). Evidence of a spontaneously reduced intussusception was noted; however, the primary pathology was localized to this 30-cm segment. The affected segment was resected (Figure 3), followed by a primary end-to-end enteroenteric anastomosis (Figure 4).

Further exploration revealed a similar 20-cm segment of intestinal thickening proximal to the anastomosis, although no palpable polyposis was detected. Incidentally, pathology, a large Meckel's diverticulum (MD) was discovered, which had not been identified on preoperative imaging and was subsequently resected (Figure 5).

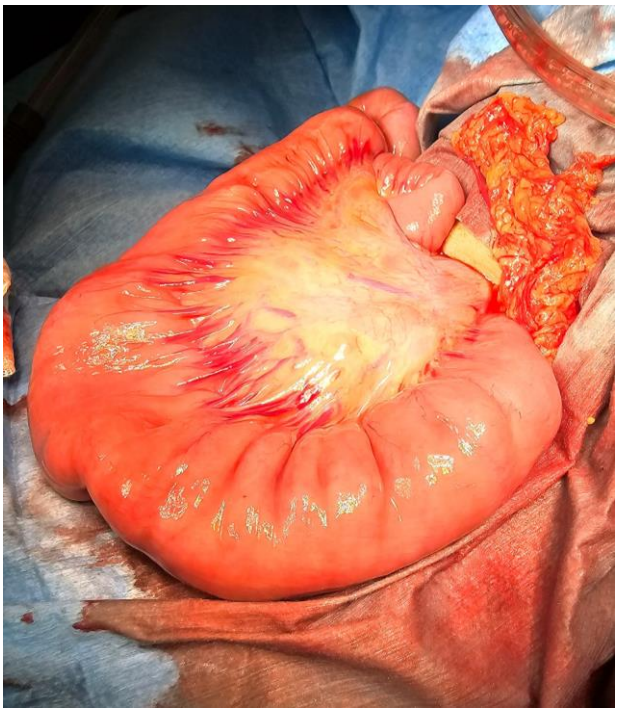

Figure-2: Intraoperative view of a thickened 30cm segment of the small intestine located 10cm distal to the ligament of Treitz. Intraluminal polyps were palpable within this segment prior to resection.

The resected specimens were sent to the pathology department. Pathology reports confirmed the presence of characteristic hamartomatous polyps with branching smooth muscle cores, consistent with Peutz-Jeghers syndrome (Figure 6). Evaluation of the diverticulum confirmed the histology of a true Meckel's diverticulum. Postoperatively, the patient was assessed for other clinical manifestations of Peutz-Jeghers syndrome. He lacked mucocutaneous hyperpigmentation, and there was no known family history of the disease. Therefore, according to the World Health Organization diagnostic criteria, this case is classified as type B1 (4) (Table 1). At bith the 6-month and 1-year postoperative follow-ups, the patient was clinically well, and upper and lower esophagogastric and intestinal endoscopic examinations were normal, with no polyps detected.

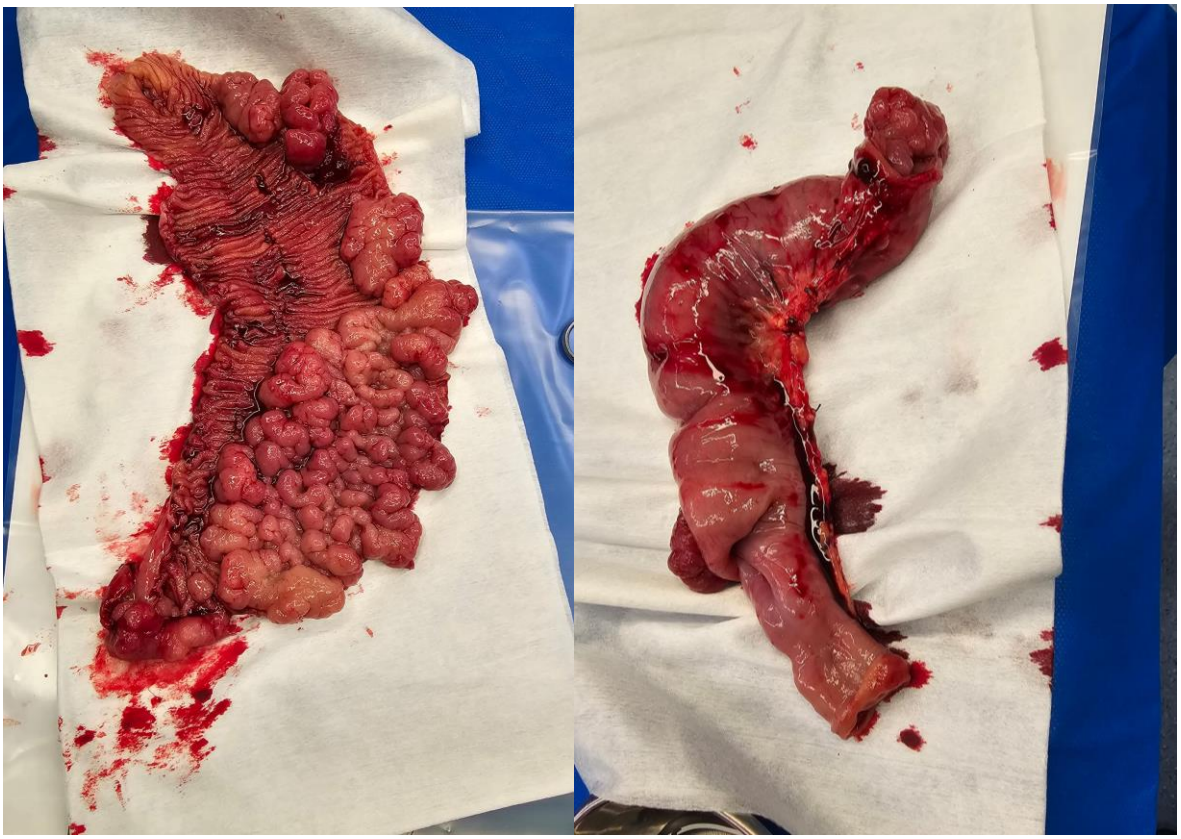

Figure-3: (A) Longitudinal section of the resected 30cm intestinal segment, revealing dense polyposis. (B) Gross specimen of the resected jejunojejunal.intussusception.

3- DISCUSSION

In 1921, Dr. Jan Peutz first reported a family with multiple affected individuals presenting with intestinal polyps and associated pigmentation. In 1949, Dr. Harold Jeghers expanded upon Peutz's findings, describing the condition syndromically as a distinct clinical entity (5). The hallmark of PJS is the widespread occurrence of hamartomatous polyposis thoughout the gastrointestinal tract. These polyps are typically smooth, lobulated growths arising from hyperplasia of the mucosal epithelium and dendritic growth of smooth muscle fiber bundles originating from the muscularis mucosae. They are primarily found in the small intestine, accounting for approximately 75% of cases (6), followed by the stomach, which accounts for 25% (7); however, other parts of the digestive system may also be affected, with the exception of the esophagus (6). Another diagnostic hallmark is mucocutaneous pigmentation, typically manifesting as dark blue or brown macules on the lips, buccal mucosa, perioral region, genitalia, and fingertips. Etiologically, PJS is an autosomal dominant inherited disorder caused by a germline pathogenic variant in the STK11 gene. Despite its hereditary nature, approximately 17% to 50% of PJS cases occur sporadically, with no family history of the disease. Consequently, several organizations have proposed clinical diagnostic criteria. The World Health Organization (WHO) criteria are listed in Table 1. To the best of our knowledge, the most recently updated diagnostic framework is from the Research Group for Improving the Medical Standard of Gastrointestinal Polyposis Syndromes, published by the Japanese Ministry of Health, Labour and Welfare in January 2018 (8), which is presented in Table 2.

Table-1. Diagnostic criteria of PJS from the point of WHO.

A	A positive family history of PJS and 1. Any number of histologically confirmed PJS polyps or 2. Characteristic prominent mucocutaneous pigmentation
B	A negative family history of PJS and 1. Three histologically confirmed PJS polyps or 2. Any number of histologically confirmed PJS polyps and characteristic prominent mucocutaneous pigmentation

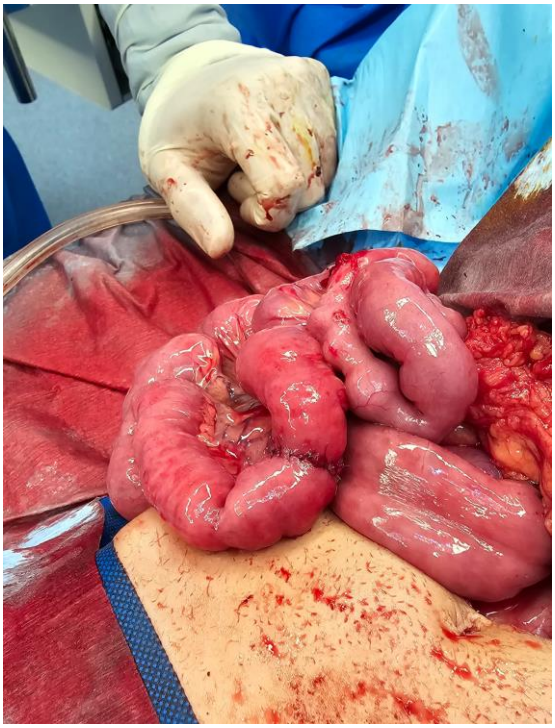

Figure-4: Jejunojejunal end-to-end anastomosis performed following the resection of the intussusception.

Table 2. Clinically, polyps larger than 15mm may act as lead points, causing intussusception and necessitating endoscopic or surgical treatment (9). While intussusception is recognized as the most common cause of intestinal obstruction and abdominal emergencies in young children, most pediatric cases are idiopathic. However, previous studies indicate that polyps are the leading pathological finding in 2.2% to 15% of pediatric intussusception cases (10-12).

In our case, it is highly likely that both PJS polyposis and Meckel's diverticulum contributed to the jejunojejunal intussusception.

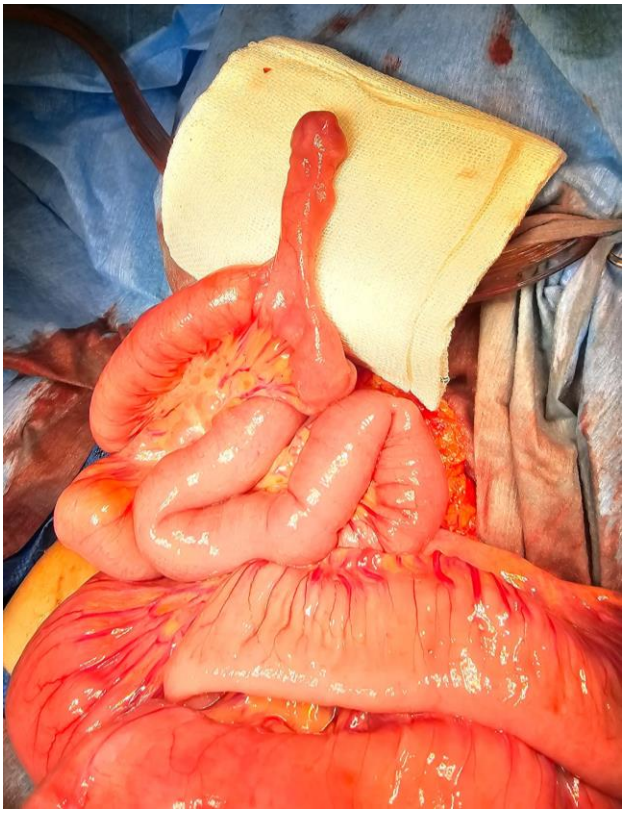

Figure-5: Gross appearance of the large, incidentally discovered Meckel's diverticulum.

A review of the medical literature reveals that the most common tumoral causes of pediatric secondary intussusception are benign polyps, PJS hamartomas, and malignant lymphomas (10, 11, 13-15). In contrast to primary intussusception—where a non-operative approach using pneumatic or hydrostatic reduction is preferred—the management of intussusception secondary to intestinal tumors requires surgical intervention (10). Although the initial presentation of PJS is rarely marked by life-threatening complications such as severe rectal bleeding, gastrointestinal obstruction, or perforation, there are documented cases in

which intestinal obstruction due to intussusception was the first sign of the disease (16, 17). Given the increased risk of recurrent obstruction, a complete small bowel survey and resection of all concerning polyps are recommended during laparotomy whenever possible (9, 18). A significant long-term concern for patients with PJS is recurrent intussusception secondary to small bowel polyps; multiple surgical resections over a patient's lifetime can ultimately lead to short bowel syndrome (9, 19). An incidental finding during the operation was a MD, the most common congenital gastrointestinal malformation. MD is an embryologic abnormality resulting from the failure of the vitelline duct to close and is present in approximately 2% of the population (20). As a true diverticulum, it contains all three layers of the small bowel wall. Its complications are well-documented and most commonly include obstruction, followed by hemorrhage, perforation, diverticulitis, and intussusception (11). Although the management of incidentally discovered MD remains somewhat controversial, a recent literature review indicates a paradigm shift toward prophylactic resection for all incidentally discovered cases or, at a minimum, for patients exhibiting high-risk factors (21).

Table-2. Diagnostic criteria of PJS from the point of the Japanese Ministry of Health, Labour and Welfare.

A	**Symptoms** 1. Mucocutaneous pigmentations of 1–5 mm on the lips, mouth, and fingertips
B	**Examination findings** **1. Endoscopic findings:** Upper gastrointestinal endoscopy, colonoscopy, and small-bowel endoscopy (capsule endoscopy or balloon-assisted endoscopy) show hamartomatous polyps in any gastrointestinal tract except the esophagus **2. Pathological findings:** Hamartomatous polyps have hamartomatous hyperplasia of the mucosal epithelium and dendritic growth of smooth muscle fiber bundles from the muscularis mucosae, which can be diagnosed as Peutz-Jeghers polyps
C	**Differential diagnosis** **Differentiate from the following diseases** Familial adenomatous polyposis, juvenile polyposis syndrome, Cowden syndrome/PTEN hamartomatous syndrome, tuberous sclerosis, inflammatory polyposis, serrated polyposis syndrome, Cronkhite-Canada syndrome, hereditary mixed polyposis syndrome, and Laugier-Hunziker-Baran syndrome
D	**Genetic testing** **Germline pathogenic variants in STK11 gene** <Diagnosis Category*> 1. A is fulfilled, two of B are fulfilled, and the differential diagnoses in C are excluded 2. A is fulfilled in an individual who has a family history of PJS in close relation, and the differential diagnoses in C are excluded 3. Two of B are fulfilled in an individual who has a family history of PJS in close relation, and the differential diagnoses in C are excluded 4. B-1 is fulfilled, B-2 is met for multiple lesions, and the differential diagnoses in C are excluded 5. D is fulfilled

*For patients who meet some of the diagnostic criteria but do not meet the above diagnostic categories (1–4) based on symptoms and laboratory findings, a search for germline pathogenic variants in STK11 should be considered for diagnosis by genetic testing. If a germline pathogenic variant in STK11 is identified, PJS can be diagnosed.

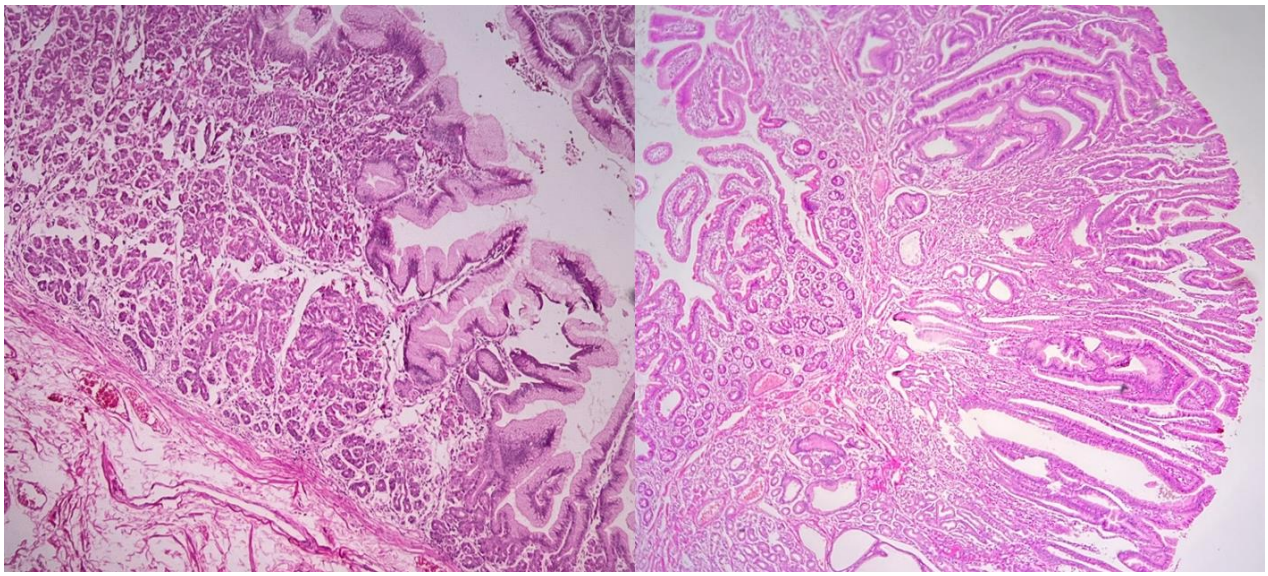

Figure-6: Hematoxylin and Eosin staining of a hamartomatous polyp of Peutz-Jeghers syndrome in ×100 and ×400 magnification; papillary texture with bundles of smooth muscle as dense arboriculture.

4- CONCLUSION

Pediatric abdominal pain often presents a diagnostic challenge due to the wide range of potential pathologies, varying severity, and the patient's possible inability to provide an accurate history or cooperate during physical examinations. While most causes are self-limiting, acute abdominal pain can indicate a surgical emergency requiring prompt and precise intervention. Carefully considering the patient's age can significantly narrow the differential diagnosis. Since idiopathic intussusception is uncommon in children older than 7 years, healthcare teams must maintain a high index of suspicion for underlying secondary pathologies in children over 3 years old. This case ultimately highlights two critical clinical points: not all pediatric intussusception cases are primary, and the absence of classic mucocutaneous pigmentation does not definitively exclude Peutz-Jeghers syndrome.

5- ETHICAL CONSIDERATIONS

Informed written consent for the publication of the patient's medical data was obtained from the patient's guardians and documented in the hospital records. As this work is a case report, approval from the institution's ethics committee was not required.

6- DATA AVAILABILITY

Data are available from the authors upon request.

7- CONFLICT OF INTEREST

The authors have disclosed no conflicts of interest.

8- FUNDING

The authors declare that they have not received any financial support.

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