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Case Report

Double Intussusception: A Case Revealing Peutz-Jeghers Syndrome

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Abstract

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant genetic disorder that features multiples mucocutaneous pigmentation, hamartomatous polyps usually in the small intestine, and often a family history of the condition. These polyps can lead to intermittent abdominal pain, chronic anemia, and, notably, bowel obstruction due to intussusception, where a segment of the intestine folds into itself. Patients suffering from this syndrome have an increased risk for common and unusual types of gastrointestinal and non-gastrointestinal tumors. Recent guidelines recommend regular small-bowel surveillance to reduce these risks associated with PJS. Imaging techniques such as CT scans are valuable in detecting intussusception caused by these polyps. However, a definitive diagnosis of Peutz-Jeghers Syndrome requires histological examination of the polyps. we present a rare case of a 17-year-old female with chronic anemia and abdominal pain, where double intussusception was identified on CT scans. Subsequent surgical resection of the polyps revealed the presence of hamartomatous Peutz-Jeghers polyps upon histological examination.

Keywords: *Peutz-Jeghers Syndrome (PJS); Intussusception; Polyps*

Introduction

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant genetic disorder characterized by specific gastrointestinal hamartomatous polyps and mucocutaneous pigmentations around the oral and anal areas [1]. The hamartomatous polyps in the intestine cause intermittent abdominal pain, chronic anemia, and can lead to complications such as gastrointestinal bleeding, obstruction due to intussusception [2]. Patients suffering from this syndrome have an increased risk for common and unusual types of gastrointestinal and non-gastrointestinal tumors [1,2]. Imaging allows the diagnosis of intussusception with these polyps, and histological examination establishes the formal diagnosis of Peutz-Jeghers syndrome (PJS).

Case Report

A 17 years old girl presented to emergency department with severe abdominal pain and vomiting. Upon examination, the abdomen was soft and without signs of peritoneal irritation. Upon inspection, there was black pigmentation on the lower lip without any other cutaneous or mucosal lesions (Figure 2F). Laboratory tests showed a slight elevation in CRP (C-reactive protein) and a known iron-deficiency anemia treated with injections. The origin of this anemia has not yet been determined. She had no known surgical or family history.

Faced with this acute abdominal symptomatology, an emergency abdominal ultrasound was performed. It revealed suggestive images of invaginations in the form of "target sign" in cross-sectional images and a "pseudo kidney sign" in longitudinal images of the periumbilical quadrant (Figure 2E). Hence, intussusception was diagnosed according to these findings.

To further the research etiologic of this intussusception, a CT scan of the abdomen and pelvis with intravenous contrast was performed. It showed a duodeno-jejunal intussusception involving the distal duodenum (D3) with the proximal segment of the jejunum, containing polyploid tissue formations, measuring approximately 40 x 33 mm, related to polyps with upstream dilatation measured at 40 mm in maximum thickness. The second invagination is jejuno-jejunal on multiple polyps with the same characteristics described above. The patient underwent the following day a duodenotomy with resection of the duodenal and jejunal polyps (Figure 1).

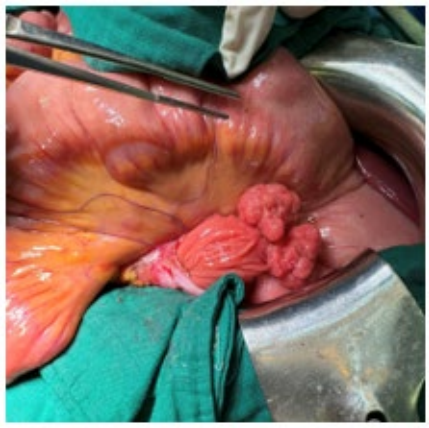

Figure 1: *Pedunculated polypoid mass with a typical multilobulated surface is seen. The head of the polyp is 4.5 cm in diameter.*

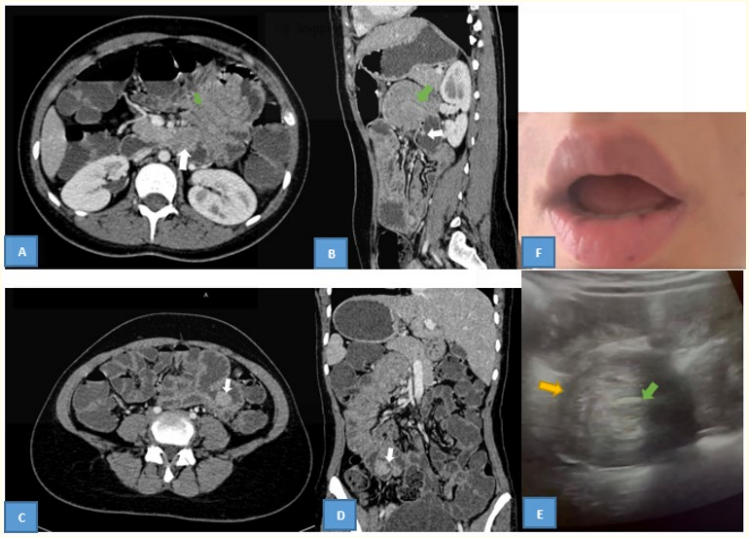

Figure 2: *CT scan of the abdomen and pelvis with intravenous contrast on the axial plan (A) with sagittal (B) and coronal reconstruction (C) showing duodeno-jejunal intussusception and jejuno-jejunal containing several polyps (white arrows). (D) Sonographic ultrasound (E) appearance of an ileo-ileal invaginations. Longitudinal section of the intussusception sausage showing successive layers of the digestive wall (yellow arrow). In the center, invaginated mesenteric fat with vessels (green arrow).*

Discussion

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant genetic syndrome characterized by multiples gastrointestinal hamartomata's polyp often accompanied by oral and anal mucocutaneous pigmentations. it is usually diagnosed at the age of 20 - 30 years [1].

Clinical manifestations of PJS are characterized by alternating asymptomatic periods and episodes of complications, including abdominal pain, recurrent intussusception leading to bowel obstruction, prolapse of rectal polyps, and often occult bleeding. Small bowel obstruction commonly presents initially, often necessitating repeated surgeries due to polyp-related complications recurring at relatively short intervals [2]. Our patient presented with symptoms of intermittent upper intestinal obstruction and anemia for two years.

Peutz-Jeghers Syndrome (PJS) not only predisposes patients to gastrointestinal polyps but also significantly elevates their risk of developing both gastrointestinal and non-gastrointestinal malignancies studies indicate that individuals with Peutz-Jeghers syndrome (PJS) face a cumulative cancer risk of up to 93% from ages 15 to 64, which is significantly higher than that of the general population [3]. They also have a relative risk of developing neoplasms that can be up to 15 times greater. The most commonly observed cancers include colonic tumors, followed by breast, pancreatic, gastric, ovarian, small intestinal, and uterine cancers [3,4]. Most authors agree that surveillance is needed in PJS patients. According to clinical guidelines from the American College of Gastroenterology, it is recommended that patients undergo an annual comprehensive health assessment. This includes a complete blood cell count and a physical examination that encompasses evaluation of the breasts, abdomen, pelvis, and tests [5].

The recommended baseline endoscopic screening of the gastrointestinal tract should commence at eight years of age and include upper gastrointestinal endoscopy, video capsule endoscopy (VCE), and colonoscopy. Subsequent screening intervals should be tailored to the findings of the initial examination: If polyps are identified during the baseline screening, upper endoscopy, VCE, and colonoscopy should be repeated every two to three years. Conversely, if no polyps are found during the initial screening, upper endoscopy, VCE, and colonoscopy should be repeated at age 18, or sooner if symptoms develop, with subsequent follow-up screenings scheduled every two to three years [6].

Polyp removal represents the cornerstone in preventing complications. Timely polypectomy can preempt the need for recurrent emergency interventions and extensive resections of the small intestine. For decades, laparotomy and bowel resection were the standard treatments for managing symptomatic gastrointestinal polyps. However, some patients may need multiple surgeries, which can increase the risk of developing short bowel syndrome. The "clean sweep" approach combines endoscopy and surgery to manage small intestinal polyps in patients with Peutz-Jeghers syndrome (PJS). This integrated treatment aims to alleviate gastrointestinal symptoms and proactively mitigate the necessity for repetitive abdominal procedures [7].

Summary

Acute intestinal invaginations are rare in Peutz-Jeghers syndrome (PJS), and their multiplicity is exceptional. However, this syndrome should be considered in cases of recurrent abdominal pain in patients with cutaneous pigmentation and chronic anemia. Ultrasound and CT scan are the imaging modalities of choice on diagnosis with sensitivity and negative predictive value close to 100%. Early detection of this genetic disorder in affected individuals and their relatives, combined with diligent cancer surveillance, is essential for achieving favorable outcomes.

Bibliography

1. Arber N and Moshkowitz M. "Small bowel polyposis syndromes". *Current Gastroenterology Reports* 13.5 (2011): 435-441.
2. Loureiro J., *et al*. "Diagnostic difficulty in Peutz-Jeghers syndrome". *Journal of Coloproctology (Rio de Janeiro)* 35.1 (2015): 67-71.
3. Chen HY., *et al*. "Cancer risk in patients with Peutz-Jeghers syndrome: a retrospective cohort study of 336 cases". *Tumor Biology* 39.6 (2017): 101042831770513.
4. Fan RY and Sheng JQ. "A case of Peutz-Jeghers syndrome associated with high-grade intramucosal neoplasia". *International Journal of Clinical and Experimental Pathology* 8.6 (2015): 7503-7505.
5. Homan M., *et al*. "Peutz-Jeghers syndrome. A case report". *Acta Dermatovenerologica Alpina Pannonica et Adriatica* 14.1 (2005): 26-29.
6. Flageole H., *et al*. "Progression toward malignancy of hamartomas in a patient with Peutz-Jeghers syndrome: case report and literature review". *Canadian Journal of Surgery* 37.3 (1994): 231-236.
7. Wang H., *et al*. "Clinical presentations and surgical approach of acute intussusception caused by Peutz-Jeghers syndrome in adults". *Journal of Gastrointestinal Surgery* 15.12 (2011): 2218-2225.

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