BJHS
Birat Journal of Health Sciences

ISSN:

2542-2758 (Print) 2542-2804 (Online)

ARTICLE INFO:

Received Date: November 19, 2024
Accepted Date: March 20, 2025
Published Date: 31 August, 2025

KEYWORDS:

Peutz-Jeghers Syndrome; PJS; Intussusceptions.

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Access the article online

DOI: 10.62065/bjhs633

CITATION:

KC S, Poudyal H. Intussusception as the initial presentation of previously undiagnosed Peutz–Jeghers Syndrome- A case report. 2025; 10 (2): 101-104.

Intussusception as the initial presentation of previously undiagnosed Peutz–Jeghers Syndrome- A case report

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ABSTRACT

Peutz-Jeghers Syndrome (PJS) is an autosomal dominant condition characterized by hamartomatous polyps within the gastrointestinal tract and mucocutaneous pigmentation. Patients with PJS have heightened risks for various malignancy, requiring regular follow up. This report discusses a 18-year-old male who initially presented to the Emergency Department with complaints of abdominal pain. Imaging and clinical findings were suggestive of PJS. This case report discusses a case of jejunal intussusception as a initial presentation in previosly undiagnosed PJS. Imaging is critical for diagnosis, identifying complications, and future surveillance.

Introduction

Peutz-Jeghers Syndrome (PJS) is characterized by the presence of gastrointestinal hamartomatous polyps, mucocutaneous hyperpigmentation, and various other tumors.[1] The syndrome was first recognized in the late 19th century and received increased attention in the 1920s, particularly when Dr. Jeghers emphasized its distinct features in 1949. The term "Peutz-Jeghers Syndrome" was introduced by Brewer and his team in 1954. This rare condition occurs in approximately 1 in 50,000 to 1 in 20000 live births, affecting individuals regardless of gender or ethnicity.[2]

The condition is mainly linked to mutations in the STK11 tumor suppressor gene found on chromosome 19p13.3; these mutations impair kinase activity and disrupt cell cycle signaling, leading to an increased risk of various cancers, with an increased risk factor of 15 to 18 times compared to the general population. Typically, these mutations are inherited in an autosomal dominant fashion, with approximately 25% arising as de novo mutations.[3] This case report discusses the clinical presentation, diagnostic criteria, genetic components, related malignancy, and imaging screening protocols in line with established guidelines.

Clinical Presentation

An 18-year-old male presented to emergency department with a history of abdominal pain and vomiting that had persisted for four days. He also gave history of recurrent mild abdominal discomfort for the past 5 to 6 years. There were no indications of hematemesis or melena, and his family medical history was unremarkable. Upon admission, his vital signs were stable. However, a physical examination revealed slight abdominal distension and generalized tenderness. Additionally, skin examination showed hyperpigmented macules on both the lips and palms. Laboratory tests were within normal limit.

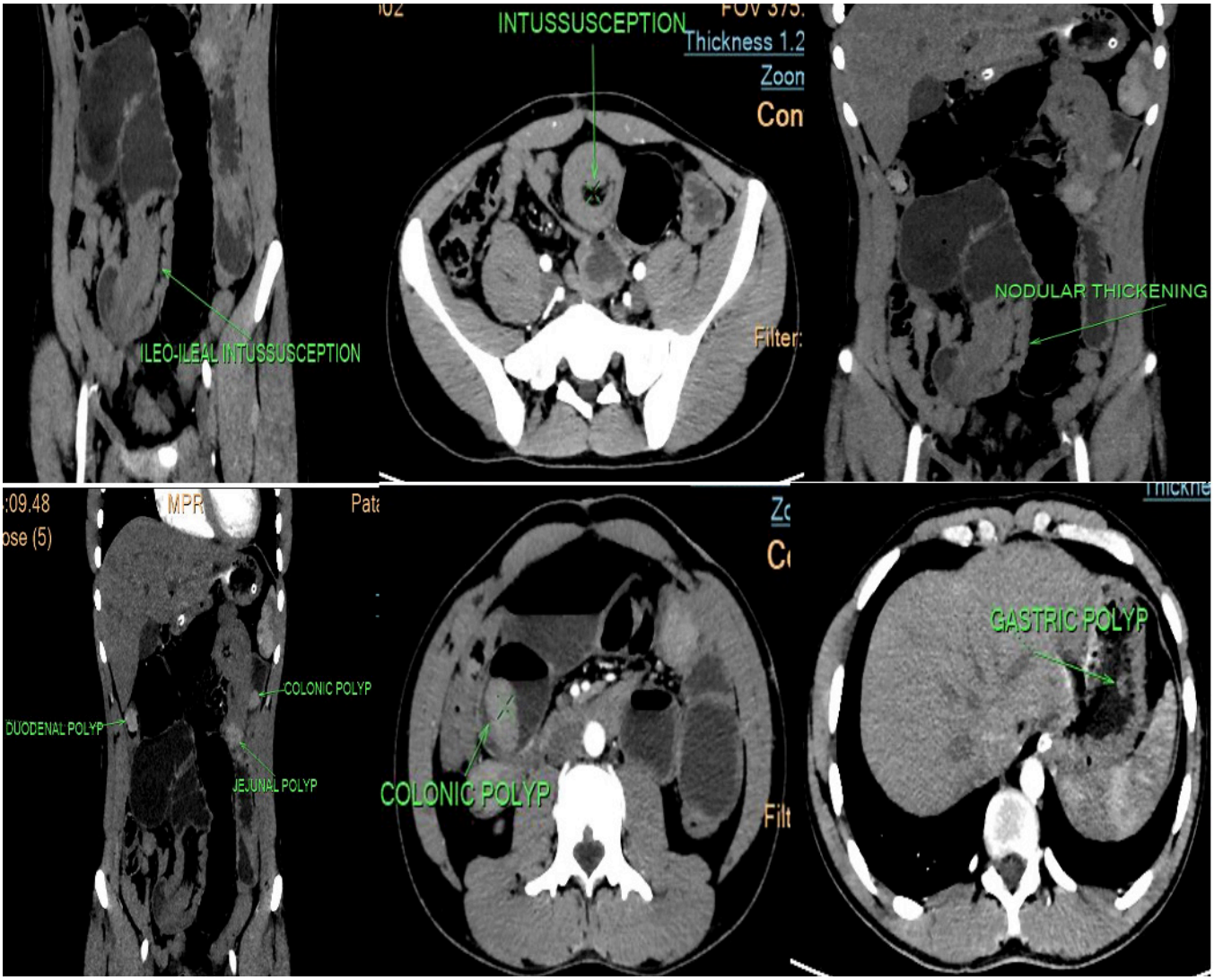
Abdominal ultrasonography(USG) demonstrated a characteristic "target sign" in periumbilical region suggestive of intussusception. Contast enhanced Computed Tomography(CECT) scan of the abdomen and pelvis showed telescoping of bowel

within bowel loop at proximal jejunum with enhancing nodular leions at region of telescoping of bowel. Dilatation of small bowel loops noted proximal to it. Multiple homogeneously enhancing endoluminal, polypoid masses with attachment to bowel wall were noted in body of stomach, second part of duodenum, jejunum and transverse colon. Based on these spectrum of mucocutaneous hyperpigmentation and multiple polyps in the gastrointestinal tract, the diagnosis of the Peutz–Jeghers syndrome (PJS) was made.

On the second day of admission, abdominal pain decreased. A follow-up ultrasound of the abdomen indicated a reduction in the previously observed intussusception, leading to the decision to postpone surgery. The patient was discharged on the sixth day after resuming a normal diet and having normal bowel movements. The patient received counseling regarding the condition and is currently under regular follow-up in the surgical department as of the writing of this manuscript

Figure Legends

Contrast enhanced Commputed Tomography(CECT) showed multiple polyps along the wall of jejunum appearing as nodular thickening of jejunum, Target sign, multiple polyps in duodenum, colon, jejunum and stomach



Discussion

Intussusception is a condition where a segment of the intestine telescopes into itself or into an adjacent segment due to peristaltic movement leading to bowel obstruction, ischaemia, necrosis and perforation if untreated. In pediatric cases, intussusception is a frequent emergency that often occurs without a indentifiable cause(90% cases). Conversely, in adults, it is relatively uncommon, accounting for only 1% to 5% of cases, with 90% linked to a specific underlying condition that serves as a lead point such as colorectal carcinoma, lymphoma, GIST, Polyps, lipoma and rarely congenital etiology such as Meckel Diverticulum, duplication cyst.[4]

The classic triad of intussusception—intermittent abdominal pain, vomiting, and a right upper quadrant mass—occur in less than 20% of cases, with 15% presenting without abdominal pain making the diagnosis difficult. Classical examination

findings include a palpable, "sausage-like" mass in the right upper quadrant and a lack of palpable bowel in the right lower quadrant. "Currant jelly" stool, a mix of blood and mucus might be seen, indicating bowel ischemia.[5]

Abdominal X-rays can show a stretched soft tissue mass in the upper right quadrant of children, along with signs of bowel obstruction and perforation. A contrast enema is the gold standard for diagnosis and treatment, revealing intussusception as a occluding mass prolapsing into the lumen with a coiled spring sign; this study however needs to be avoided in case of perforation.[6] USG is a reliable screening method for low-risk children with intussusception. Key USG features include the target sign, pseudo-kidney sign, crescent in a doughnut sign, vascularity, and occasionally a lead point.[7] For adults, CT is the preferred method for evaluating acute abdominal conditions, classically showing bowel within bowel configuration(equiavalent to target sign on USG) as shown in our case.

One of the rare cause of intussusception in adult is PJS. The World Health Organization (WHO) has established the following criteria for diagnosing PJS clinically:[8]

- Three or more histologically confirmed Peutz–Jeghers polyps,
- Any number of Peutz–Jeghers polyps with a family history of PJS,
- Characteristic mucocutaneous pigmentation with a family history of PJS,
- Any number of Peutz–Jeghers polyps and characteristic mucocutaneous pigmentation.

Our patient had multiple polyp along with typical mucocutanoeus pigmentation. These pigmentations are hamartomatous macules, particularly in the oral cavity and genital areas, starting in infancy and potentially diminishing in adolescence, though oral lesions may persist into adulthood.[1]

Gastrointestinal symptoms typically arise around age 13, with half of patients experiencing issues by early 2nd decade, including intussusception, bowel obstruction, rectal bleeding, and anemia due to hamartomatous polyps, mainly in the jejunum which occurs in roughly 70% of PJS patients often requiring surgical intervention. The risk of intussusception increases with increasing age and polyp size of > 15 mm.[9]

PJS is associated with both gastrointestinal and non-gastrointestinal malignancy, with gastrointestinal malignancies being the most frequent, representing nearly two-thirds of all cases.

Colorectal cancer is the most common gastrointestinal cancer linked to PJS, with a cumulative risk of 39% as individuals age. Recommended screening methods include endoscopy, video capsule endoscopy, and CT or MR enterography, with fluoroscopic gastrointestinal series as an alternative for those unable to undergo endoscopy. CT scans are the most effective pre-operative diagnostic tool for intussusception related to PJS, with about 83% accuracy. Initial screenings should start at age 8, with follow-up every 2 to 3 years if polyps are found; if no polyps are detected, screenings can be delayed until age 18.[2,10]

Breast cancer is the second most common cancer in PJS, affecting 32–54% of patients, with an average diagnosis age of 37. High-risk management includes monthly self-exams starting at 18, biannual clinical assessments, and annual mammograms from age 25, with MRI recommended to reduce radiation exposure. Prophylactic mastectomies and genetic counseling are advised, though no standard management protocols exist.[2,10]

Pancreatic maligancy is also associated with PJS, affecting 36–40% of individuals, usually in their 50s to 60s. Screening is recommended between ages 25-35yrs using MRCP or EUS, with follow-ups every 2 to 3 years.[1,2,10] Additionally, PJS patients face an increased risk of gynecological maliganancy, particularly ovarian malignancy, which has a 21% lifetime risk and an average onset age of 28, similar to BRCA mutation risks. Annual transvaginal ultrasounds and CA-125 tests are recommended starting at age 25 for PJS patients, despite limited evidence for screening. PJS patients are at increased risk for sex cord tumors with annular tubules (SCAT), with over one-third of cases occurring in this group. Cervical cancer screening should follow standard guidelines, as PJS patients may also face adenoma malignum, a rare cervical adenocarcinoma that can resemble complex Nabothian cysts on ultrasounds. Sertoli cell tumors, the most common testicular cancer associated with PJS, typically appear around age 9, with USG recommended only if abnormalities or early puberty are present.[1,2,10]

Individuals with STK11 mutations face a 7% risk of lung cancer by age 60, significantly higher than the 1% risk in the general population. However, there are no specific lung cancer screening guidelines for those with STK11 mutations, aside from standard screenings for smokers. Pulmonary lesions are often discovered incidentally or during metastasis evaluations.[11]

Genetic testing and comprehensive evaluations are essential for diagnosing PJS, and relatives of affected individuals should also be tested.[10]

Treatment for gastrointestinal polyps depends on their size and number. The malignant potential of PJS polyps remains unconfirmed, and it is uncertain whether endoscopic polypectomy can effectively prevent cancer or lower the risk of developing it.[12] Endoscopic polypectomy is preferred for polyps over 1 cm, while larger or symptomatic polyps may require laparotomy. Removing polyps is crucial to reduce the need for frequent emergency surgeries that can lead to short gut syndrome.[13] An integrated approach combining endoscopic and surgical methods is ideal for small intestinal polyps presenting with intussusception, but was not necessary in our case due to spontenous reduction of intussusception

Studies are ongoing for mTOR inhibitors (rapamycin, everolimus), COX-2 inhibitors, aromatase inhibitors, and metformin.[2] Currently, there is no established pharmacological prevention for this syndrome.

Conclusions

Radiological imaging not only plays a crucial role in confirming the diagnosis but also guides treatment planning, screening, surveillance including the consideration of other associated malignancy. Established imaging protocols are limited, but guidelines and multi disciplinary consensus can guide management.

Funding Information: This article did not receive any grants.

Conflict of Interest Statement

No conflict of interests.

Data Availability Statement

All the findings are present within the manuscript.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editor in chief of this journal on request

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