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Case Report

Colocolic Intussusception: A Case Report of an Uncommon Manifestation of Peutz-Jeghers Syndrome

Qasem Alyhari[1], Saleh Al-Wageeh[1], Faisal Ahmed[2*], Hanan Mohammed[1], Noha Dahan[1], Asad Alhadi[3]

1 Department of General Surgery, School of Medicine, Ibb University, Ibb, Yemen.
2 Department of Urology, School of Medicine, Ibb University, Ibb, Yemen.
3 Department of Radiology, School of Medicine, Ibb University, Ibb, Yemen.
* Corresponding author's email: fmaaa2006@yahoo.com

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ABSTRACT

Peutz-Jeghers syndrome is a rare genetic disorder resulting from defects in signaling pathway regulation, marked by gastrointestinal hamartomas and mucocutaneous pigmentation. This condition can lead to complications such as intussusception, with colocolic intussusception being particularly rare. We present the case of a 30-year-old female who exhibited acute on chronic abdominal pain and progressive distension, along with digital and perioral hyperpigmentation. Unfortunately, her diagnosis of Peutz-Jeghers syndrome was delayed until she developed intestinal obstruction. Imaging revealed large bowel obstruction with proximal bowel dilatation, prompting a left hemicolectomy and end-to-end colocolic anastomosis. The patient experienced an uneventful postoperative recovery and remained symptom-free during follow-up. This case highlights the rarity of colocolic intussusception as a manifestation of Peutz-Jeghers syndrome and emphasizes the importance of early recognition and intervention to prevent bowel obstruction and the need for extensive surgical procedures.

Introduction

Peutz-Jeghers Syndrome (PJS) is an autosomal dominant disorder caused by germline mutations in the LKB1 gene, leading to disruption of the threonine kinase pathway signaling.[1] Such aberrations manifest clinically with mucocutaneous hyperpigmented lesions and gastrointestinal (GI) hamartomatous polyps.[2] Although these polyps are generally benign, individuals with PJS have an increased risk of developing GI malignancies, as well as cancers of the genital tract and breast. While PJS patients may be asymptomatic at the time of diagnosis, several GI complications have been reported which include GI bleeding, bowel obstruction, and intussusception [2,3]. Intussusception is particularly notable, occurring in more than 60% of PJS patients. However, the majority of cases involve the small intestine, predominantly the ileum or jejunum.[3,4] This predilection has been attributed, in part, to the higher density of polyps in these regions compared to the duodenum and colon, which typically have a lower polyp density and are partially retroperitoneal in location.[5] Here, we present a case of a young female who presented with chronic abdominal pain consistent with imaging revealing evidence of large bowel intussusception complicated with bowel obstruction.

Unfortunately, the diagnosis of her PJS was delayed until she experienced a gastrointestinal (GI) obstruction.

Case Presentation

A 30-year-old female presented with worsening colicky lower abdominal pain, accompanied by abdominal distension and constipation, which had developed over the preceding four days. She presented to our hospital (Almanar Hospital, Ibb Governorate, Yemen). The patient reported a history of recurrent, intermittent left flank pain and sporadic rectal bleeding following defecation, managed conservatively without definitive diagnostic investigation. Over the past three years, she sought medical attention from a general practitioner, receiving multiple diagnoses and various medications, all without discernible therapeutic benefit. She had undergone two prior hospitalizations for similar symptomatology. Following both admissions, the patient had been discharged prior to initiation of comprehensive investigations to determine the etiology of her rectal bleeding, secondary to financial constraints. Her medical, surgical, and family history were non-contributory.

The patient appeared ill, with no pallor or palpable lymphadenopathy. She was underweight (Body Mass Index of 17 kg/m²), afebrile, and hemodynamically stable. The patient's facial examination revealed dark brown pigmentation in the perioral region, lower lips, and buccal mucosa, without any distinctive facial features (Figure 1).

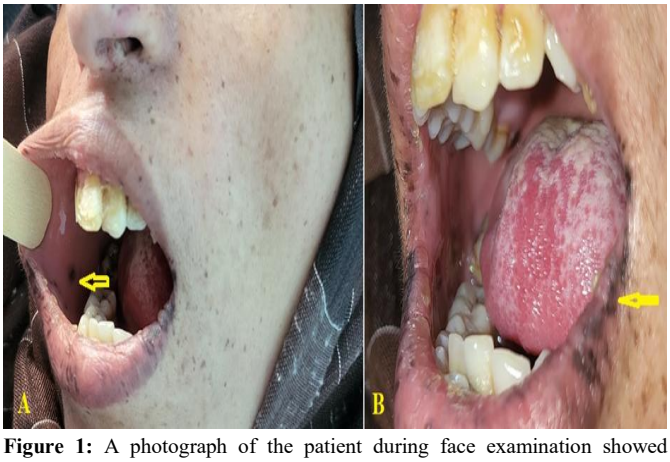

Figure 1: A photograph of the patient during face examination showed exhibited black pigmentation in the palmar surfaces of her hands (A) and feet (B) (Yellow arrows).

The upper and lower extremities exhibited dark brown hyperpigmentation on the palmar surfaces and feet, without clubbing or peripheral cyanosis (Figure 2).

The abdominal examination indicated mild distention, intact hernial orifices, and a tender palpable mass in the left iliac fossa. The per rectal examination was unremarkable. The patient was subsequently admitted for further evaluation.

Initial laboratory tests revealed a hemoglobin level of 13 gm/dL and a white blood cell count of 13,500/µL, with neutrophil-predominance. Immunoserology showed an elevated C-reactive protein of 30 mg/L and an erythrocyte sedimentation rate of 83 mm/hr. Hepatic viral markers were negative. Alkaline phosphatase was elevated at 510 U/L (normal: <300 U/L), while liver and renal function tests, coagulation profile, and electrolyte panels were otherwise unremarkable.

Abdominal ultrasonography identified a large, heterogeneous solid intestinal mass measuring 88 × 55 mm, causing luminal narrowing and proximal bowel segment dilation. Abdominopelvic computed tomography (CT) scan revealed a large, well-defined soft tissue density measuring 7.5 × 6 cm, along with an inability to separate it from the descending colon with dilated bowel loops proximally and gas-fluid levels. Additionally, mesenteric vessel enhancement and hypodense mesenteric fat were noted (Figure 3).

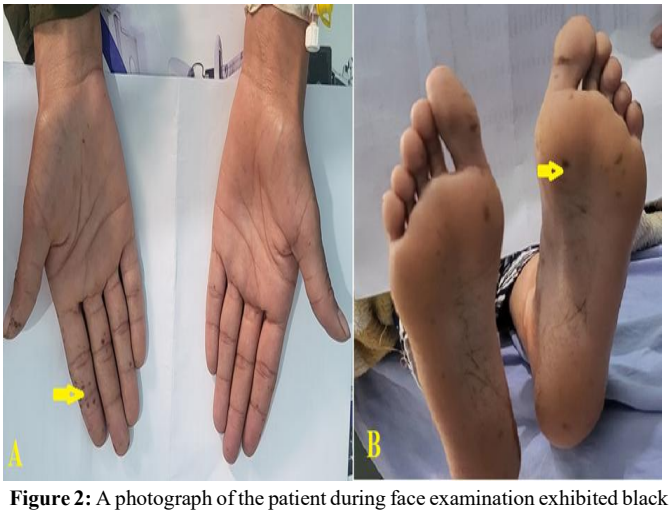

Figure 2: A photograph of the patient during face examination exhibited black pigmentation in her oral cavity (A), and perioral region (B) (Yellow arrows).

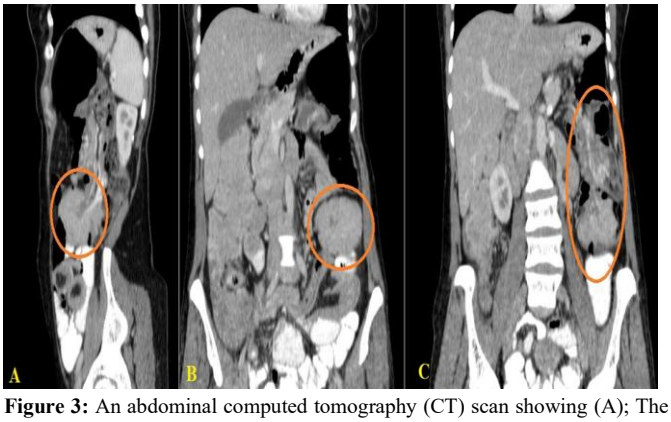

Figure 3: An abdominal computed tomography (CT) scan showing (A); The longitudinal axis representing intussusceptions (Orange circle line) and linear-enhancing structures within mesenteric fat in mesenteric blood vessels (Yellow arrow). (B); The longitudinal axis shows an extended and well-defined mass of fluid density at the tip of intussusception in the descending colon, as well as curvilinear calcification on the cystic lesion wall (Yellow arrow). (C); The longitudinal axis representing a gas-fluid level in dilated proximal loops is indicative of a small intestinal obstruction (Yellow arrow).

Colonoscopy demonstrated a collapsed lumen with an irregular polypoid mass measuring approximately 7 × 6 cm, along with two large sessile polyps, each approximately 2 cm in diameter, located 40 cm from the anal verge. Biopsies were obtained from these lesions. However, advanced imaging studies, such as MRI enterography, DBE (double-balloon enteroscopy), or CE (capsule endoscopy), were not available in our setting.

The patient's course was notable for clinical deterioration with worsening abdominal pain, distention and persistent vomiting. She underwent an exploratory laparotomy, which revealed an intussuscepted mass in the descending colon, a dilated proximal bowel loop, and a collapsed distal segment. A small intraluminal mass was identified in the proximal ileum, along with multiple mesenteric lymphadenopathies. Segmental resection of the affected bowel with an adequate safety margin was done and a left hemicolectomy accompanied by end-to-end colocolic anastomosis was performed (Figure 4).

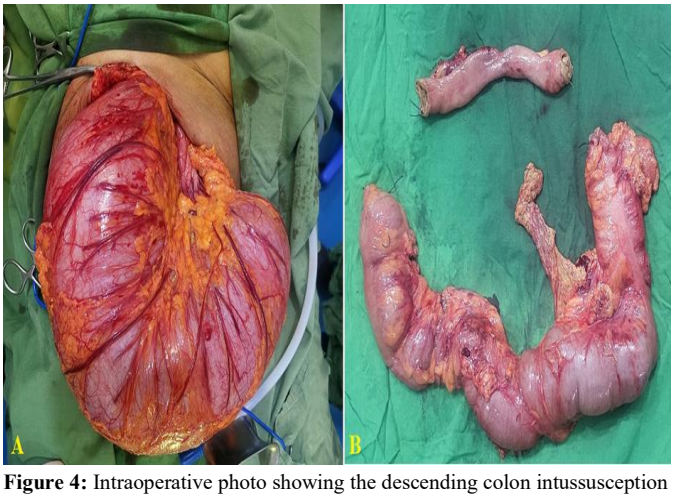

Figure 4: Intraoperative photo showing the descending colon intussusception (A), resected bowel lesion after left hemicolectomy accompanied by end-to-end colo-colic anastomosis (B).

Histopathological examination revealed existence of hamartomatous polypoid formations characterized by the proliferation of smooth muscle bundles, which were lined with regular mucosecreting cylindrical epithelium and hyperplastic glands, all devoid of any signs of malignant transformation, accompanied by reactive lymphadenopathy, which was suggestive of Peutz-Jeghers syndrome (Figure 5).

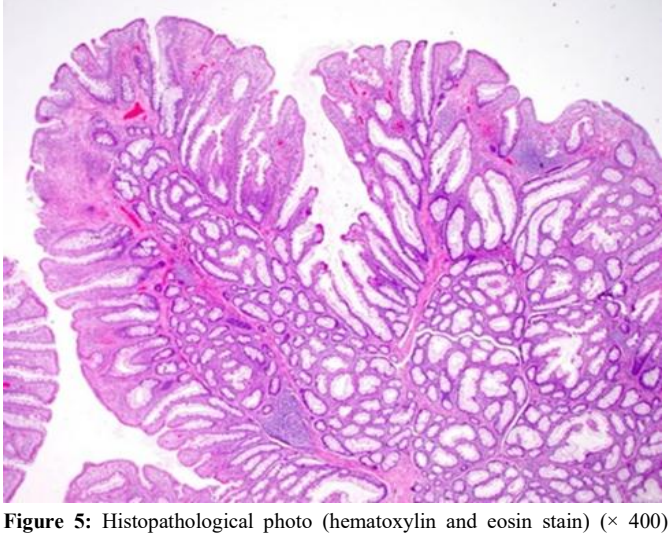

Figure 5: Histopathological photo (hematoxylin and eosin stain) (× 400) showing a polyp with proliferation of the smooth muscle bundle without signs of malignancy.

The patient had an uneventful recovery, with symptomatic improvement. She was discharged for four days post-operatively, with outpatient follow-up and periodic GI follow-up was recommended. A genetic study to detect mutations in the STK11 tumor suppressor gene was recommended. However, these testing, unfortunately, was unavailable in our settings.

Her familial constituents have subsequently been referred to as a colonoscopy due to the absence of any previous colonoscopic evaluations among them. Various tests, including esophagogastroduodenoscopy, mammography, and breast ultrasonography, found no pathological abnormalities. At one year of follow up, she has no clinical signs of recurrence.

Discussion

Intussusception is a common complication of Peutz-Jeghers syndrome and can be one of its presenting manifestations. [5] Notably, the majority of these cases involve the small intestine, particularly the jejunum and ileum, which have the highest polyp density [6]. Conversely, colo-colonic intussusception, as seen in our case, is exceedingly rare and has been scarcely documented in the literature. [4,7,8] These cases may be misdiagnosed or underrecognized due to their rarity. The World Health Organization (WHO) proposed diagnostic criteria for PJS, which include the presence of ≥ 3 hamartomatous polyps confirmed histopathologically. However, any polyp or mucocutaneous pigmentation, along with a positive family history of PJS, suffices to establish the diagnosis. [6] In our case, the diagnosis was established based on the presence of hamartomatous small intestinal polyps and mucocutaneous pigmentation across the perioral and buccal mucosa. There was no family history of PJS in this patient, suggesting a possible de novo mutation, which has been previously reported among PJS patients.

The gastrointestinal manifestations of PJS exhibit notable variability. Prior reports indicate that up to 50% of patients are diagnosed incidentally or as part of familial screening. Other clinical manifestations include intermittent abdominal discomfort or rectal bleeding due to polyp ulceration or intussusception. [7,9] While intussusception is rare among the adult population, it has been reported in up to 69% of PJS patients, presenting non-specifically with vague abdominal pain or findings of intestinal obstruction. [10] In our case, the patient reported occasional abdominal pain and frequent rectal bleeding. These findings, along with notable signs of abdominal tenderness and a palpable abdominal mass on physical examination, are consistent with colo-colonic intussusception, which typically presents more indolently. [11] In our case, despite the patient's symptomatic presentation spanning several years, the delayed diagnosis can be attributed to several factors: inadequate physician awareness regarding the syndrome and its diverse clinical manifestations, coupled with socioeconomic constraints, including limited access to advanced diagnostic tools such as imaging and endoscopy, particularly within primary care settings. Therefore, to mitigate future diagnostic delays and optimize patient outcomes, it is recommended that clinical practices undergo refinement. This includes a strengthened emphasis on comprehensive clinical evaluations and the integration of advanced diagnostic capabilities, particularly within resource-constrained environments.

Given the less specific clinical presentation, radiologic examination with abdominopelvic CT scan is the most effective imaging modality. This approach provides morphological and structural evaluation of polyps and concomitant obstructive findings. Additionally, it can differentiate between lead-point and non-lead-point intussusception, which is crucial for determining appropriate therapy and reducing the likelihood of unnecessary surgery. [12] Classic radiological findings of intussusception include crescentic attenuation of mesenteric fat within the intussusception, resulting in a characteristic tri-layered appearance. Enhanced vascular structures within the mesenteric adipose tissue may also be observed, as seen in our patient. [3,8]

The management of colo-colonic intussusception in PJS patients typically requires surgical intervention, particularly when a lead point, such as a small bowel polyp, is identified. This approach has been endorsed by the American Gastroenterological Association. [13] Radiological or endoscopic reduction is considered inappropriate for cases of intussusception secondary to PJS. [14] During laparotomy, intraoperative enteroscopy is recommended to facilitate the identification and removal of additional PJS polyps in the small bowel. [3,14] There is a lack of definitive consensus guidelines for the management of PJS polyps, necessitating a tailored approach for each case. However, in instances of significant hemorrhage or intussusception, polypectomy is warranted. [8,12] Colonoscopy should be employed to excise polyps, particularly those that are large and singular and within accessible range, while enterotomy should be utilized to surgically remove short segments of the intestine that are heavily affected. [3,5,8] In our patient, pre-operative colonoscopy was performed to provide further therapeutic assessment.

Endoscopic management of PJS necessitates polyp resection, frequently employing snare polypectomy for diminutive lesions. Larger or concerning polyps warrant Endoscopic Mucosal Resection (EMR) or Endoscopic Submucosal Dissection (ESD). EMR is indicated for sizable polyps exhibiting potential for early-stage malignancy or high-grade dysplasia, while ESD is reserved for lesions with elevated suspicion of submucosal invasion or confirmed early-stage carcinoma, facilitating precise staging and complete en bloc resection. [15] Contraindications encompass bleeding diatheses, patient non-compliance, and significant cardiorespiratory compromise. EMR/ESD are judiciously employed when substantial perforation risk exists, in the context of prior surgical intervention, or when lesion dimensions preclude effective intervention. [16] Regular endoscopic surveillance is paramount, and a multidisciplinary approach, integrating the expertise of gastroenterologists, surgeons, and pathologists, is deemed essential for optimal patient care. [15,16]

Individuals with PJS exhibit an increased predisposition to various malignancies, including those of the breast, gastrointestinal (GI) tract, pancreas, stomach, small intestine, liver, lung, testicles, ovaries, cervix, and uterus. [5,8,12] In the absence of adequate medical surveillance, the cumulative lifetime risk for all malignancies can reach up to 93%. GI cancers are the most prevalent malignancies among individuals with PJS, accounting for 63% by the age of 70. [17] Therefore, screening for GI malignancies should commence as early as 8–10 years of age with upper and lower endoscopy, allowing for direct visualization, tissue sampling, or polypectomy. [8,12] Our patient colonic intussusception within the descending colon, attributable to a colonic hamartoma. In addition to GI surveillance, individuals with PJS should consider annual mammograms and breast MRI starting at age 25–30, ovarian cancer screening with ultrasound and CA-125 at age 25–30, and annual pancreatic imaging (ultrasound or MRCP). [8, 12] Genetic testing for the STK11 gene should be conducted for all newly diagnosed patients and their families; positive results may warrant earlier and more frequent surveillance. [18] These testing, unfortunately, were unavailable in our settings. A multidisciplinary team management and tailored protocols according to risk and medical guidelines are recommended.

Conclusion

Colo-colonic intussusception is a rare manifestation of PJS that can present indolently with chronic abdominal pain. An elevated index of suspicion is essential for timely diagnosis, particularly in patients with chronic, refractory abdominal symptoms or characteristic pigmentation findings. Early identification, coupled with proactive polyp management and surveillance, is critical to preventing complications and optimizing clinical outcomes.

Consent for Publication:

Written informed consent was obtained from the patient for the 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 upon request.

Ethical Approval:

Ethical approval for this study was provided by the Research Ethics Committees.

Patient Perspective:

Throughout her course of treatment, the patient expressed satisfaction with the care she received.

Patient Consent for Case Report Publication

The patient has been thoroughly informed about the nature and purpose of the case report, including the potential for her clinical information and images to be published in a medical journal. She has provided her voluntary, written informed consent for the publication of her anonymized medical data, ensuring her privacy and confidentiality are maintained. The patient understands that her identity will not be disclosed, and all identifying information will be omitted or anonymized to protect her privacy.

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Conflict of Interest

Authors declare no conflict of interest.

Data availability

Data are available upon reasonable request.

All authors meet the ICMJE criteria for authorship and agree to be accountable for all aspects of the work.

ORCID

Qasem Alyhari 0000-0003-1349-0945
Saleh Al-Wageeh 0000-0001-6132-623X
Faisal Ahmed 0000-0001-7188-2715

References

[1] Fraser JD, Briggs SE, St. Peter SD, De Petris G, Heppell J. Intussusception in the adult: an unsuspected case of Peutz–Jeghers syndrome with review of the literature. Familial cancer. 2009 Jun;8(2):95-101. https://doi.org/10.1007/s10689-008-9212-x

[2] Shah ZA, Zeb M, Ilyas M, Hamid H, Fatima K, Batool M, Abbas M. Peutz–Jeghers syndrome with polyps in the stomach, duodenum, and small and large intestine: a case report. Journal of Medical Case Reports. 2024 Mar 5;18(1):86. https://doi.org/10.1186/s13256-023-04335-9

[3] Pham HD, Nguyen TH, Tran TQ, Bui VG, Duc PH, Hoa NT, Van GB. Multidetector Computed Tomography Findings of Acute Abdominal Intussusception Due to Peutz–Jeghers Syndrome. Cureus. 2021 Jul 15;13(7). https://doi.org/10.7759/cureus.16401

[4] Mabrouk MY, Guelil A, Bouzayan L, Slimi Y, Jabi R, Bouziane M. Intestinal occlusion revealing Peutz Jeghers syndrome: A rare case report. International Journal of Surgery Case Reports. 2024 Sep 1;122:110128. https://doi.org/10.1016/j.ijscr.2024.110128

[5] Duan SX, Wang GH, Zhong J, Ou WH, Fu MX, Wang FS, Ma SH, Li JH. Peutz–Jeghers syndrome with intermittent upper intestinal obstruction: a case report and review of the literature. Medicine. 2017 Apr 1;96(17):e6538. http://dx.doi.org/10.1097/MD.0000000000006538

[6] Barhmji H, Alsalehi A, Kammasha A, Alkheder A. Uncommon manifestation of Peutz-Jeghers syndrome: a case of jejuno-jejunal intussusception and volvulus leading to small bowel obstruction. Journal of Surgical Case Reports. 2024 May;2024(5):rjae335. https://doi.org/10.1093/jscr/rjae335

[7] Jaremko JL, Rawat B. Colo-colonic intussusception caused by a solitary Peutz-Jeghers polyp. The British Journal of Radiology. 2005 Nov 1;78(935):1047-9. https://doi.org/10.1259/bjr/25000606

[8] Shah J, Sunkara T, Xiao P, Gaduputi V, Reddy M, Razia S. Peutz-Jeghers syndrome presenting as colonic intussusception: a rare entity. Gastroenterology research. 2018 Apr 7;11(2):150. https://doi.org/10.14740/gr964e

[9] Chen XD, Yu YY, Yang L, Rui YY, Zhou ZG. Duodenal Intussusception due to a giant Neuroendocrine carcinoma in a patient with Peutz–Jeghers syndrome: case report and systematic review. European journal of gastroenterology & hepatology. 2012 Jun 1;24(6):722-6. https://doi.org/10.1097/meg.0b013e328351c1df

[10] Latchford AR, Clark SK. Gastrointestinal aspects of Peutz-Jeghers syndrome. Best Practice & Research Clinical Gastroenterology. 2022 Jun 1;58:101789. https://doi.org/10.1016/j.bpg.2022.101789

[11] Wang N, Cui XY, Liu Y, Long J, Xu YH, Guo RX, Guo KJ. Adult intussusception: a retrospective review of 41 cases. World journal of gastroenterology: WJG. 2009 Jul 14;15(26):3303. https://doi.org/10.3748/wjg.15.3303

[12] Klimkowski S, Ibrahim M, Ibarra Rovira JJ, Elshikh M, Javadi S, Klekers AR, Abusaif AA, Moawad AW, Ali K, Elsayes KM. Peutz–jeghers syndrome and the role of imaging: Pathophysiology, diagnosis, and associated cancers. Cancers. 2021 Oct 13;13(20):5121. https://doi.org/10.3390/cancers13205121

[13] T. Chand J, R R, Ganesh MS. Adult intussusception: a systematic review of current literature. Langenbeck's Archives of Surgery. 2024 Jul 31;409(1):235. https://doi.org/10.1007/s00423-024-03429-2

[14] Latchford A, Cohen S, Auth M, Scaillon M, Viala J, Daniels R, Talbotec C, Attard T, Durno C, Hyer W. Management of Peutz-Jeghers syndrome in children and adolescents: a position paper from the ESPGHAN Polyposis Working Group. Journal of pediatric gastroenterology and nutrition. 2019 Mar;68(3):442-52. https://doi.org/10.1097/MPG.0000000000002248

[15] Syarifuddin E, Masadah R, Lusikooy RE, Uwuratuw JA, Faruk M. Peutz-Jeghers syndrome in a woman presenting as intussusception: A case report. International Journal of Surgery Case Reports. 2021 Feb 1;79:286-90. https://doi.org/10.1016/j.ijscr.2021.01.053

[16] Raju GS. Complications of endoscopic mucosal resection and endoscopic submucosal dissection. Techniques in Gastrointestinal Endoscopy. 2011 Jan 1;13(1):91-4. https://doi.org/10.1016/j.tgie.2011.02.018

[17] Edmondson E, Mihalopulos G, Kwok PE, Dellert A, Olsen L, Zhang ZJ, Mihalopulos GJ, Kwok PE, Dellert AA. A case of solitary Peutz-Jeghers syndrome leading to chronic small bowel obstruction due to intussusception from a large hamartomatous polyp. Cureus. 2023 Apr 17;15(4). https://doi.org/10.7759/cureus.37679

[18] Wagner A, Aretz S, Auranen A, Bruno MJ, Cavestro GM, Crosbie EJ, Goverde A, Jelsig AM, Latchford AR, van Leerdam ME, Lepisto AH. The management of Peutz–Jeghers syndrome: European hereditary tumour group (EHTG) guideline. Journal of clinical medicine. 2021 Jan 27;10(3):473. https://doi.org/10.3390/jcm10030473