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Adult Intussusception as a Presentation of Peutz-Jeghers Syndrome: A Case Report

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Abstract

Background: Adult intussusception is a rare cause of bowel obstruction and is usually associated with an identifiable pathological lead point. Peutz–Jeghers syndrome (PJS) is a rare hereditary disorder characterized by mucocutaneous pigmentation and multiple hamartomatous gastrointestinal polyps, with intussusception being one of its most frequent surgical complications. **Case:** A 20-year-old male presented with intermittent abdominal pain and recurrent vomiting for one month. Imaging studies demonstrated jejuno-jejunal intussusception. Emergency exploratory laparotomy confirmed jejuno-jejunal intussusception with multiple pedunculated polyps acting as lead points. Surgical reduction with segmental resection was performed. **Conclusion:** PJS should be considered in young adults presenting with intussusception, particularly in the presence of mucocutaneous pigmentation. Early diagnosis allows appropriate surgical management and long-term surveillance to prevent recurrence and malignancy-related complications.

Keywords: Bowel obstruction, intussusception, LKB1/STK11 mutation, Perioral pigmentation, Peutz-Jeghers syndrome, Emergency Medicine

Introduction

Adult intussusception is an uncommon cause of intestinal obstruction, constituting approximately 1% of all bowel obstructions and about 5% of all intussusception cases. Unlike pediatric intussusception, which is often idiopathic, adult intussusception usually has an identifiable

pathological lead point, frequently associated with benign or malignant lesions. Peutz–Jeghers syndrome (PJS) is a rare autosomal dominant inherited disorder characterized by distinctive mucocutaneous hyperpigmentation and the development of multiple hamartomatous polyps throughout the gastrointestinal tract, predominantly affecting the small intestine. Individuals with PJS are at increased risk of recurrent gastrointestinal complications, including chronic or acute bleeding, anaemia, bowel obstruction, and intussusception. Recurrent intussusception is one of the most common surgical emergencies in patients with PJS and often represents the initial manifestation of the disease. We present a case of adult intussusception secondary to Peutz–Jeghers syndrome, highlighting its clinical presentation, diagnostic challenges, and management considerations.[1]

Case Report

A 20-year-old male presented to the emergency department with complaints of intermittent abdominal pain and recurrent episodes of vomiting for one month. There was no history of altered bowel habits or overt gastrointestinal bleeding. On general examination, multiple hyperpigmented macules were noted on and around the lips. Abdominal examination revealed a distended abdomen with diffuse tenderness.

Laboratory evaluation revealed marked hepatic dysfunction. Serum bilirubin was significantly elevated with a total/direct/indirect fraction of 25.3/16.1/9.2 mg/dL, indicating predominantly conjugated hyperbilirubinemia. Liver enzymes were deranged, with SGOT and SGPT levels of 63 U/L and 96 U/L respectively, along with an elevated alkaline phosphatase of 194 U/L, suggestive of a mixed hepatocellular–cholestatic pattern of liver injury. Serum albumin was reduced to 2.5 g/dL, reflecting impaired synthetic liver function. Coagulation parameters were prolonged with a prothrombin time of 23.1 seconds and an INR of 2.0, further indicating significant hepatic synthetic dysfunction.

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Iron studies demonstrated markedly elevated serum ferritin (2,671 ng/mL) and transferrin saturation (85%), with serum iron of 142 µg/dL, suggesting significant iron overload. Serum ammonia was elevated at 166 mg/L, consistent with hepatic insufficiency and risk of hepatic encephalopathy. Immunological workup revealed elevated serum IgG levels (1813 g/L) with positive anti–smooth muscle antibodies (ASMA), while antimitochondrial antibody (AMA), antinuclear antibody (ANA blot), and anti–liver kidney microsomal (anti-LKM) antibodies were negative, supporting an autoimmune-mediated liver process. Wilson's disease and hereditary hemochromatosis were excluded by negative ceruloplasmin and HFE gene testing, respectively.

Overall, the biochemical and immunological profile was suggestive of severe liver dysfunction with features favouring autoimmune hepatitis and secondary iron overload.

Ultrasonography of the abdomen revealed a well-defined bowel-within-bowel appearance measuring approximately 10.1 × 5 × 7 cm in the left para-umbilical region, suggestive of intussusception.

Contrast-enhanced computed tomography of the abdomen confirmed the diagnosis of jejuno-jejunal intussusception with features of bowel obstruction.

The patient underwent emergency midline laparotomy. Intra-operatively, a jejuno-jejunal intussusception was identified. Manual reduction was successfully performed, and the affected segment revealed multiple pedunculated intraluminal polyps acting as lead points. Segmental resection with removal of multiple polyps was carried out. The resected specimen was sent for histopathological examination and STK11 (LKB1) gene analysis.

Histopathology revealed hamartomata's polyps consistent with Peutz–Jeghers type. On further evaluation, the patient gave a positive family history of similar mucocutaneous pigmented lesions on the maternal side. Based on the clinical features, operative findings, histopathology,

and family history, a final diagnosis of Peutz–Jeghers syndrome was made. The postoperative period was uneventful, and the patient was discharged with advice for genetic counselling and long-term endoscopic surveillance.

Discussion

Peutz–Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by mucocutaneous lentiginosis, multiple hamartomatous gastrointestinal polyps, and a markedly increased lifetime risk of malignancy. The estimated prevalence is 1 in 50,000 to 200,000 individuals, with most patients becoming symptomatic in childhood or early adulthood. However, adult presentation with acute intestinal obstruction or intussusception, as in the present case, is uncommon and often leads to delayed diagnosis.[2]

Giri et al. reported a young adult with Peutz–Jeghers syndrome presenting with recurrent abdominal pain and multiple small-bowel intussusceptions. Their patient, similar to the present case, was in early adulthood and presented emergently with features of intestinal obstruction. Both cases underline that in India, PJS is frequently diagnosed only after an acute surgical emergency rather than during routine screening.[3]

Anuragi et al. also described a young patient who presented with acute intestinal obstruction due to PJS polyps, reinforcing that emergency presentation remains the most common clinical scenario in Indian reports.[4]

Bhat et al. highlighted the importance of mucocutaneous pigmentation as an early diagnostic clue, reporting prominent perioral and buccal lentiginosis in their case.[5]

Similarly, Sharma et al., in an Indian pediatric surgical series, noted that almost all patients had characteristic pigmentation, but it was often overlooked until careful physical examination was performed.[6]

Searchavilli et al. conducted one of the earliest Indian molecular studies on Peutz–Jeghers syndrome and reported novel STK11 mutations, while also observing that a proportion of clinically confirmed patients lacked detectable mutations, highlighting genetic heterogeneity and the limitations of molecular testing alone. Similar to their findings, the diagnosis in the present case was established primarily on the basis of classical clinical features, including periorificial lentiginosis, hamartomatous gastrointestinal polyps, and presentation with intussusception, supported by histopathology rather than genetic confirmation. This similarity reinforces the observation that, in the Indian setting, Peutz–Jeghers syndrome can be reliably diagnosed using clinical and pathological criteria, particularly where access to genetic testing is limited.[7]

Conclusion

This case highlights the importance of considering Peutz–Jeghers syndrome in young adults presenting with intussusception, particularly when accompanied by characteristic mucocutaneous pigmentation. As demonstrated in our patient, PJS in the Indian setting often comes to clinical attention only after an acute surgical emergency, rather than through proactive surveillance. Prompt surgical intervention is essential in adult intussusception due to the high likelihood of a pathological lead point.[8] Furthermore, this case reinforces that a confident diagnosis of PJS can be made on the basis of classical clinical features and histopathology, even in the absence of molecular confirmation. Given the markedly increased lifetime risk of gastrointestinal and extra-intestinal malignancies, early recognition, genetic counselling, family screening, and institution of structured long-term surveillance are crucial to reduce morbidity and improve outcomes in patients with Peutz–Jeghers syndrome.

Recommendations

Young patients presenting with intussusception or recurrent abdominal pain should be carefully evaluated for mucocutaneous pigmentation to

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allow early recognition of Peutz–Jeghers syndrome.[9] Adult intussusception warrants prompt surgical management due to the high likelihood of a pathological lead point. Once diagnosed, patients should be enrolled in a structured lifelong surveillance program with regular endoscopic evaluation and age-appropriate cancer screening. Genetic counselling and family screening are essential to identify at-risk relatives and to reduce long-term morbidity and malignancy-related mortality.[10]

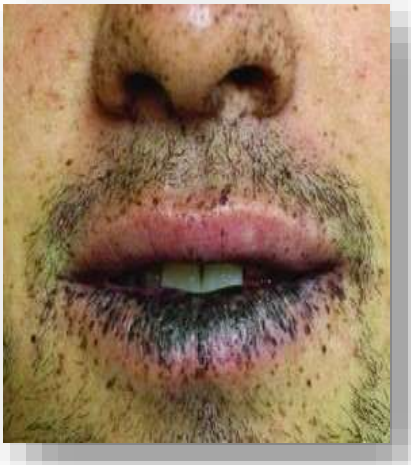


Fig 1: Depicts multiple hyperpigmented macules on and around the lips.

Fig 2 : Contrast-enhanced computed tomography of abdomen confirming the diagnosis of jejuno-jejunal intussusception.

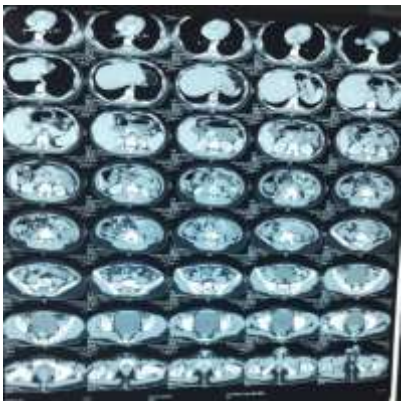
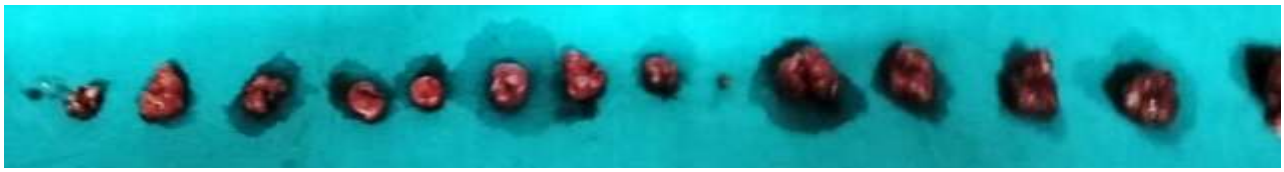


Fig3: Multiple polyps post-segmental resection



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