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Case Report

Double Intussusceptions in Peutz-Jeghers Syndrome Patient: A Case Report

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Abstract

Background: *Peutz-Jeghers Syndrome is one of the hereditary gastro-intestinal cancer syndrome with characteristic mucocutaneous pigmentation and histologically distinctive hamartomatous polyps in gastro-intestinal tract. Although it is characteristically benign hamartomatous polyp, majority of affected individuals develop symptoms starting from their second decades. We reported a known Peutz-Jeghers Syndrome case developed recurrent polyps leading to double intussusceptions required bowel resection. Multidisciplinary management and patient compliance to surveillance regime are important in managing PJS patients with potential gastro-intestinal tract complications and relative high risk of developing syndrome specific cancers.*

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Introduction

Peutz-Jeghers Syndrome (PJS) is one of the familial polyposis syndrome with triad of classical mucocutaneous pigmentations, benign hamartomatous polyps with rare potential of malignant transformation and autosomal dominant inheritance. Jeghers first described the disease with correlation to the autosomal dominance inheritance in 1949. (1)

The prevalence of PJS is reported to be rare with 1 in 100,000 people. (2) Its prevalence is unknown in Malaysia although it was first reported in 1978 by Joishy et al. (3) The patient has typical mucocutaneous pigmentation and intestinal obstruction. Note worthily, the patient came with recurrent intussusception required small bowel resections. A small group of them presented with recurrent intussusceptions signifying the important features of the polyps of which progression by segmental spurts with period of quiescence lasting for months. (4)
We hereby reported an interesting case of double intussusceptions in a known case of PJS patient.

Case Presentation

We reported a 24 years old Malaysian lady with background history of PJS She was diagnosed with PJS when she was 12 years old with small bowel intussusception. Emergency laparotomy, small bowel resection and primary anastomosis was done for the small bowel intussusception.

She presented with symptoms of intestinal obstruction and clinically distended abdomen with tenderness over right iliac fossa. Her blood investigations were non-remarkable and abdominal radiography showed paucity of small bowel gas and absence of rectal gas. (Figure 1) Due to her obstructive symptoms and hostile abdomen, contrasted computed tomography (CT) scan showed 2 intussusceptions: one at right lumbar represented ileo-colic intussusception and one at left lumbar represented jejuno-jejunal intussusception. (Figure 2)

We went to emergency laparotomy due to her obstructive symptom correlated with CT scan findings. Intra-operatively, we noted ileo-colic intussusception from cecum up to proximal transverse colon and jejuno-jejunal intussusception with intramural polyp. (Figure 3) We proceeded to right hemicolectomy, primary ileo-colic anastomosis, wedge resection of jejuno-jejunal intussusception and primary anastomosis. Gross specimen of right hemicolectomy revealed 2 pedunculated polyps both measured 3cm respec-

tively from small bowel while 1 pedunculated polyp was seen at the wedge resection of jejunum. As for microscopic examination, all polyps behaved as Peutz -Jeghers polyps without evidence of dysplasia or malignancy.

Her post-operative period was uncomplicated and she was discharged home on day 8 post operatively.

Discussion

We described a young patient with recurrent intussusceptions 10 years from the first incident. PJS is a rare disease, however, its presentation is simply pathognomonic with its distinctive mucocutaneous pigmentation as presented by the patient. (Figure 4) Often, the diagnosis of the disease occurs when the complications such as intussusception, gastro-intestinal bleeding and intestinal obstruction arise.

Repeated abdominal surgeries are the major concern of the nature progression of the disease. Ninety percent of the PJS patients will have polyps in small intestine in their lifetime. (5) The intestinal polyp is often described as benign. However, PJS is associated with higher risk of gastrointestinal and non-gastrointestinal malignancies. (6) A meta-analysis has shown that an affected individual has 15 times relative risk of neoplasm in any body region as compared to general population. (7,8)

In order to avoid unnecessary repeated abdominal surgeries and risk of short bowel syndrome, an affected individual must comply to surveillance regime according to American College of Gastroenterology clinical guideline. The algorithm of management started with dedicated genetic testing of at-risk individuals and family members. Subsequently, systemic review and throughout physical examinations are important to rule out any syndrome specific cancers includes colon, stomach, small bowel, etc.

Upper GI endoscopy and colonoscopy should be done at the age of 8 years based on guideline and if presence of polyps, there should be repeated procedures every 3 years. If none are found, both examinations should be done at age of 18 and then every 3 years. (9) Our patient has her last upper and lower scope done the year before but did not show any

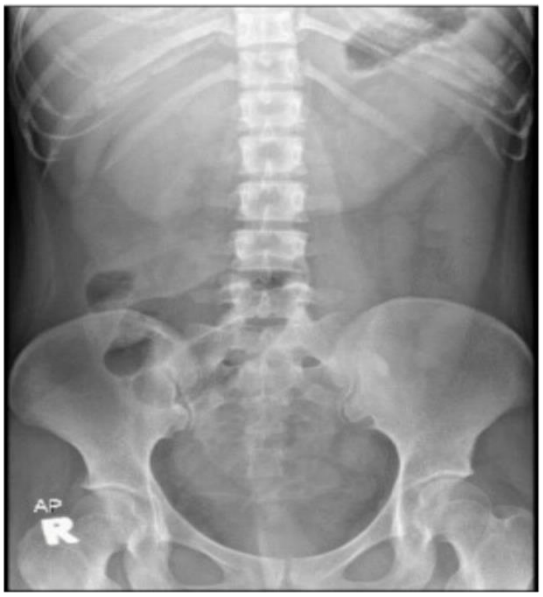


Figure 1. Demonstrated plain abdominal radiography with paucity of small bowel gas and absence of rectal gas.

abnormalities. Small bowels are often involved in 96% of affected individuals. (10) Hence, much efforts should be focus in surveillance of small bowels including video capsule endoscopy and CT enterography. (9)

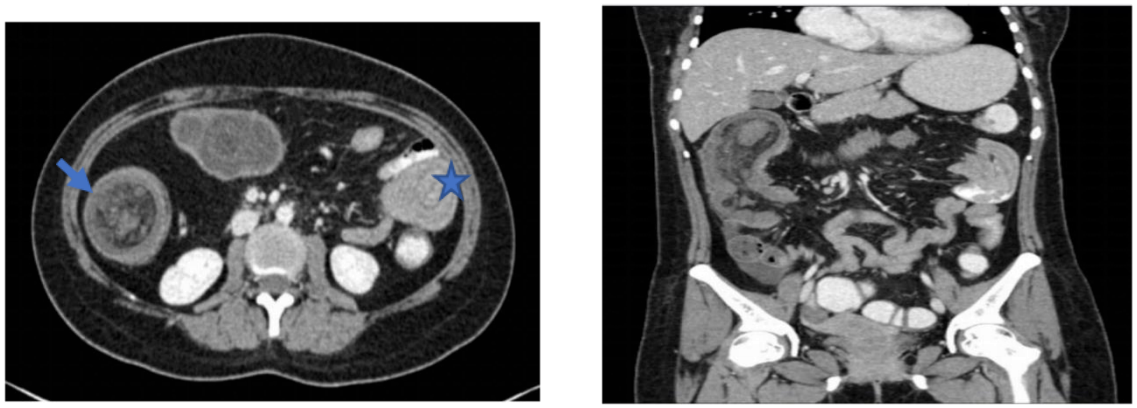
Prevention of short bowel syndrome is ultimately the aim of the management of intestinal polyps. Moving forward, as the patients presented with intestinal obstruction due to intramural polyp, careful examination of entire GI tract and removal of all identifiable polyps through endoscopic polypectomy are essential.

Advancement of technologies has allowed surveillance and non-operative removal of polyps. For examples, video capsule endoscopy, double balloon enteroscopy and CT enterography.

Conclusion

PJS is an unbending disease that will affect an individual lifetime psychologically. Timely diagnosis and family counselling are initial steps in the algorithm. Well-timed surveillance scheme will ensure early discovery of asymptomatic polyps and polypectomy if needed to prevent unnecessary bowel resection.

More modalities should be developed for surveillance of GI tract especially small bowel for early diagnosis and treatment of polyp without putting them into multiple abdominal surgeries. To date, PJS is still tough to be handle due to its rarity and high recurrences.



A) *B)*

Figure 2. (A) CT axial view demonstrated ileocolonic intussusception (arrow) and jejuno-jejunal intussusception (star). (B) CT coronal view demonstrated similar findings particularly showing the ileo-colic intussusception until proximal transverse colon.

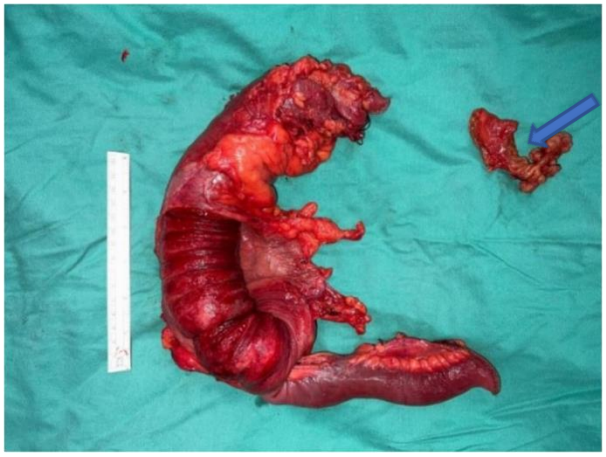


Figure 3. Specimen demonstrated ileocolonic intussusception
on the left and wedge resection of jejunum with intramural polyp on the right. (arrow)

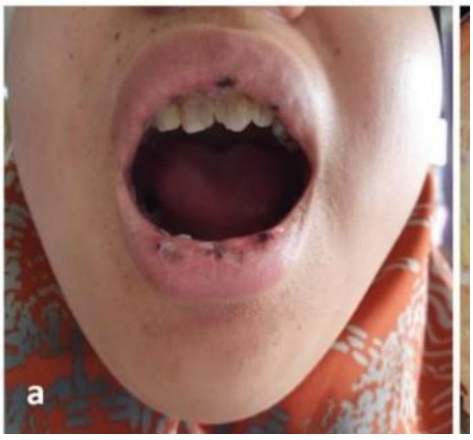


Figure 4. Pigmentation **at lips**

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