OMCR

*Oxford Medical Case Reports*, 2019;6, 274–277

**doi: 10.1093/omcr/omz051**
Case Report

CASE REPORT

# Sporadic Peutz–Jeghers syndrome: a rare cause of intussusception in a toddler with no medical history

Mhmmad Nassif[1], Bardisan Gawrieh[1,*], Aras Abdo[1], Zuheir Alshehabi[2] and Wajih Ali[1]

[1]Pediatric Surgery Department, Tishreen University Hospital, Lattakia, Syria, [2]Pathology Department, Faculty Of Medicine, Tishreen University, Lattakia, Syria

*Correspondence address. Pediatric Surgery Department, Tishreen University Hospital, Lattakia, Syria. Tel: +963-988568777; E-mail: gawriehb@gmail.com

## Abstract

Peutz–Jeghers syndrome (PJS) is an unusual hamartomatous polyposis of the gastrointestinal tract associated with melanocytic mucocutaneous hyperpigmentation. This research paper examines the case of an 18-month-old Syrian female who had been diagnosed with intussusception. The patient underwent laparotomy, and multiple small bowel polyps were found to act as the lead point. For this reason, small bowel resection (~15 cm), with end-to-end anastomosis, were performed. Although PJS diagnosis was histopathologically confirmed, the patient had no pigmented lesions on the face, the lower lip or the buccal mucosa and neither had any history of hospitalization or family history of the disease. This case was examined and is reported in the present study because PJS is rarely present at this early age when significant medical history is lacking.

## INTRODUCTION

Peutz–Jeghers syndrome (PJS) is an autosomal dominant disorder characterized by hamartomatous polyps and pigmented macules [1]. Hamartomatous polyps are most commonly found in the small intestine but may also be seen in the stomach and colon. The average age for the onset of gastrointestinal symptoms is 13 [2], and ~50% of patients will suffer from polyps complications—such as intussusception, obstruction and rectal bleeding—by the age of 20 [2, 3]. Common in more than 90% of affected individuals, mucocutaneous pigmented lesions tend to arise in infancy, occurring around the mouth, nostrils, perianal area, fingers and toes and may be the first signs of PJS [2, 3]. The most common symptom is recurrent colicky abdominal pain caused by transient intussusception or obstruction [2]. The diagnosis of PJS is assumed if any of the following criteria is met: (i) two or more PJS polyps; (ii) any number of PJS polyps and a history of PJS in a close relative; (iii) characteristic mucocutaneous pigmentation in an individual with a history of PJS in a close relative; or (iv) any number of PJS polyps with characteristic mucocutaneous pigmentation [4, 3]. The majority of individuals with PJS have been found to have genetic alterations in LKB1, and up to 25% of those diagnosed have *de novo* STK11 mutations with no family history of the disease [4]. PJS is associated with a progressively increasing lifetime risk of developing malignancies—by about 1% to 2% by the age of 20, more than 30% by 50 and more than 80% by 70 [4].

**Received:** January 16, 2019. **Revised:** March 24, 2019. **Accepted:** April 21, 2019

## CASE REPORT

An 18-month-old Syrian female baby was admitted to our hospital with complaints of acute abdominal pain, several episodes of non-bilious, non-bloody vomiting and red diarrhea. She had neither prior hospitalization nor any significant symptoms in her own medical history, and her family's history for PJS was negative. On admission, her vital signs were normal, and physical examination did not reveal any peculiar pigmented lesions. Abdominal examination showed generalized tenderness, diminished bowel sounds and a possible palpable epigastric mass. Blood analysis indicated low hemoglobin concentration of 9.4 g /dl (n:12 to16 g /dl), a mean cell volume of 63.2 F /L (N:72–100 F /L), but other initial laboratory results were normal. Abdominal ultrasonography, in turn, revealed typical 'target and pseudo-kidney' signs. Accordingly, the patient was diagnosed with small bowel intussusception (Fig. 1). Consequently, laparotomy was performed, and surgical examination detected jejunal palpable masses in the intussusceptum. Enterotomy revealed multiple polyps on the mucosal surface (Fig. 2), and small bowel resection (~15 cm), with end-to-end anastomosis, were performed. Postoperative course was uneventful. Histopathologic study was conducted on the resected portion, which consisted of a 15-cm specimen from the small bowel containing more than 100 pedunculated polyps on mucosal surface ranging from 1 to 2.5 cm in diameter (Fig. 3).

Microscopic examination of polyps showed proliferation of papillary and glandular formations lined by hyperplastic epithelial cells. Stroma was rich in smooth muscle fiber arising from the muscularis mucosae and infiltrated with patchy mononuclear inflammatory cell infiltrate (Fig. 4). Neither adenomatous change nor malignancy was observed after a thorough examination. On the basis of two or more PJS polyps, the diagnosis of PJS syndrome was confirmed.

## DISCUSSION

PJS is an autosomal dominant condition, which was first described by Peutz in 1921, and it was not until 1949 that Jeghers defined the relation between pigmented lesions and gastrointestinal polyposis, on the one hand, and an increased risk of carcinoma, on the other [5]. Our patient underwent laparotomy following clinical symptoms of intussusception—confirmed by ultrasonography, and PJS was diagnosed with the help of histopathologic evaluation of the excised bowel. Here we ought to mention that laparoscopy is increasingly the preferred approach for diagnosis and management of acute abdominal and can be performed safely for bowel resection

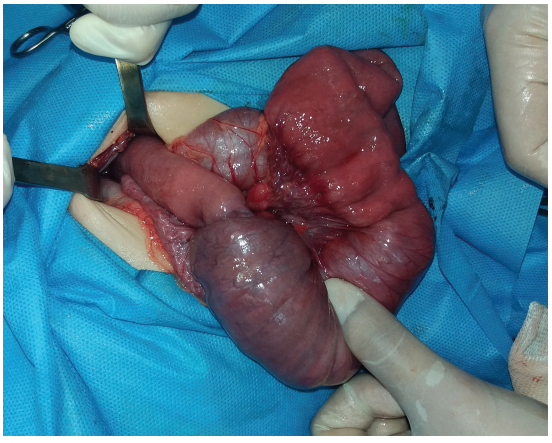

**Figure 1:** Small bowel intussusception.

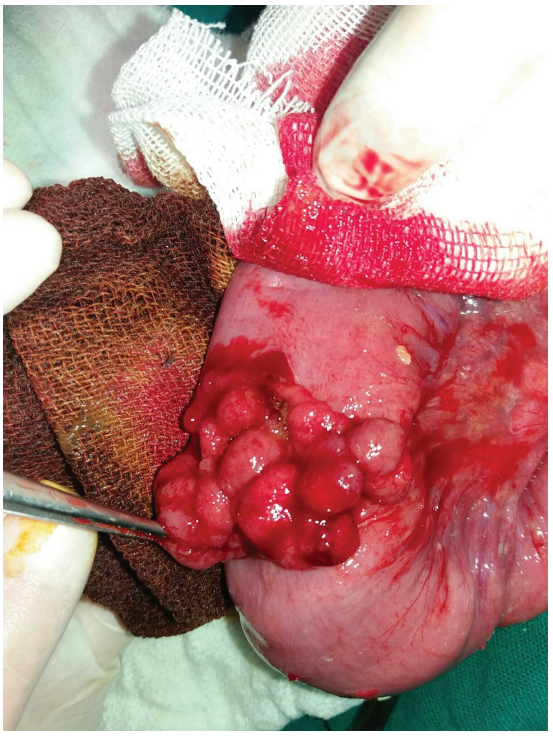

**Figure 2:** Enterotomy.

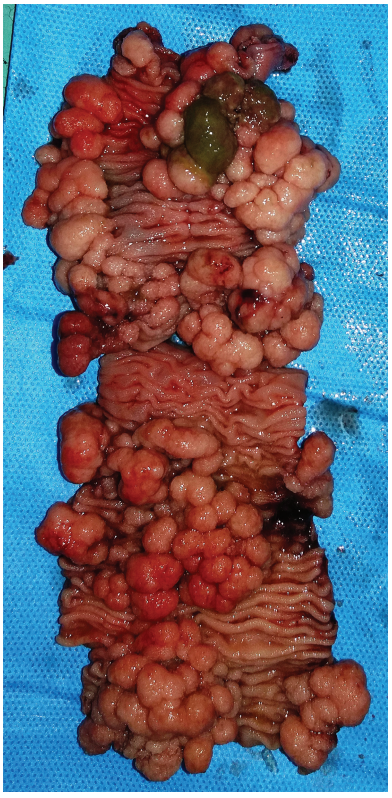

**Figure 3:** Multiple polyps on the mucosal surface.

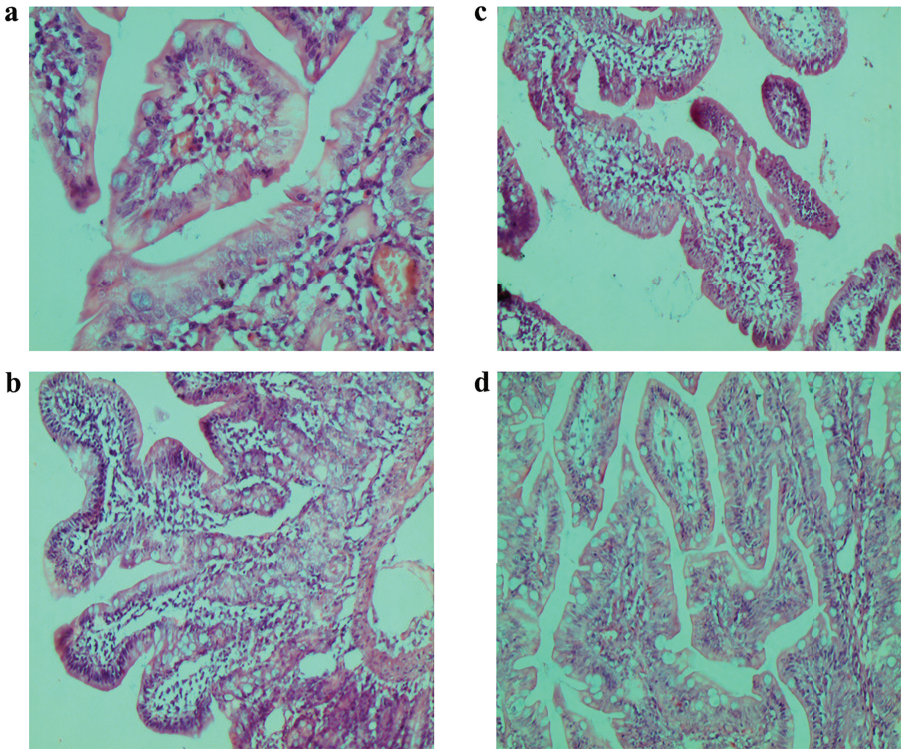


**Figure 4:** (**a**) Microscopic image; (**b**) microscopic image; (**c**) microscopic image.

and anastomosis. However, the procedure was not available in the hospital where the study was conducted. Gastrointestinal hamartomatous polyps are characteristic of PJS, and they are most commonly found in the small intestine but can also be found in other intestinal or extraintestinal sites [2, 4]. The median time to first presentation with polyps is about 11–13 years of age, and ~50% will have experienced intussusception, obstruction or rectal bleeding by the age of 20 [1, 4, 6]. Although the association between PJS and intussusception is not uncommon, this complication is rarely presented as a first clue of PJS. What is peculiar about our patient is that she presented with intussusception at the age of 18 months as the first complication of PJS.

Mucocutaneous pigmentation is typically found in over 90% of PJS cases but fades with age [4].The phenotype varies considerably from diffuse, dark freckling over the face to light pigmentation in characteristic areas [2]. Physical examination of our patient, however, did not reveal any pigmented lesions. PJS is caused by mutations in the STK11 gene (also called LKB1) located at 19p13.3, as it encodes a serine/threonine kinase that functions as a tumor suppressor [7, 8]. Mutations in this gene are detected in 50—90% of individuals with PJS [2]. Although PJS is inherited in an autosomal dominant fashion, up to 25% of cases do not have any family history [9, 10]. Similarly, our patient did not have history of PJS in a close relative, and thus her case should be categorized as sporadic PJS, especially given that the authors of this study lacked the means to conduct genetic testing of the case under examination.

PJS is associated with a significantly increased risk of both intestinal and extraintestinal cancer [9, 10]. It rises with age, by about 1% to 2% by the age of 20, more than 30% by 50 and more than 80% by 70 [4]. Clinical guidelines for the surveillance and management of PJS-affected individuals have been established; upper gastrointestinal endoscopy and colonoscopy should be performed every 2–3 years, starting from late teens. Additionally, small bowel visualization should start at the age of 8–10 and should be repeated every 2–3 years. Other screening procedures include clinical examination and specific imaging to screen for lesions of the breast, female genital tract, testis and pancreas [6]. However, management and follow-up methods for PJS patients continue to be somewhat controversial.

In conclusion, intussusception may be the first lead for PJS regardless of age, family history or pigmentation.

## CONFLICT OF INTEREST

None declared.

## FUNDING

The authors did not receive any funding to conduct this study.

## ETHICAL APPROVAL

No ethical approval was needed.

## CONSENT

Consent has been duly obtained.

## REFERENCES

1. Kopacova M, Tacheci I, Rejchrt S, Bures J. Peutz–Jeghers syndrome: diagnostic and therapeutic approach. *World J Gastroenterol* 2009;**15**:5397–408.

2. Gammon A, Jasperson K, Kohlmann W, Burt RW. Hamartomatous polyposis syndromes. *Best Pract Res Clin Gastroenterol* 2009;**23**:219–31.
3. Beggs AD, Latchford AR, Vasen HF, Moslein G, Alonso A, Aretz S, *et al*. Peutz–Jeghers syndrome: a systematic review and recommendations for management. *Gut* 2010;**59**:975–86.
4. Achatz MI, Porter CC, Brugières L, Druker H, Frebourg T, Foulkes WD, *et al*. Cancer screening recommendations and clinical management of inherited gastrointestinal cancer syndromes in childhood. *Clin Cancer Res* 2017;**23**:e107–14.
5. Jeghers H, McKusick VA, Katz KH. Generalized intestinal polyposis and melanin spots of the oral mucosa, lips and digits; a syndrome of diagnostic significance. *N Engl J Med* 1949;**241**:1031–6.
6. Rosty C. The role of the surgical pathologist in the diagnosis of gastrointestinal polyposis syndromes. *Adv Anat Pathol* 2018;**25**:1–13.
7. Mehenni H, Blouin JL, Radhakrishna U, Bhardwaj SS, Bhardwaj K, Dixit VB, *et al*. Peutz–Jeghers syndrome: confirmation of linkage to chromosome 19p13.3 and identification of a potential second locus, on 19q13.4. *Am J Hum Genet* 1997;**61**:1327–34.
8. Hemminki A, Tomlinson I, Markie D, Järvinen H, Sistonen P, Björkqvist AM, *et al*. Localization of a susceptibility locus for Peutz–Jeghers syndrome to 19p using comparative genomic hybridization and targeted linkage analysis. *Nat Genet* 1997;**15**:87–90.
9. Broadman LA, Thibodeau SN, Schaid DJ, Lindor NM, Mcdonnell SK, Burgart LJ, *et al*. Increased risk for cancer in patients with the Peutz–Jeghers syndrome. *Ann Intern Med* 1998;**128**:896–9.
10. Živković V, Pejović S, Nagorni A, Petrović B, Petrović A, Ilić I. Heredity hamartomatous gastrointestinal polyposis syndrome *Sci J Fac Med Niš* 2010;**27**:93–103.