PEUTZ-JEGHER'S POLYPOSIS: CHILDHOOD PRESENTATION OF ACUTE AND CHRONIC INTERMITTENT BOWEL OBSTRUCTION CAUSED BY INTESTINAL POLYPS IN RURAL SOUTH AFRICA: A CASE SERIES

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Keywords

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Abstract

Introduction: Peutz-Jegher's Syndrome (PJS) is a rare hereditary condition characterized by the development of gastrointestinal polyps and mucocutaneous freckling. These polyps can cause intussusception and acute bowel obstruction, requiring urgent intervention due to the high risk of bowel ischemia. This paper aims to report two different presentations of PJS in children who presented to the pediatric surgery department. One case involved the acute onset of bowel obstruction, while the other involved chronic intermittent bowel obstruction.
Case presentation: We present 2 cases of PJS that presented to our department. A 9-year-old boy with an acute history of symptoms of intestinal obstruction. Diagnostic laparoscopy revealed intussusception. We elected to convert to an open laparotomy and manual reduction of the intussusception. The polyp was resected, the bowel repaired, and the patient was subsequently discharged home and did not have any recurrence. The second was a 6-year-old boy with a 3-year history of abdominal pain and intermittent bowel obstruction. A large polyp was discovered and resected during laparotomy.
Conclusion: PJS may present differently in patients due to the size and

	location of the polyps. It is essential to maintain high clinical suspicion and refer these patients for long-term surveillance by a pediatric surgeon.
Abbreviations	PJS: Peutz-Jegher's GP: General practitioner CT: Computed tomography

INTRODUCTION

Peutz-Jegher's Syndrome (PJS) is an autosomal dominant condition characterized by the presence of multiple polyps in the gastrointestinal tract. The estimated incidence is 1 in 150,000. [1,2] It is associated with hyperpigmented mucocutaneous macules, which are usually present in the first year of life and commonly occur on the lips, mucous membranes of the mouth, hands, and feet, and occur in about 90% of cases. [3]

The etiology is unknown but postulated to be caused by germ-line mutations in the *STK11* (*LKB1*) on chromosome 19p13.3. [4] The STK11 is a tumor suppressor gene that regulates cell polarity, and it encodes serine/threonine kinase 11, which plays a crucial role in controlling the cell cycle. Mutations in STK11 are detected in 50% to 80% of families with PJS; for the remaining patients, the syndrome resulted from a de novo mutation. [5]

Patients with PJS are at risk of developing intussusception, which is a condition where the proximal part of the intestine telescopes into the distal intestine, causing intestinal obstruction, bowel edema, and necrosis, and can result in intestinal perforation. [6] We report two cases of PJS with contrasting clinical presentations, the first being an acute emergency and the other presenting as chronic intermittent intestinal obstruction.

CASE REPORT

Case 1: Patient A

Patient A first presented to our unit with a history of an incarcerated umbilical hernia, which was reduced and subsequently booked for elective umbilical hernia repair. The day before the surgery, the patient presented with symptoms and signs of intestinal obstruction, which included bilious vomiting associated with colicky abdominal pain. On clinical examination,

the hernia was not incarcerated. There was no history of hematemesis, mucoid stools mixed with blood, or any other clinical signs associated with intussusception.

The patient had previously been seen by multiple general practitioners (GP) with symptoms of abdominal pain, associated with vomiting since 2016. The diagnosis given by the GP's varied from constipation to colic and psychosomatic pain. The patient had no family history of congenital abnormalities, including PJS.

On examination, the patient appeared alert, with mild pallor, and was comfortable on room air. Obvious mucocutaneous lesions were noticeable on the lower lip, mouth, palms, and feet. (Fig 1) On abdominal examination, there was an obvious umbilical hernia, which was associated with abdominal distension and a positive 'Dance sign' in the right Iliac fossa. The rectal exam was normal, and other systems and vitals were normal. An ultrasound was ordered, and intussusception was diagnosed before surgical intervention.

A diagnostic laparoscopy was performed, and intussusception was visualized. (Fig 2) Due to difficulties encountered while doing a laparoscopic reduction and the associated colonic perforation, the operation was converted to laparotomy, and manual reduction of an ileocolic intussusception was performed. An umbilical hernia was also repaired in the same setting, and the patient was discharged 3 days later.

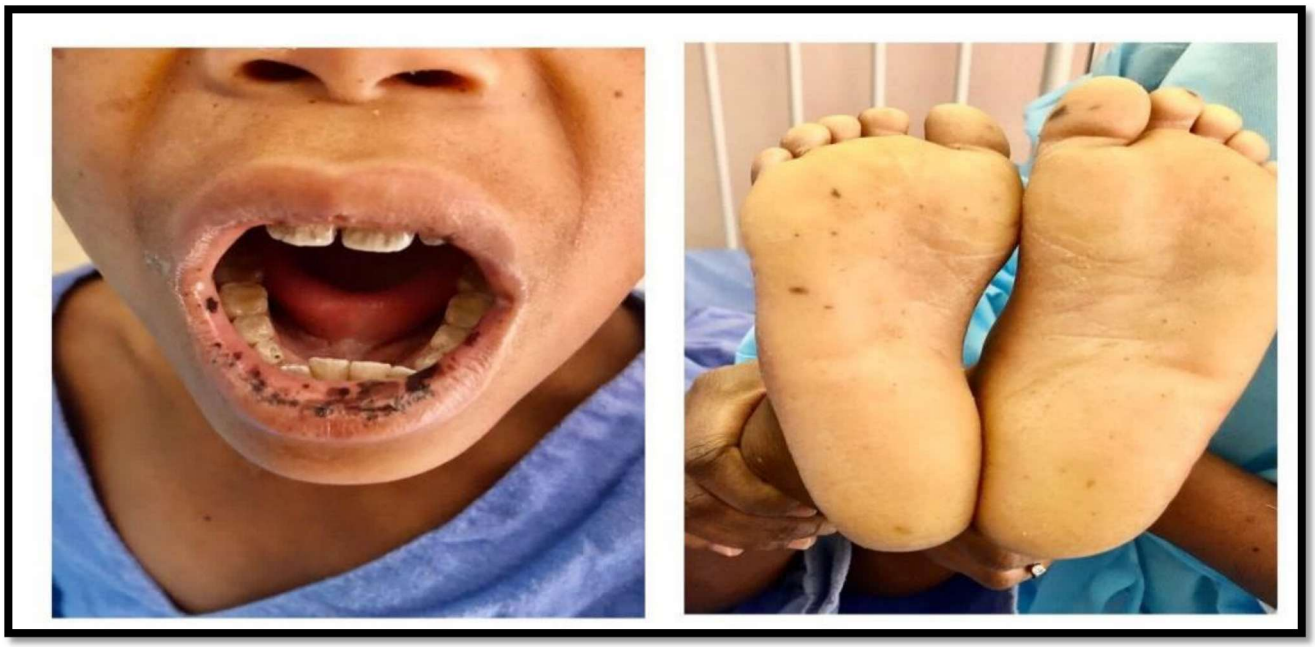

Fig 1. Mucocutaneous lesions

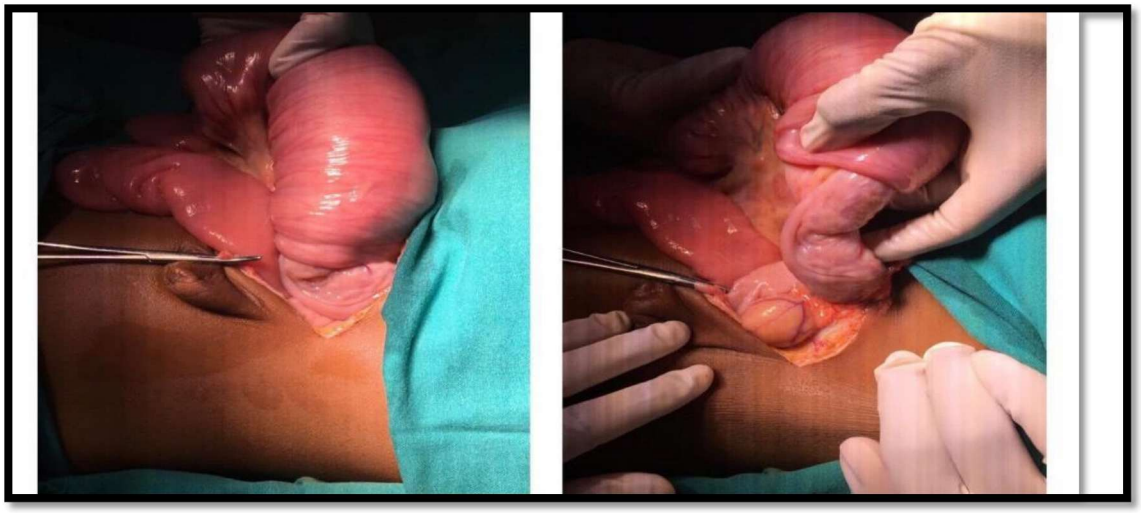

Fig 2. Intussusception

Case 2: Patient B

Patient B presented to our unit in March 2021, as a referral from a rural hospital with a 3-year history of abdominal pain, associated with vomiting, abdominal distension, and constipation. He presented with a history of generalized body weakness and PICA that had been treated by pediatricians previously.

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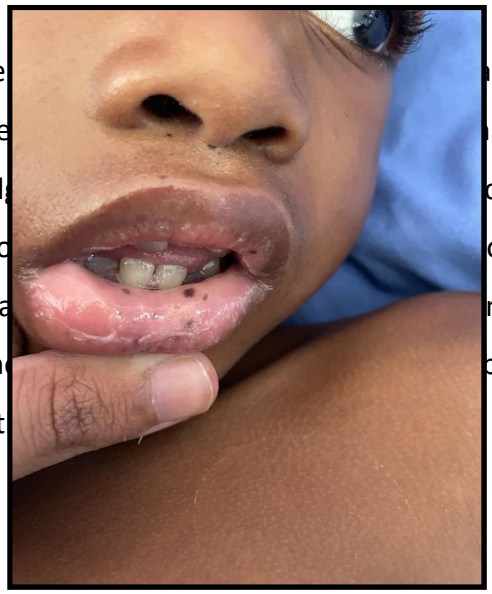

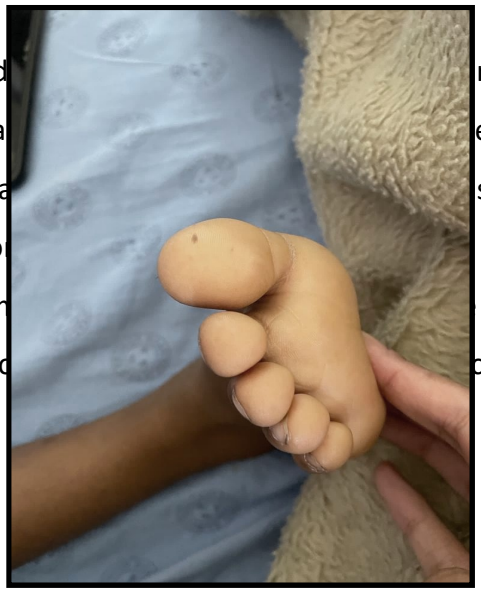

Fig 3. Mucocutaneous lesions Patient B

Abdominal X-Ray showed features in keeping with bowel obstruction, and a computed tomography (CT) of the abdomen was ordered, with a target sign suggestive of intussusception. (Fig 4)

The patient was booked for laparotomy via a Rocker-Davis incision. A large polyp (10x10 cm) was discovered in the terminal ileum, which was thought to have been the cause of the

intermittent obstruction, resulting in intermittent intussusception episodes. (Fig 4) A resection of that segment of the bowel with a primary anastomosis and a lymph node biopsy was done. (Fig 5) Three days later, the patient was discharged home, subsequently followed up at our surgical outpatient department, and scheduled for further screening.

Fig 4. CT scan

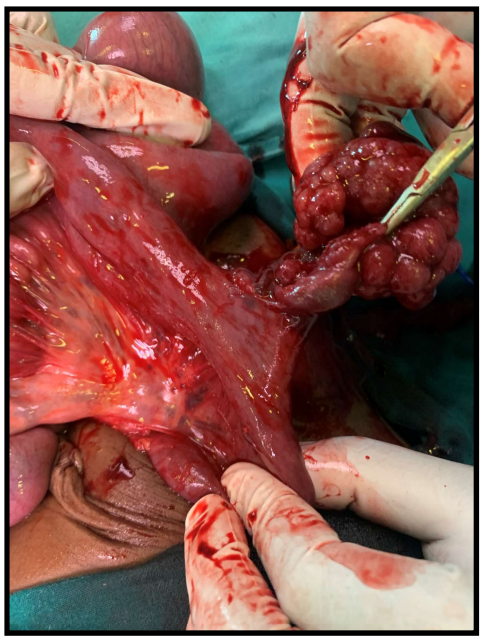

Fig 4. Polyp in terminal ileum

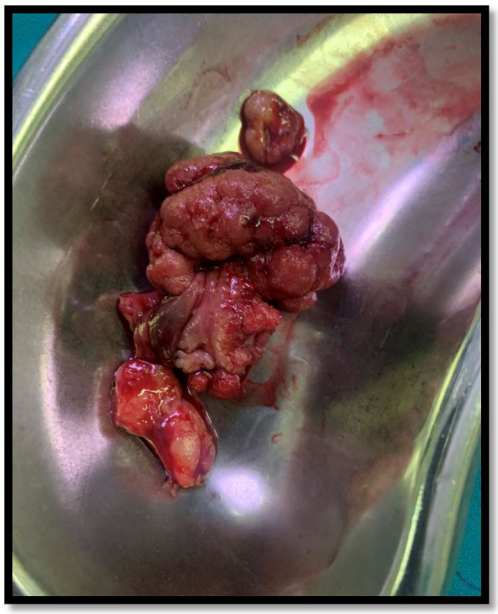

Fig 5. Excised polyp

DISCUSSION

PJS in children is rare and clinical presentation may differ, with intestinal obstruction accounting for most of the cases (43%), followed by abdominal pain (23%), blood in the stools (14%), and protruding anal polyps (7%). [7] Huang et al. noted the age of onset of symptoms to be 2 years and 4 months, and Tse et al. reported that a third of patients are

symptomatic by the age of 10, and half of the patients develop intussusception by the age of 20. [8] The most common location of the development of hamartomata's polyps is the small bowel (64%), followed by the colon (53%), stomach (49%), and rectum (32%). [7]

Most patients with PJS present with recurrent intussusception and require close observation to avoid multiple laparotomies. [9] Van Lier et al. showed that the cumulative risk of intussusception in PJS is 15% by the age of 10 years, and the size of the polyps is important [10], thus determining the presentation of both acute and chronic cases. Polyps that are >15mm are likely to cause intussusception. Most polyps telescope and reduce by themselves without any sequelae, but in certain cases, this does not happen, and patients present with classic symptoms and signs of intestinal obstruction caused by intussusception, such as colic, abdominal pain, red-current bloody diarrhea, abdominal mass, and in some cases a Dance sign. [11] Whereas patients with smaller polyps may develop intermittent bowel obstruction, symptoms might be resolved. This is why surveillance of these patients is important, with endoscopy and colonoscopy starting at 8 years of age, with repeats dependent on the presence of polyps and their sizes. [12]

CONCLUSION

PJS may present differently in patients, which can be attributed to the size and location of the polyps. A high clinical suspicion is important with early referral of these patients for long-term surveillance by a pediatric surgeon.

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