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Original Article

Surgical Management of Peutz-Jeghers Syndrome-Related Complications in Children

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Running Title: Complicated Peutz-Jeghers: Surgical Management

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Abstract:

Objective: Peutz–Jeghers syndrome is a rare hamartomatous polyposis disorder characterised by recurrent gastrointestinal polyps, mucocutaneous pigmentation, and complications including intussusception, intestinal obstruction, and gastrointestinal bleeding in children. Surgical management aims for effective polyp clearance while preserving bowel length. We report our institutional experience in managing paediatric PJS patients with complications, emphasising intraoperative enteroscopy.

Material and Methods: A retrospective analysis was performed on seven patients diagnosed with Peutz–Jeghers syndrome (PJS) between 2014 and 2025. The study examined demographics, clinical presentations, and surgical interventions including intraoperative enteroscopy, genetic testing for STK11 mutation, and outcomes.

Results: Seven children (five girls and two boys; median age, 11 years) with PJS were analysed retrospectively. Abdominal pain and vomiting were the most common presentations. Genetic testing for STK11 mutation was performed in three patients, all of whom tested positive. Intraoperative enteroscopy was performed in five patients to facilitate polyp clearance. Histopathology confirmed hamartomatous polyps in all cases, with no evidence of malignancy. Surgical management was tailored according to the site and presentation. Most patients had favourable postoperative outcomes, although one required reoperation for adhesive intestinal obstruction, while recurrent polyps were identified in two patients during follow-up. No patient developed recurrent intussusception, gastrointestinal bleeding, or malignancy.

Conclusion: Surgical management of PJS should be individualised, with intraoperative enteroscopy playing a key role in minimising bowel resections through effective polyp clearance. Open excision may be considered for selected patients with large or complex duodenal polyps. Given the risk of recurrence and malignancy, structured long-term surveillance is essential for optimal outcomes.

Keywords: enteroscopy, hamartoma, Peutz-Jeghers syndrome, polyps, resection, tumors

Introduction

Peutz–Jeghers syndrome (PJS) is a rare autosomal dominant hereditary polyposis disorder[1], characterised by hamartomatous polyps throughout the gastrointestinal tract, most commonly involving the small intestine, along with distinctive mucocutaneous pigmentation,

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which often presents in childhood and may fade after puberty, serving as an important early clinical clue[2]. Children commonly present with complications related to polyp burden[3], including intussusception, intestinal obstruction, gastric outlet obstruction, or gastrointestinal bleeding. PJS is also associated with a markedly increased lifetime risk of both gastrointestinal and extra-intestinal malignancies, with cancers reported at a relatively young age, necessitating vigilant long-term surveillance[4]. Although intraoperative enteroscopy (IOE) has been described previously, it continues to play an important role in contemporary paediatric surgical practice by facilitating complete polyp clearance while minimising repeated bowel resections and preserving bowel length. In this context, we present our institutional experience in the surgical management of paediatric patients with PJS presenting with complications, with particular emphasis on less commonly reported manifestations such as upper gastrointestinal obstruction secondary to large duodenal polyps.

Material and Methods

A retrospective analysis of electronic medical records spanning 12 years (2014–2025) was conducted in our paediatric surgery department following approval from the institutional review board. Cases were identified retrospectively from institutional surgical records and hospital medical registries. The study included seven children with Peutz–Jeghers syndrome who presented with varied gastrointestinal manifestations and underwent surgical management for disease-related complications. The diagnosis of Peutz–Jeghers syndrome was established based on the World Health Organization (WHO) diagnostic criteria, including characteristic mucocutaneous pigmentation, histopathological confirmation of hamartomatous polyps, family history, and/or genetic testing for STK11 mutation where available. In selected patients, intraoperative enteroscopy (IOE) was performed using a colonoscope to facilitate complete small bowel evaluation and polyp clearance. Pathology reports of the excised specimens were reviewed, and outcomes were noted. Genetic testing for Serine/Threonine Kinase 11 mutation was performed in three patients. Genetic counselling regarding the hereditary nature of the disease, need for surveillance, and screening of at-risk family members was provided as part of routine clinical care. Given the descriptive nature of this retrospective case series and the small sample size, no inferential statistical analyses were performed, and results are presented using descriptive statistics only.

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Results

Between 2014 and 2025, seven children were diagnosed with Peutz-Jeghers syndrome (PJS), comprising two boys and five girls. Their ages ranged from 3 to 15 years, with a mean age at diagnosis of 10 years and 2 months and a median age of 11 years. A positive family history of PJS was present in five patients. The most common presenting symptoms were abdominal pain and vomiting, observed in five cases. One child presented with melaena requiring multiple blood transfusions, and another reported an abdominal mass, with a history of prior surgery elsewhere for PJS (sigmoid resection with Hartmann's procedure), complicated by postoperative abdominal wound dehiscence requiring subtotal colectomy and excision of rectal stump polyp with enucleation of left ovarian mass, and presented to us with recurrence of an ovarian sex cord–stromal tumour. Characteristic mucocutaneous pigmentation was detected in six patients on the oral mucosa, in five on the lips, four on the hands, and in one on the feet. Two children presented as emergencies with gastrointestinal manifestations without a prior PJS diagnosis, while the remaining five were known cases. Intussusception was the most frequent mode of presentation, and two of our patients had presented with gastric outlet obstruction. A summary of the clinical characteristics is provided in Table 1. Genetic testing for STK11 mutation was performed in three patients, and all three tested positive. Genetic testing was not performed in the remaining four patients due to limited availability during the earlier study period, financial constraints, and loss to follow-up. The identified variants are summarized in **Table 2.** Endoscopic evaluation (upper and lower gastrointestinal) (Figure 1) was conducted preoperatively in three patients and postoperatively in two. The most common polyp sites were the jejunoileal region (six patients), followed by gastric and duodenal (four patients)(Figure 2) and colonic (three patients) locations. Intraoperative enteroscopy (IOE) was performed in five cases, achieving complete removal of all the macroscopically visible and palpable polyps identified during surgical exploration and enteroscopy. Polypectomy was performed in five children (Figure 3), with histopathological examination revealing hamartomatous polyps as the predominant subtype. No evidence of malignancy was identified in any of the polypectomy specimens. One child required resection and anastomosis for necrotic jejunojejunal intussusception, and another underwent a similar procedure for a large sessile jejunal polyp causing obstruction. Out of the seven patients, five underwent follow-up through outpatient visits at our institute, one continued follow-up with the local treatment team and was additionally contacted telephonically, while one patient was lost to follow-up. The median follow-up duration was 10 months (range: 5–84 months). Follow-up

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outcomes, surveillance findings, recurrence, and re-interventions are summarized in **Table 3.** On follow-up, one child required reoperation after one year due to adhesive intestinal obstruction, while another developed recurrent gastric and duodenal polyps associated with biliary obstruction and gallstone pancreatitis and is awaiting further surgical intervention. Four children remained asymptomatic and were scheduled for surveillance endoscopy at three years according to institutional protocol. During the available follow-up period, no patient developed recurrent intussusception, gastrointestinal bleeding, or malignancy. Recurrent polyps were identified in two patients during surveillance, including gastric and rectal polyps in one patient and gastric and duodenal polyps causing biliary obstruction and gallstone pancreatitis in another. Surveillance practices in our institution were based on available ESPGHAN recommendations and institutional protocol, including upper gastrointestinal endoscopy, colonoscopy, and small bowel surveillance where indicated.

Discussion

Peutz–Jeghers syndrome is a rare hamartomatous polyposis syndrome characterised by mucocutaneous pigmentation, gastrointestinal polyps, and an increased risk of malignancy, with an estimated incidence of 1:50,000 to 1:200,000 live births. Children commonly present with abdominal pain, vomiting, gastrointestinal bleeding, or complications related to intussusception and bowel obstruction secondary to polyp burden. In our series, abdominal pain and vomiting were the most frequent presenting symptoms. Female preponderance was observed, although the small sample size limits any interpretation regarding sex distribution[5]. In our series, 71% of patients had a family history of PJS.

Mucocutaneous pigmentation remains an important clinical clue for diagnosis and was identified in the majority of patients in our cohort, most commonly involving the lips and oral mucosa. Similar to previous reports, gastrointestinal manifestations were the predominant reason for presentation in our patients[6–9].

Asymptomatic PJS patients are surveilled with a combination of oesophago-gastro-duodenoscopy, colonoscopy, and MRI enterography or video capsule endoscopy. The ESGE (European Society of Gastrointestinal Endoscopy) guidelines recommend the removal of small bowel polyps >15-20 mm to prevent intussusception and small polyps in symptomatic patients using device-assisted enteroscopy[10]. This can be combined with a surgical approach in cases where patients present with acute complaints such as intussusception or in an elective setting when numerous polyps are identified on pre-operative screening. At our institute, IOE was safely performed in paediatric patients using a 12.8 mm colonoscope with a hot snare for

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polypectomy. In our study, the five patients who underwent IOE and clean sweep had all macroscopically visible and palpable polyps removed during surgical exploration and enteroscopy. IOE was not performed in all patients because of emergency presentation, prior interventions, or technical considerations. Resection and anastomosis were required in one patient with gangrene of the intussuscepted segment and in another due to the proximity of a large sessile polyp obstructing the lumen adjacent to the enterotomy site.

There are a few comprehensive protocols for managing this rare condition. As polyposis is known to recur, extensive resection and anastomosis have been abandoned in favour of IOE and polypectomy[11–14]. Oncel et al. reinforce the benefits of a 'clean sweep' where all small bowel polyps are removed in one sitting[15]. Although IOE has been described for several decades, it remains a valuable adjunct in paediatric patients with extensive or recurrent polyposis, particularly in reducing repeated bowel resections and preserving bowel length. Laparoscopy-assisted polypectomy has been described in the literature as well; however, we feel a limited laparotomy with IOE in an emergency scenario is less morbid than multiple enterotomies, as described in the study by Goneidy et al[16]. Two of our patients presented with gastric outlet obstruction due to large duodenal polyps that could not be removed using IOE. Based on our limited experience, upfront duodenotomy and polyp excision may be considered in selected patients with large sessile polyps in the D1 and D2 regions when endoscopic or enteroscopic removal is not feasible. However, further studies with larger patient cohorts are required to better define the role of this approach. Common malignancies include duodenal and colonic tumours and extra-intestinal cancers. Hearle et al. conducted a meta-analysis in which GI tract malignancies were the most common[17]; however, a newer study by Latchford et al. has shown that pancreatic and breast cancers have overtaken these in terms of frequency[18]. Ovarian tumours such as sex cord tumours with annular tubules (SCTAT) can present with precocious puberty. They are benign but can have malignant transformation and warrant salpingo-oophorectomy. One child in our series presented with a history of having undergone anoplasty and subsequent colonic resections and polypectomy at another centre. At the time of colectomy, the child had enucleation of a left ovarian mass. The biopsy was reported as a sex cord tumour. Subsequently, the child presented to us with a recurrence of a left adnexal tumour. A computerised tomography scan revealed a 7x5x3.2 cm solid-cystic left adnexal mass. She underwent a left salpingo-oophorectomy. The specimen was a pT1a sex cord stromal tumour with annular tubules and a concomitant mucinous cystadenoma. At a multidisciplinary meeting,

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it was decided to go for cytotoxic therapy given the risk of secondary malignancies and to keep her on follow-up.

Recent guidelines based on the systematic review by Beggs et al. have reduced the age for starting endoscopic follow-up from ten to eight years of age[19]. As per the European Society of Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN) recommendations, which are followed at our institute, gastrointestinal surveillance with upper gastrointestinal endoscopy, colonoscopy, and small bowel evaluation using video capsule endoscopy or MR enterography is recommended from eight years of age, or earlier in symptomatic children. Surveillance is generally repeated every three years, although the interval may be individualised based on symptoms and polyp burden. It is crucial to review these patients periodically as part of a structured surveillance programme. There are various guidelines for the screening of extra-intestinal organs (breast, ovary, testis, pancreas and lung), such as the Japanese and European expert groups, the American College of Gastroenterology (ACG)[20], the National Comprehensive Cancer Network (NCCN)[21], and the American Association for Cancer Research (AACR)[22].

Guidelines recommend annual physical examinations from eight years and ultrasounds from 18–25 years. However, one patient in our study developed a sex cord tumour at four years, highlighting the need for strict screening and evaluation of at-risk family members with features of PJS. Our institutional protocol follows these guidelines. However, the findings of our study should be interpreted in light of the small sample size and retrospective single-centre design, which may limit the generalizability of the observations. Genetic testing was available in only a subset of patients, and detailed mutation reports could not be retrieved for all tested individuals. In addition, the relatively short follow-up duration in some patients may limit assessment of long-term polyp recurrence and malignancy risk. Routine postoperative endoscopic or radiological confirmation of complete polyp clearance was not performed in all patients; therefore, small residual lesions cannot be entirely excluded.

Conclusion

PJS in children can present with a wide spectrum of manifestations, ranging from intestinal obstruction (due to intussusception or gastric outlet obstruction) to gastrointestinal bleeding. The presence of characteristic mucocutaneous melanotic pigmentation of the lips can aid in diagnosis, and these findings should be actively sought in young patients presenting with abdominal pain or intestinal obstruction. Intra-operative enteroscopy with a ‘clean sweep’ may

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be considered in patients undergoing surgical intervention to facilitate effective polyp clearance while preserving bowel length. Based on our limited experience, duodenotomy and excision may be considered in selected patients with large duodenal polyps in the D1/D2 regions when endoscopic management is not feasible. A multidisciplinary approach is essential for optimal management and long-term surveillance. Family screening of the relatives of children diagnosed with PJS may facilitate early detection and prevention of disease-related complications.

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Conflicts Of Interest

All authors have completed the ICMJE uniform disclosure form. The authors have no conflicts of interest to declare.

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Table 4: Clinical features of patients with Peutz-Jeghers syndrome. Age, sex, presenting symptoms, and polyp location are summarized.

Patient No	Age (in years)	Sex	Presenting symptoms	Cutaneous pigmentation (Mucocutaneous melanotic macule)	Location of polyps / Intra-op findings	Procedure Performed	Family History	Malignanc
1	6	Female	Recurrent Melena	Lips, oral mucosa	Stomach, duodenum (large sessile D2 polyp causing gastric outlet obstruction), jejunum, ileum (polyp as lead point for intussusception), colon.	Laparotomy + intraoperative enteroscopy (IOE) with polypectomy + duodenotomy and excision of duodenal polyp	No	No
2	3	Female	Blood in stools abdominal mass	Lips, oral mucosa	Well-encapsulated left ovarian mass Prior surgery elsewhere for PJS (sigmoid resection with Hartmann's procedure), complicated by burst abdomen requiring subtotal colectomy and excision of rectal stump polyp.	Laparotomy + left salpingo-oophorectomy	Yes	No
3	10	Female	Abdominal pain, Vomiting	Lips , hands	Ileal polyp acting as lead point for intussusception.	Laparotomy + IOE with polypectomy	No	No
4	11	Female	Abdominal pain, Vomiting	Oral mucosa	Stomach, duodenum, jejunum, ileum (polyps with intussusception) .	Laparotomy + IOE and polypectomy, along with segmental resection of the jejunum containing a large obstructing polyp, followed by	Yes	No

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						primary jejunojejunal anastomosis		
5	12	Female	Abdominal pain, Vomiting	Oral mucosa, lips, feet	Stomach, duodenum (large sessile D2 polyp causing gastric outlet obstruction), jejunum, ileum (intussusception).	Laparotomy + IOE with polypectomy + duodenotomy and duodenal polyp excision	Yes	No
6	14	Male	Abdominal pain, Vomiting	Lips, oral mucosa and hands	Jejunal polyp causing intussusception with gangrene.	Laparotomy + resection of gangrenous jejunum with anastomosis + feeding jejunostomy	Yes	No
7	15	Male	Abdominal pain, Vomiting	Lips, oral mucosa and hands	Stomach, duodenum, jejunum, ileum (intussusception), colon.	Laparotomy + IOE with polypectomy	Yes	No

Table 5: STK11 genetic variants identified in patients with Peutz–Jeghers syndrome.

Patient No	STK11 testing	Variant	Mutation type
2	Positive	Heterozygous likely pathogenic frameshift deletion	STK11 c.871delG (p.E291Sfs*44)
4	Positive	Heterozygous likely pathogenic frameshift duplication	STK11 c.1008dup (p.Val337CysfsTer23)
5	Positive	Heterozygous likely pathogenic CNV deletion	STK11 exon 2–3 deletion

Table 6: Follow-up outcomes, surveillance findings, assessment of polyp clearance, and re-interventions in children with Peutz–Jeghers syndrome.

Patient	Follow-up (months)	IOE/Polyp clearance assessment	Surveillance findings	Re-intervention/Outcome
1	14	IOE performed. complete	Surveillance endoscopy	Clinically well.

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Patient	Follow-up (months)	IOE/Polyp clearance assessment	Surveillance findings	Re-intervention/Outcome
		intraoperative clearance achieved.	not yet due (planned at 3 years)	
2	6	IOE not performed	Gastric and recurrent rectal polyps detected on surveillance endoscopy.	Under local surveillance; planned gastric and rectal polypectomy.
3	84	IOE performed. complete intraoperative clearance achieved.	No residual polypoid lesion on CT.	Adhesive intestinal obstruction requiring adhesiolysis. No documented polyp recurrence, currently well.
4	5	Endoscopic gastric polypectomy performed followed by IOE and complete intraoperative clearance achieved.	Surveillance endoscopy not yet due (planned at 3 years).	Clinically well.
5	6	IOE performed; complete intraoperative clearance achieved.	Surveillance endoscopy not yet due (planned at 3 years).	Clinically well.
6	44	IOE not performed at index surgery.	Multiple gastric, duodenal, and colonic polyps detected on surveillance.	Endoscopic polypectomy performed. Recurrent duodenal polyps causing biliary obstruction and gallstone pancreatitis; planned cholecystectomy with IOE/polypectomy.
7	Lost to follow- up	IOE performed; complete intraoperative clearance achieved.	Not available.	Lost to follow-up

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Figure 1 Upper gastrointestinal endoscopy showing gastric pre-antral (A) and duodenal (B) polyps. Colonoscopy (C) showing a colonic polyp. All polyps are indicated by black arrows (Patient 6).

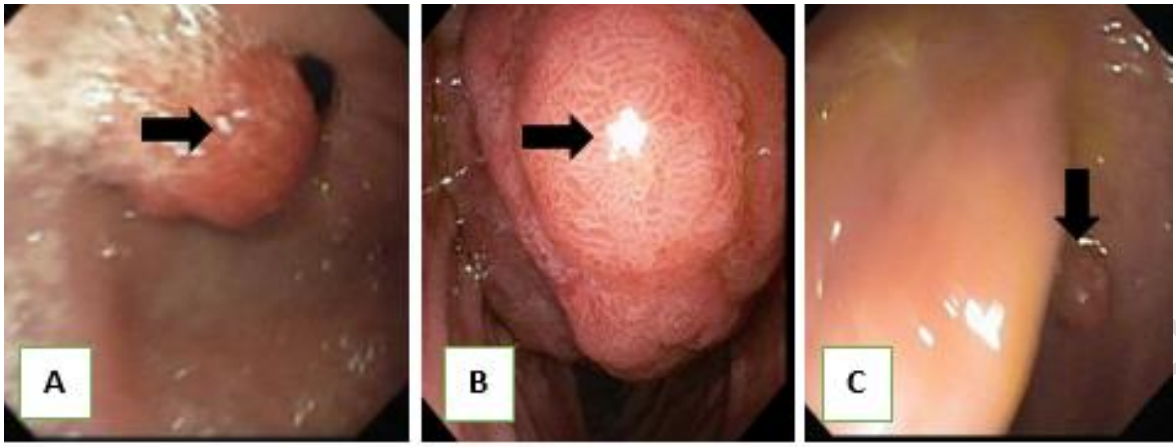
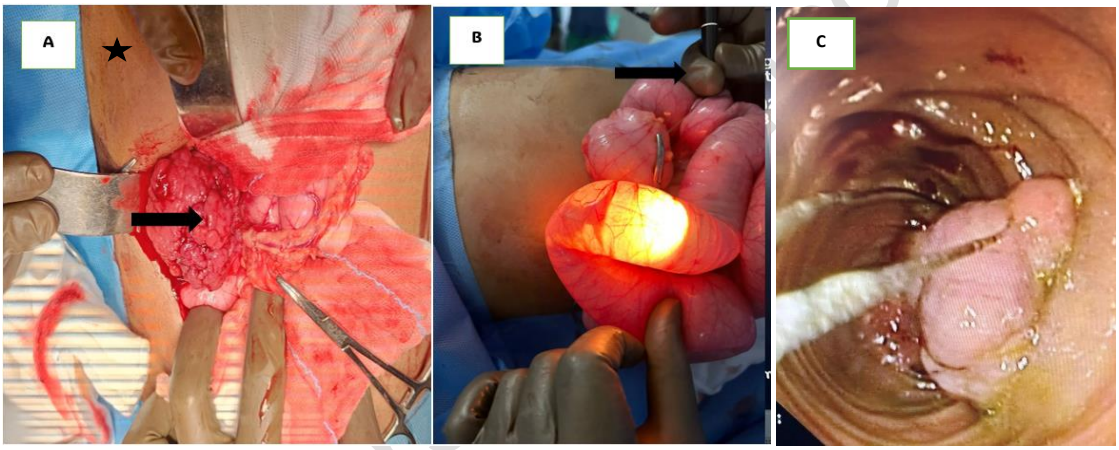


Figure 2 (A) Large duodenal polyp (arrow) causing gastric outlet obstruction. Cranial end is indicated by the star symbol. (B) Intraoperative enteroscopy showing a jejunal polyp (arrow). (C) Snare polypectomy being performed (Patient 5).



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Figure 3 Completed polypectomy of duodenal, jejunal, and ileal polyps (Patient 5).



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