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TITLE OF CASE

Ileo-ileal intussusception secondary to a Peutz-Jeghers hamartomatous polyp in an infant.

SUMMARY

We report the case of ileo-ileal intussusception secondary to a Peutz Jeghers Syndrome (PJS) hamartomatous polyp in a male infant. The patient presented with non-bilious vomiting and a single episode of passing blood in his stool. Upper gastrointestinal contrast study showed proximal bowel obstruction. At laparotomy, ileo-ileal intussusception was identified with a papillary mass acting as a lead-point. The mass was resected and a primary anastomosis was performed. The patient recovered well and was discharged on post-operative day five. Histological assessment diagnosed a PJS hamartoma. The patient was well at one month follow-up. This case report describes a rare cause of intussusception in an infant that should be considered in the differential diagnosis. Diagnosis of PJS in infancy is uncommon and requires long-term follow-up.

BACKGROUND

Intussusception describes the invagination of a segment of intestine into a section of adjacent bowel, causing luminal obstruction. The intussusceptum includes the bowel mesentery and its blood supply. If the intussusception fails to resolve the vascular supply to the prolapsed bowel can be compromised, leading to intestinal ischaemia, infarction, and perforation. Intussusception is the most common cause of small bowel obstruction in infants and children, with a peak incidence at 4 – 10 months of age [1]. Children present with abdominal pain, vomiting and a palpable ‘sausage-like’ abdominal mass. Blood per rectum, or ‘redcurrant jelly’ stool, is a late feature and signifies that bowel ischaemia has occurred.

In contrast to adults, intussusception in children is rarely caused by a pathological lead point. It is thought that intussusception occurs secondary to hypertrophic intestinal lymphoid tissue in the context of a gastrointestinal or systemic infection [2]. The absence of a pathological lead point means that children typically respond well to pneumatic or hydrostatic reduction [3]. Rarely, intussusception in infants is caused by a Meckel’s diverticulum, duplication cyst or an intestinal mass [4, 5]. Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterised by the presence of multiple gastrointestinal polyps and mucocutaneous pigmentation [6]. Here we report the case of a male infant presenting with intussusception secondary to a PJS hamartomatous polyp.

CASE PRESENTATION

A male infant presented to his local hospital with four episodes of non-bilious vomiting. The patient was born at term via caesarean section with no antenatal concerns. He was formula fed, growing well and was prescribed no routine medications. That morning, he had undergone his routine eight-week immunisations: diphtheria, tetanus, pertussis, polio, *Haemophilus influenzae* type b and hepatitis B, oral rotavirus and meningococcal group B. The local hospital team attempted to reintroduce feeds, but the patient continued to vomit. An abdominal plain film radiograph was performed, which showed dilated loops of bowel in the central abdomen (Figure 1). He was discussed with the paediatric surgical team and was transferred urgently to our tertiary paediatric surgery centre for further management of suspected bowel obstruction.

On arrival to our surgical centre, the infant was alert but irritable, and showed features of dehydration. On examination, his abdomen was soft and non-distended, with no palpable abdominal masses. He had normal external male genitalia and a normally sited anus. Altered blood, mixed in with the stool, was found in the nappy. His mother also had evidence of a bilious vomit on his clothes that had occurred on transfer. In the emergency department his observations were within age-defined normal limits. A nasogastric tube was passed. He was given a 20ml/kg fluid bolus and intravenous antibiotics were started.

INVESTIGATIONS

Blood tests on admission showed: haemoglobin of 97g/L, white cell count of $11.9x10^9$/L, platelet count of $523x10^9$/L, C-reactive protein of 58mg/L, serum sodium 135mmol/L, serum potassium of 5.2mmol/L and serum creatinine of 18µmol/L. A plain film abdominal radiograph from the local hospital showed dilated loops of bowel in the central abdomen (Figure 1). An upper gastrointestinal contrast study was performed to exclude malrotation with midgut volvulus. This showed passage of contrast through a normally positioned duodenum and the duodenojejunal (DJ) flexure to the left of the midline (Figure 2A). However, the contrast filled dilated and aperistaltic loops of proximal small bowel, suggesting a proximal obstruction (Figure 2B).

DIFFERENTIAL DIAGNOSIS

Pre-operatively, the differential diagnosis consisted of malrotation with midgut volvulus or proximal bowel obstruction, likely caused by intussusception. These diagnoses were considered based on the age of the patient and the results of the radiological investigations.

TREATMENT

The patient was taken to theatre for an emergency exploratory laparotomy. There was dilated proximal small bowel with a transition point at the site of ileo-ileal intussusception. A 10cm section of intussuscepted bowel was gently reduced, revealing a hard intraluminal mass in the ileum, approximately 30cm distal to the DJ flexure (Figure 3A). The intussusceptum was viable, so the mass was resected with a 3cm margin of distal and proximal small bowel, and a primary ileo-ileal anastomosis was performed. After resection of the lesion, the specimen was inspected, revealing a papillomatous intra-luminal mass (Figure 3B). The small and large bowel were examined to exclude synchronous lesions.

OUTCOME AND FOLLOW-UP

Post-operatively the patient was admitted to the high-dependency unit with a central venous line, trans-abdominal plane block and morphine nurse-controlled analgesia. Intravenous cefuroxime and metronidazole were given for a total of 72-hours. Clear fluids were started on post-operative day two and full enteral feeds were resumed on post-operative day four. The patient was discharged home on day five.

Histological assessment of the resected lesion diagnosed a PJS hamartomatous polyp. Macroscopic assessment showed a well circumscribed lesion with a papillary element, measuring 31mm by 15mm by 18mm. On microscopy, the lesion showed features typical of a PJS small bowel polyp. These included a papillary villous architecture with arborising smooth muscle bundles, dilated glands within the epithelium and expansion of the lamina propria with acute and chronic inflammatory cells (Figure 4).

The patient was reviewed in outpatients four weeks after discharge and was doing well. The laparotomy wound had healed without issue. The patient was feeding and stooling normally. He has been referred to a national polyposis clinic for further management and long-term follow-up.

DISCUSSION

PJS is an autosomal dominant polyposis syndrome caused by mutations in the *STK11* gene. Multiple polyps form in the gastrointestinal tract and associate with mucocutaneous pigmentation. Incidence is estimated to be 1 in 200,000 live births. Familial inheritance is observed in up to 70% of cases, with incomplete penetrance. De novo mutations are responsible for 30% of cases [7].

Hamartomatous PJS polyps show characteristic histological features, including a papillary epithelium with dilated cystic glands and arborisation of smooth muscle. The polyps are found throughout the gastrointestinal tract, most commonly in the jejunum, but can also be found in the bronchi, bladder and gallbladder [6]. Pigmented lesions are present in 95% of patients and typically develop during infancy, occurring on the mouth, nostrils, perianal area, hands and feet. These may regress during puberty but typically persist in the buccal mucosa [6].

Diagnosis of PJS relies on the presence of the following: 1) ≥2 histologically confirmed PJS polyps; 2) detection of any number of polyps in an individual with a first degree relative with PJS; 3) characteristic pigmentation in an individual with a first degree relative with PJS; or 4) detection any number of polyps in an individual with characteristic PJS pigmentation [8].

PJS associates with an increased risk of malignancy, which increases with age. Patients with PJS have a 50% risk of developing a malignancy by 60 years of age [9]. In particular, PJS patients are at risk of gastric, colorectal, pancreatic and gynaecological cancers [7]. However, identifying dysplasia or atypia in resected PJS polyps is very rare (incidence of 0.24%), suggesting that a hamartoma-adenoma-carcinoma sequence is not responsible for the development of malignant gastrointestinal tumours [10].

The natural history of PJS in children is not well described. Approximately 33% of patients develop clinical manifestations of PJS in the first decade of life and 50% in the second decade [11]. In children, PJS most commonly presents with small bowel intussusception, requiring emergency surgery and bowel resection [8]. In one retrospective study, 68% of children diagnosed with PJS had undergone a laparotomy for intestinal obstruction by 18-years of age. Median age at laparotomy was 10 years [12]. Predictive genetic testing and

gastrointestinal surveillance are recommended to identify children at risk of developing complications from PJS [8]. Rarely, cases of adenocarcinoma have been identified in children with PJS [13, 14].

Presentation of PJS in infants is very rare and complications from polyposis may occur before the development of pigmentation. Infants may present with gastric outlet obstruction, prolapsing rectal polyps or intussusception. Management is invariably surgical. Burgmeier *et al*, report a case of a neonate admitted to a tertiary hospital with recurrent episodes of non-bilious vomiting and an abdominal mass. Ultrasound and an upper gastrointestinal contrast study identified two large PJS polyps in the stomach causing obstruction. The neonate underwent a laparotomy and excision of both lesions. In this case, a family history of PJS was present from both parents [15]. Huang and colleagues reported a nine-month-old female infant who presented with recurrent prolapsing rectal polyps [16]. There was no family history of PJS. Five polyps were resected during screening colonoscopy. Post-procedure the child became unwell and large bowel perforation was diagnosed. The patient underwent a laparotomy and the perforated segment of descending colon was resected. Histological examination identified PJS polyps and genetic testing identified a mutation in *STK11*. Howel *et al.* reported a case of colo-colic intussusception secondary to a sigmoid PJS polyp in a four-month-old boy [17]. This was successfully managed through hydrostatic reduction but recurred after 16 hours. At laparotomy a sigmoid colectomy was performed with a primary anastomosis.

Intussusception is the commonest cause of small bowel obstruction in infants. Rarely it is caused by a hamartomatous PJS polyp. In infants, PJS may present with intussusception, gastric outlet obstruction or rectal polyp prolapse, and in children with PJS the risk of emergency surgery secondary to intussusception is high. PJS associates with a lifetime increased risk of malignancy, and close follow-up is recommended to prevent future complications.

LEARNING POINTS/TAKE HOME MESSAGES

- Peutz-Jeghers Syndrome (PJS) hamartomatous polyps are a rare cause of intussusception in infants. In this age group, PJS can also present as gastric outlet obstruction or with rectal polyp prolapse. The characteristic PJS pigmentation may not have developed at this age.
- The risk of surgery in children with PJS is high. Surgical resection with primary anastomosis is safe. The gastrointestinal tract should be thoroughly assessed for synchronous polyps.
- Due to the risk of complications caused by gastrointestinal polyps, and the association of PJS with malignancy, children with PJS require lifelong follow-up.

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FIGURES

Figure 1: Abdominal plain film radiograph.

Figure 2: Upper gastrointestinal contrast study. A) opacification of stomach and D1 – 4. B) dilated and obstructed proximal small bowel.

Figure 3: Intra-operative findings. A) Intra-luminal mass acting as a pathological lead point for intussusception. B) Cross-section of resected small bowel showing the hamartomatous polyp.

Figure 4: Patient histology images from the PJS polyp showing a papillary architecture with arborising smooth muscle fibres.

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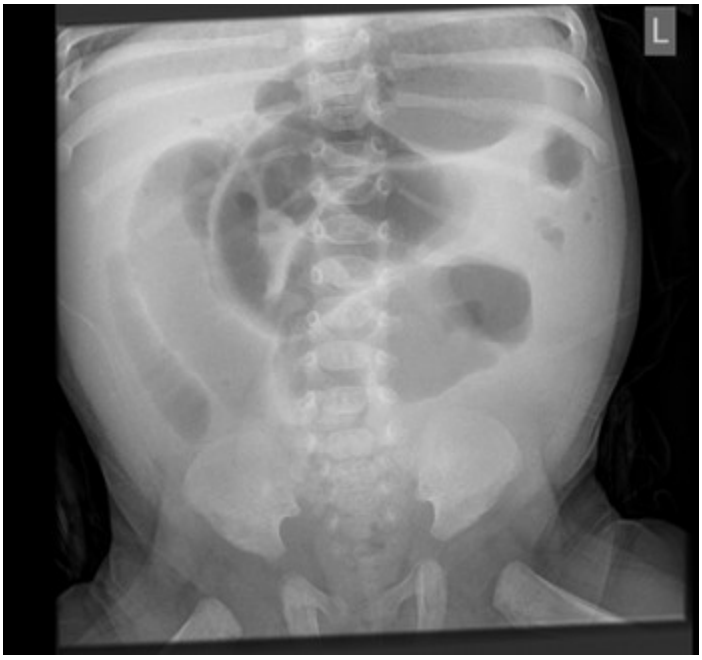

Figure 1: Abdominal plain film radiograph.

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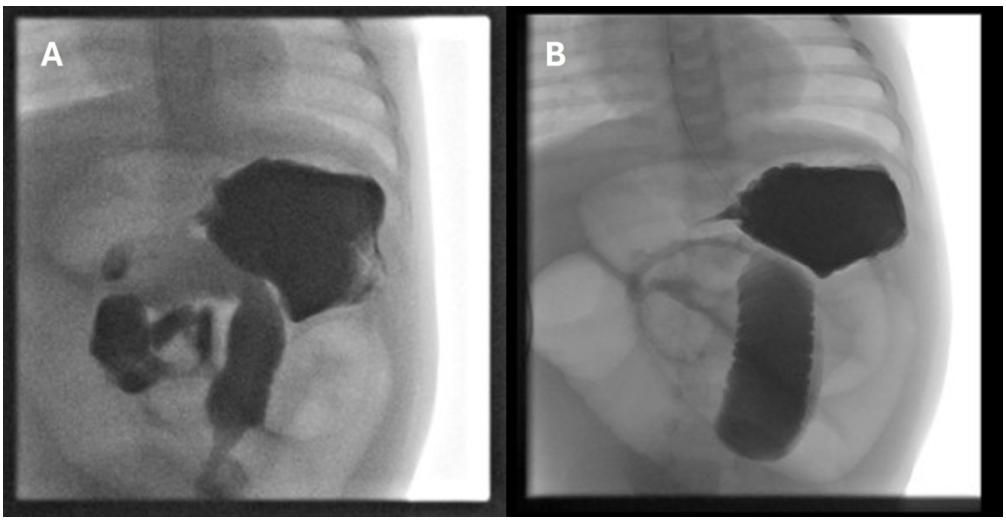

Figure 2: Upper gastrointestinal contrast study. A) opacification of stomach and D1 – 4. B) dilated and obstructed proximal small bowel.

176x89mm (300 x 300 DPI)

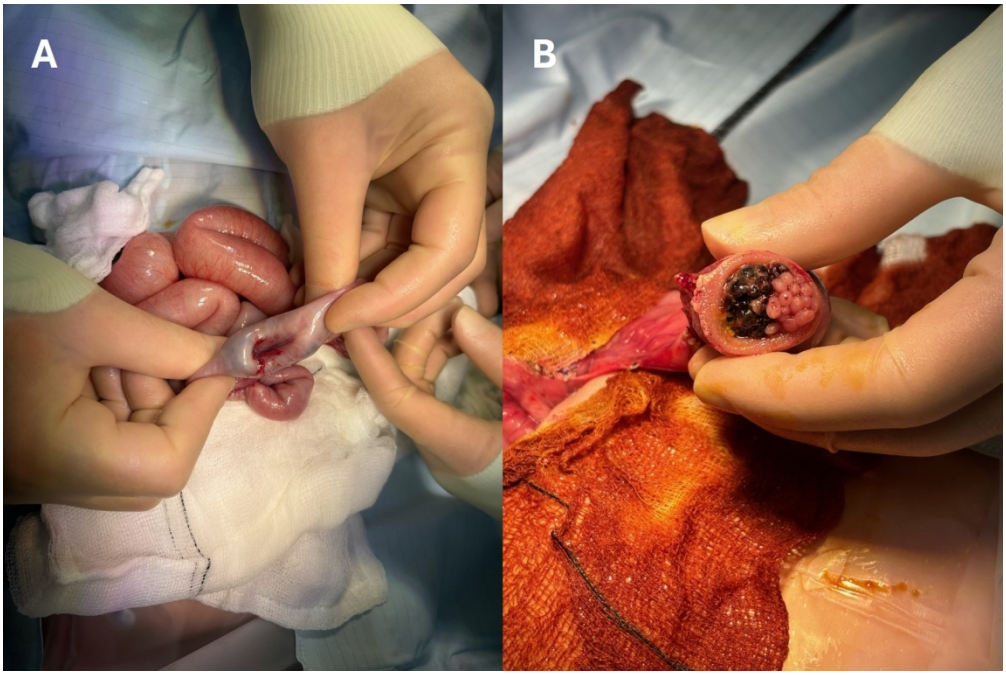

Figure 3: Intra-operative findings. A) Intra-luminal mass acting as a pathological lead point for intussusception. B) Cross-section of resected small bowel showing the hamartomatous polyp.

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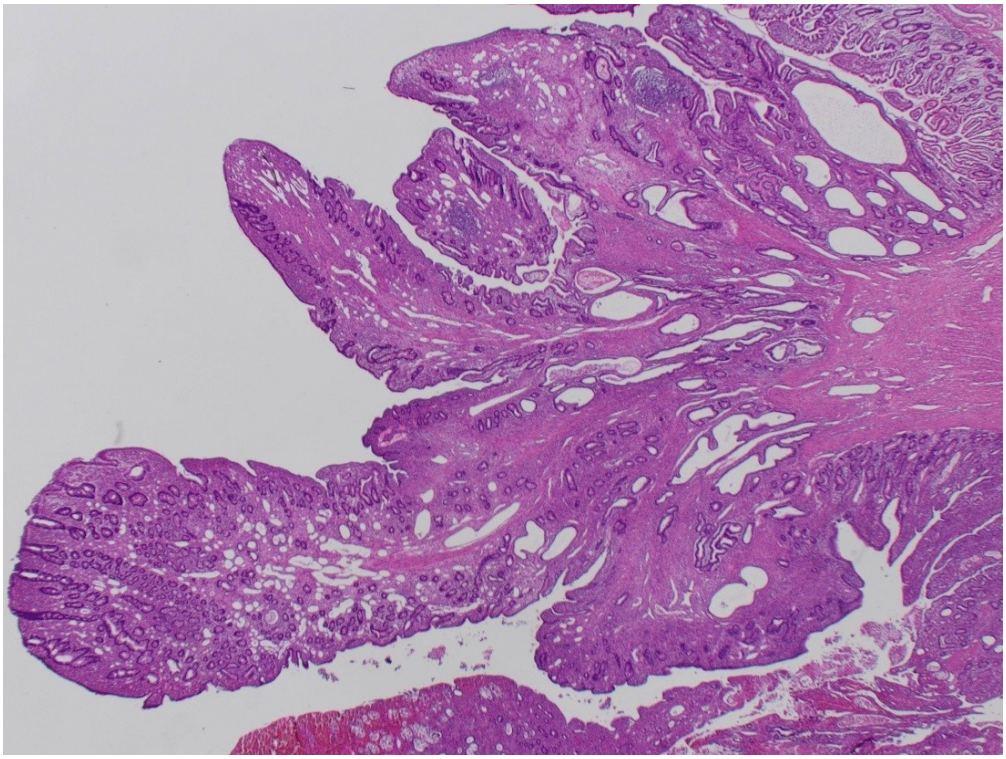

Figure 4: Patient histology images from the PJS polyp showing a papillary architecture with arborising smooth muscle fibres.

126x95mm (220 x 220 DPI)