**CASE REPORT**

Fred Boseto · Edward Shi · John Mitchell · John Preddy
Susan Adams

**Gastroduodenal intussusception due to Peutz-Jeghers Syndrome in infancy**

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**Abstract** A case of Peutz-Jeghers Syndrome (PJS) presenting in infancy with gastric-outlet obstruction is described. PJS may become symptomatic at any age and should be suspected when there are obstructive symptoms or gastrointestinal blood loss in a baby with a positive family history. Contrast studies and endoscopy are useful in diagnosis and surveillance. Treatment requires a combination of endoscopy and laparotomy/laparoscopy. Because of the reported cancer risk, we recommend surveillance from the age of diagnosis.

**Keywords** Peutz-Jeghers syndrome · Gastric-outlet obstruction · Gastroduodenal intussusception

**Introduction**

Peutz-Jeghers syndrome (PJS) is a rare, autosomal-dominant disorder characterised by gastrointestinal (GI) hamartomatous polyps and pigmented macules on the skin and oral mucose [1, 2]. Most cases present in the 2nd or 3rd decade of life [3], but in one-third the diagnosis is made before the age of 10 years [4]. PJS presenting in infancy is rare. In most cases, colonic or small-bowel intussusception or intestinal bleeding leads to the diagnosis [4, 5]. We found only four reported cases of gastric-outlet obstruction (GOO) in PJS: the youngest patient was 15 years old [6, 7, 8]. We describe an infant with PJS who presented with GOO due to an intussuscepting gastric polyp, and discuss the management of PJS presenting in childhood.

**Case report**

A 14-month-old female presented to a regional hospital with a 1-month history of symptoms of progressive GOO: non-bilious vomiting, abdominal pain, and weight loss of 1 kg, on a background of a 4-month history of failure to thrive. There was a positive family history of PJS (mother and maternal grandmother and possibly maternal great-grandfather) and breast cancer (maternal grandmother). On examination, she was lethargic and dehydrated. There was no mucocutaneous pigmentation and no palpable abdominal mass. Electrolyte examination revealed a severe hypochloremic, hypokalaemic metabolic alkalosis (Na 133 mmol/l, K 2.8 mmol/l, Cl 68 mmol/l). During the first 24 h after admission, she had two generalized seizures. Once her condition stabilised, a contrast meal confirmed GOO (Fig. 1). The child was transferred to Sydney Children’s Hospital. On arrival, she was well-hydrated, the electrolytes were fully corrected, and there were no further seizures.

Forthy-eight hours post-admission, endoscopy showed severe ulcerative oesophagitis. A sessile, polypoid lesion partially obstructing the gastric outlet was demonstrated. It was possible to pass the endoscope into the duodenum, and no distal polyps were seen. An attempt to endoscopically snare the polyp was unsuccessful because of its large size and broad base. A laparotomy was therefore performed. The large, distal gastric polyp was found to be loosely intussuscepting into the first part of the duodenum (Fig. 2) and was manually reduced. A gastrostomy revealed a sessile polyp in the distal antrum measuring 85 × 42 × 20 mm. There were two additional smaller polyps measuring 20 and 3 mm, respectively. No further intestinal polyps were discerned by visualising and palpating the small and large bowel. All polyps were completely excised by submucosal dissection and the mucosal defect was oversewn. Nasojejunal feeding and nasogastric tubes were inserted and the gastrostomy closed.

The post-operative course was uneventful. Omeprazole was commenced and feeding via the nasojugal tube introduced on day 2. Oral intake resumed on day 4 and she was discharged on day 9. On follow-up at 1 month she had an excellent appetite, no abdominal pain or vomiting, and was gaining weight. Periortal pigmentation was noted at this visit.

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F. Boseto
Department of Surgery, Prince of Wales Hospital, High Street, Randwick 2031, Australia

E. Shi · S. Adams (✉)
Department of Paediatric Surgery, Sydney Children’s Hospital, High Street, Randwick 2031, Australia

J. Mitchell
Department of Paediatric Gastroenterology, Sydney Children’s Hospital, High Street, Randwick 2031, Australia

J. Preddy
Wagga Specialist Medical Centre, 1/325 Edward Street, Wagga Wagga 2650, Australia
Discussion

PJS may present in a number of ways. Utsunomiya et al. [6] reviewed the initial symptoms in 222 patients; 42.8% were obstructed, 23.4% had abdominal pain, 13.5% presented with rectal bleeding, 7.2% with rectal prolapse, and in 23.4% mucocutaneous pigmentation led to the diagnosis. Intussusception was present in 46.9%, mostly in the small intestine. Only 1 of the 222 patients had a gastric intussusception. Bartholomew et al. [9] reported a series of 182 patients with PJS, with a 24.2% incidence of gastric polyps. It is rare for these polyps to cause mechanical problems. To our knowledge, there have been only 4 reported cases of GOO due to PJS polyps [6–8]; the youngest patient was 15 years old [8].

It is most common for clinical complications of PJS to begin in the 2nd or 3rd decade of life [3], but one-third of patients present under the age of 10 years [4]. There are a few reports in the literature of PJS presenting in infancy. In most of these cases a colonic or small-bowel intussusception or bleeding led to the diagnosis [4, 5]. To our knowledge, ours is the only reported case of a gastro-duodenal intussusception due to PJS polyposis presenting in infancy. It is rare for mucocutaneous pigmentation to be present in these young patients [4], and this was absent in our case.

The initial radiological investigation of choice for GOO is a contrast meal. Typical X-ray findings have been described by Hobbs and Cohen [10], and include: (1) sharp cut-off of contrast in the mid-stomach; (2) converging axial striations of the stomach and duodenum running parallel to their long axis; (3) a duodenal filling defect and foreshortening of the antrum; and a (4) coiled-spring appearance in the duodenum. To this, Meyers [17] added (5) pre-pyloric collar-shaped outpouching; and (6) widening of the pyloric channel. Some of these features are seen in the barium study of our