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Case Report

Familial Peutz–Jeghers Syndrome Presenting as Jejunojejunal Intussusception in an 11-Year-Old Boy: A Case Report from Bangladesh

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Abstract:

Peutz–Jeghers syndrome (PJS) is a rare autosomal dominant hamartomatous polyposis syndrome with an estimated prevalence of 1 in 8,300 to 1 in 280,000; only isolated adult cases have been reported from Bangladesh. We describe an 11-year-old boy admitted to the surgical intensive care unit (ICU) with three days of colicky abdominal pain, bilious vomiting, absolute constipation and abdominal distension. He was in hypovolaemic shock with metabolic acidosis. Multiple brown-to-black macules on the lips, buccal mucosa and fingers identified at the bedside, and ultrasonography showed a target sign. After six hours of goal-directed resuscitation, laparotomy revealed a jejunojejunal intussusception with a 3.5-cm pedunculated polyp as the lead point. The intussusception was reduced and two polyps were removed through enterotomies without bowel resection. Histopathology confirmed Peutz–Jeghers hamartomas. His mother had identical, previously unrecognised pigmentation. Recovery was uneventful, and the family entered structured surveillance. Inspection of the lips and digits of every child with intestinal obstruction can establish this lifelong diagnosis in seconds; preoperative resuscitation, bowel-preserving surgery and family screening are the cornerstones of management.

Keywords: *Intussusception; Intestinal obstruction; Mucocutaneous pigmentation; Paediatric intensive care; Peutz–Jeghers syndrome.*

Introduction

Peutz–Jeghers syndrome (PJS) is an autosomal dominant disorder caused by germline mutation of the STK11 (LKB1) tumour-suppressor gene on chromosome 19p13.3. It is defined by mucocutaneous melanin pigmentation and gastrointestinal hamartomatous polyps.[1] Its prevalence is estimated at between 1 in 8,300 and 1 in 280,000 people, most often quoted as 1 in 50,000 to 1 in 200,000; roughly half of cases are familial and half arise de novo.[1,2]

No population-based data exist for Bangladesh. Applying these estimates to a population of about 170 million would predict roughly 850 to 3,400 affected individuals, yet a search of BanglaJOL and PubMed identified only two published Bangladeshi cases, both diagnosed in adulthood: a woman with small-bowel intussusception[3] and a 29-year-old man with recurrent melaena, reported in this Journal.[4] The syndrome is therefore almost certainly under-recognised in our setting.

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Polyps are most numerous in the jejunum, where they act as lead points for intussusception. In a large Dutch cohort, 69% of patients had at least one intussusception, at a median age of 16 years, and the cumulative risk reached 50% by the age of 20 years.[5] Lifetime cancer risk, mainly gastrointestinal, pancreatic, breast and gynaecological, is estimated at 37–93%.[6] We report a child in whom PJS was recognised at ICU admission from the appearance of his lips, leading to diagnosis of his mother. The report follows the SCARE 2023 guideline.[7]

Case Report

An 11-year-old boy was referred to a tertiary-care teaching hospital in Bangladesh with three days of colicky peri-umbilical pain, repeated bilious vomiting, absolute constipation for 24 hours and progressive abdominal distension. He had had two milder, self-limiting episodes of colic in the preceding year. Dark spots had appeared on his lips at about two years of age. There was no gastrointestinal bleeding, weight loss or previous abdominal surgery.

On admission to the surgical ICU he weighed 26 kg and was dehydrated and pale. Heart rate was 138 beats/min, blood pressure 88/54 mm Hg, respiratory rate 32 breaths/min, temperature 37.8 °C, SpO_2 96% on room air and capillary refill time 4 s. Multiple discrete brown-to-black macules, 1–5 mm in diameter, covered the vermilion border of both lips, the perioral skin, the buccal mucosa and gingivae, and the volar surfaces of several fingers (Fig. 1). The abdomen was distended and tender with guarding, and a tender, sausage-shaped mass was palpable in the right para-umbilical

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region. Rectal examination revealed an empty rectum with blood-stained mucus.



Fig. 1: Mucocutaneous pigmentation in the patient. (A) Discrete brown-to-black macules, 1–5 mm in diameter, along the vermilion border of the lips and perioral skin. (B) Extension of the macules on to the labial and buccal mucosa. (C) The same lesions after induction of anaesthesia, with the orotracheal tube in situ.

Investigations showed anaemia, neutrophil leucocytosis, hyponatraemia, hypokalaemia and a high-anion-gap metabolic acidosis with hyperlactataemia (Table I). Erect abdominal radiography showed multiple air–fluid levels. Ultrasonography demonstrated a target sign in the right para-umbilical region, with dilated proximal loops and a small volume of interloop fluid. Computed tomography was not performed, as it would not have altered management.

Table I: Physiological and laboratory parameters on ICU admission and after six hours of goal-directed resuscitation

Parameter	On admission	After 6 h	Reference range
Heart rate (beats/min)	138	108	70–110
Blood pressure (mm Hg)	88/54	104/64	≥ 90/60
Capillary refill time (s)	4	2	< 2
Urine output (mL/kg/h)	0.3	1.2	≥ 1.0
Haemoglobin (g/dL)	8.6	9.4*	11.5–15.5
Total leucocyte count ($\times 10^9$/L)	16.8	14.2	4.5–13.5
C-reactive protein (mg/L)	78	—	< 6
Serum sodium (mmol/L)	131	136	135–145
Serum potassium (mmol/L)	3.2	3.9	3.5–5.1
Serum creatinine (µmol/L)	78	54	27–62
Arterial pH	7.29	7.37	7.35–7.45
Serum bicarbonate (mmol/L)	17	22	22–26
Arterial lactate (mmol/L)	3.1	1.4	< 2.0

*Notes: *After transfusion of one unit of packed red cells. Reference ranges are those of the reporting laboratory for children aged 10–12 years.*

Abbreviations: ICU, intensive care unit.

Management followed a goal-directed protocol: nasogastric decompression (350 mL of bilious aspirate in the first hour), urinary catheterisation, two 20 mL/kg boluses of balanced crystalloid followed by potassium-supplemented maintenance fluid, intravenous ceftriaxone and metronidazole, transfusion of one unit of packed red cells, and analgesia with intravenous paracetamol and a low-dose fentanyl infusion titrated to a FLACC score below 4. Lactate clearance, urine output and capillary refill, rather than blood pressure alone, were the resuscitation endpoints. All targets were met within six hours (Table I), and he was then transferred to theatre.

At laparotomy, an 8-cm jejunojejunal intussusception was found 45 cm distal to the duodenojejunal flexure. It was reduced by gentle retrograde milking; the congested segment regained colour, peristalsis and mesenteric pulsation after warm packs, and resection was avoided (Fig. 2). The lead point was a pedunculated polyp on the antimesenteric wall. Bidigital palpation of the entire small bowel identified a second pedunculated polyp 60 cm distally and two sessile lesions smaller than 5 mm. The two pedunculated polyps were excised through longitudinal enterotomies, which were closed transversely in two layers (Fig. 3). Histopathology showed an arborising smooth-muscle core covered by normal small-intestinal epithelium, without dysplasia, typical of Peutz–Jeghers hamartomas.



Fig. 2: Intraoperative view after reduction of the jejunojejunal intussusception, showing the pedunculated hamartomatous polyp that formed the lead point (between the surgeon's fingers). The previously intussuscepted segment is congested but viable.



Fig. 3: Excised small-bowel polyps. The lead-point polyp (left) measured 3.5 × 3.0 × 2.0 cm and the mid-jejunal polyp (right) 2.0 × 1.5 × 1.0 cm; both show the lobulated surface typical of Peutz–Jeghers hamartomas.

His 34-year-old mother had identical pigmentation of the lips, perioral skin, gingivae and fingers (Fig. 4), recurrent uninvestigated colic since adolescence and one episode of unexplained anaemia. She was counselled and referred for endoscopic, breast and gynaecological surveillance. His two younger siblings had no pigmentation and were enrolled for clinical review. The patient met two of the World Health Organization diagnostic criteria for PJS: histologically confirmed polyps with characteristic pigmentation, and characteristic pigmentation with an affected first-degree relative.[1] Germline STK11 testing was unavailable.

A



B



Fig. 4: Mucocutaneous pigmentation in the patient's mother. (A) Brown macules along the vermilion border and perioral skin, identical to her son's phenotype. (B) Pigmented macules on the volar surfaces of the fingers.

He was extubated in theatre and monitored in the ICU for 48 hours. Oral fluids were started on the third and a full diet on the fifth postoperative day, and he was discharged on day 7 with haemoglobin 10.6 g/dL on oral iron. At three months, upper endoscopy showed two hamartomatous gastric polyps smaller than 5 mm, and colonoscopy a single 8-mm sigmoid polyp, which was removed by snare polypectomy. He remained asymptomatic at six months and was enrolled in the surveillance programme shown in Table II.[8,9]

Table II: Surveillance programme adopted for the patient and family, adapted from ESPGHAN and EHTG recommendations[8,9]

Modality	Start	Interval
Upper GI endoscopy and colonoscopy	Age 8 y (earlier if symptomatic)	3-yearly; sooner if polyps found
Small-bowel VCE or MR enterography	Age 8 y (earlier if symptomatic)	1–3-yearly; remove polyps > 15–20 mm
Clinical review, growth, haemoglobin; testicular examination in boys	At diagnosis	Annually
Genetic counselling; examination of first-degree relatives	At diagnosis	Once; cascade screening

Abbreviations: ESPGHAN, European Society for Paediatric Gastroenterology, Hepatology and Nutrition; EHTG, European Hereditary Tumour Group; GI, gastrointestinal; VCE, video capsule endoscopy; MR, magnetic resonance.

Discussion

Mucocutaneous pigmentation is present in about 95% of patients with PJS but is often dismissed as freckling;[1] in our patient it had gone unremarked for nine years, and in his mother for three decades. A brief inspection of the lips, mouth and fingertips established the diagnosis before imaging and should be routine in any child admitted with an acute abdomen. Intussusception beyond early childhood, or recurrent colic as in our patient, should prompt a search for a

pathological lead point, which requires surgery rather than hydrostatic reduction.[5]

Protocolised resuscitation guided by lactate, urine output and capillary refill allowed surgery under normalised perfusion; this interval is justified only when strangulation and perforation are absent. Because polyp complications recur over decades, surgery should preserve bowel length by reduction, enterotomy and clearance of as many polyps as possible at one laparotomy.[9] Complications are frequent in the first decade,[10] so surveillance is advised from 8 years of age, with enteroscopic removal of polyps larger than 15–20 mm.[8,9] Limited access to enteroscopy and STK11 testing in Bangladesh remains the main barrier to such care.

Given a cumulative cancer risk of 37–93%[6] and a 50% risk to each first-degree relative, recognising the mother's phenotype was arguably the greatest benefit of this admission; clinical examination of relatives is a valid, inexpensive screen where genetic testing is unaffordable.[2] Unlike the two previous Bangladeshi cases, diagnosed in adulthood,[3,4] our patient was diagnosed at his first surgical emergency. Limitations include the absence of molecular confirmation and small-bowel enteroscopy, and follow-up of only six months.

Conclusion

Peutz–Jeghers syndrome should be considered in every child with intussusception or unexplained intestinal obstruction, particularly beyond infancy or when episodes recur. The diagnosis is often visible on the lips. Structured preoperative resuscitation, bowel-preserving surgery, histopathological confirmation, examination of first-degree relatives and lifelong surveillance form the basis of care; in resource-limited settings, clinical vigilance and family screening deliver most of the achievable benefit at negligible cost.

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Consent: Written informed consent for treatment, clinical photography and publication was obtained from both parents, with assent from the patient, and separately from the patient's mother for her own photographs.

Ethical approval: Not required for a single de-identified case report at the reporting institution. The report conforms to the Declaration of Helsinki.

Conflict of interest: The authors declare no conflict of interest.

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