Duodeno-jejunal intussusception due to a solitary Peutz–Jeghers polyp in a 3-year-old Malaysian girl

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\textbf{A B S T R A C T}

Peutz–Jeghers syndrome (PJS) is a rare autosomal dominant disorder occurring in 1 in 150 000 people. It is characterized by familial mucocutaneous pigmentation (dark freckles on face, lips, buccal mucosa, palm and soles) and hamartomatous polyposis in the gastrointestinal tract. An isolated duodenal Peutz–Jeghers type polyp causing pediatric intussusception is extremely unusual. It is unclear if a solitary duodenal Peutz–Jeghers polyp is the first sign of PJS or a completely separate clinical entity existing by itself. Here, we describe a 3-year-old Malaysian girl with a 3-month history of upper abdominal distension. A single Peutz–Jeghers type duodenal polyp, causing duodenal–jejunal intussusception and upper gastrointestinal obstruction, was subsequently visualized by ultrasonography. Submucosal resection and anastomosis of the duodenum was performed. The pathology report described a solitary 3 cm × 4 cm duodenal polyp consistent with PJS. The girl was eventually followed up in clinic. We conclude that PJS is a differential diagnosis to consider when an acute pediatric intussusception is found. Appropriate surgical techniques are necessary to reduce morbidity, mortality and the need for secondary operations. Prophylactic surveillance such as timely gastrointestinal investigations, screening of at-risk individuals and their family members, genetic testing, and lifelong patient follow-up, are recommended.

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was performed (Fig. 2). A 3 cm by 4 cm duodenal villous-like tumor arose approximately 1 cm proximal to the ampulla of Vater (Figs. 2 and 3). However, the common bile duct was not obstructed.

A part of the duodenum was submucosally resected. Initially, a 3 ml mixture of noradrenaline and normal saline in the ratio of 1:2, was injected into the mucosal layer, to raise the villous-like duodenal tumor slightly from the submucosa layer (Fig. 4). Then the tumor was marginally resected above the submucosa layer using diathermy. On gross inspection, there was no visible tumor invasion into the submucosa. An intraoperative evaluation of the small and large bowel was done at the time of the original procedure. A ‘clean sweep’ performed by manual palpation excluded any additional polyps. There were no other palpable masses. The common bile duct remained untouched. Finally, the edge of the mucosa layer was approximated using a 4/0 absorbable suture and the laparotomy incision closed (Fig. 5). Histopathology report commented that the resected specimen, measuring 3 cm by 4 cm, was nonmalignant. It showed characteristic features of PJS, such as smooth muscle hyperplasia, with elongated and arborized pattern of polyp. Post-operatively, the patient progressed well with eventual hospital discharge and prophylactic surveillance follow-up in clinic.

2. Discussion

Interestingly, studies show a benefit to resecting polyps found by manual palpation of the small and large intestines during the first operation, reducing the need for subsequent operations due to bleeding polyps and repeated intussusceptions [2,4]. Appropriate surgical techniques are also necessary to reduce morbidity, mortality and the need for secondary operations. Our patient is physiologically unprepared for a major operation so we decided upon submucosal resection of the duodenum, rather than Whipple’s procedure (duodenopancreatectomy) or local segmental resection of duodenum. These surgical techniques are discussed later. We also kept in mind the need for a second operation if histology revealed malignancy.

It is important to diagnosis PJS early in patients, because the relative risk of developing gastrointestinal carcinoma as an adult increases 18-fold compared to the general population [2]. The commonest gastrointestinal cancers associated with PJS are the esophagus, stomach, small intestine, colon and pancreas. Reports suggest that 64% of all Peutz–Jeghers polyps occur in the small intestine, while malignant changes typically occur in less than 4% of individuals with duodenal polyps due to PJS [2,5]. Carcinogenesis in PJS follows the presumptive pathway of a hamartoma developing into an adenoma, which eventually becomes a carcinoma; although hamartoma polyps have negligible malignant potential in children [3]. By understanding growth patterns of different types of polyps, intraoperatively, one can have an idea whether an intestinal polyp looks benign or cancerous.

We consider the clinical existence of a PJS, whether there is an increased malignancy risk, and reflect upon the future multidisciplinary team approach for children with atypical presentations. Management for pediatric patients with PJS includes 1) screening at-risk individuals (first degree relatives of PJS patients) and 2) surveillance of affected individuals [2]. First degree relatives of PJS patients can be screened by annual history and physical examination, and genetic testing of individuals and their affected family members. Next is patient cancer surveillance follow-up. Surveillance recommendations include history, physical examination and routine blood tests from birth to 12 years of age annually, inclusive of upper gastrointestinal endoscopy and small bowel series (or capsule endoscopy) at the age of 8 (if positive, repeated every 2–3 years). From the age of 18 onward, colonoscopy, upper gastrointestinal endoscopy and small bowel series (or capsule endoscopy) are recommended every 2–3 years [2]. In regards to our patient, we screened her immediate family members (by history and examination) with cancer surveillance follow-up.

The gold standard to detect small bowel polyps is double contrast gastrointestinal studies preceding MRI in an anaesthetized child [6]. However, capsule endoscopy may prove more useful, avoiding X-ray exposure and the need to sedate the child in an MRI procedure. Genetic analysis is also recommended for atypical cases [1,2]. Experts suggest that the mean age for Peutz–Jeghers polyps causing intussusception is 9 years old [2]. Therefore, pediatric
patients with PJS or Peutz–Jeghers type polyps require active prophylactic surveillance. This patient has been enrolled in the PJS surveillance program and gained significant weight since.

Surgery is indicated in patients presenting with recurrent intussusceptions due to polyps. Two other surgical techniques are discussed. Local segmental resection of the polyp and Whipple’s procedure (commonly known as duodenopancreatectomy) are available options. Local segmental resection is simpler, quicker and less invasive compared to Whipple’s procedure. The former is used to remove a polyp with a diameter of 3 cm or less whereas Whipple’s procedure is carried out in a polyp larger than 4 cm in diameter [7]. This ensures proper surgical clearance of anatomical margins. There are also fewer postoperative morbidity and mortality in local excision. A malignant duodenal polyp in a 3-year-old is very unlikely. However, this case is an exception. The young child is unprepared for a major operation so local segmental resection of the polyp is preferred. However, we have adapted the endoscopic submucosal resection technique in the patient.

Recently in the 1990s, Japanese surgeons treated cancerous intestinal masses by endoscopic submucosal resection (EMR), after confirmation of no submucosal invasion using endoscopic ultrasound [8]. The possibility of future laparotomies or adhesions resulting in unclear anatomical margins for surgical resection, led us to modify the EMR technique to resect the tumor intraoperatively. The advantage of having submucosal resection minimizes the morbidity resulted by segmental resection, or a more complex procedure such as Whipple’s procedure. PJS polyps in children differ in complexity and subsequent outcomes when compared to adult. This has formed the basis for the operative choice in our patient.

In contrast to typical cases of intestinal obstruction related to PJS in pediatric patients, the 3-year-old girl did not fulfill the World Health Organization criteria of PJS such as mucocutaneous pigmentation, a positive family history of PJS or three or more PJS polyps [9]. Our case is the youngest patient reported without clinical manifestations of PJS, presenting with a histological diagnosis of a PJS type duodenal polyp. It is still unclear if this is the first sign of PJS or a separate sub-type of PJS. Such rare findings are uniquely unusual with few literature reports discussing Peutz–Jeghers type polyps in the duodenum [10–12].

3. Conclusion

Isolated Peutz–Jeghers type polyp of the duodenum is extremely rare. When a pediatric small bowel intussusception is visualized, one should include PJS as a differential diagnosis. Polyps in the duodenum can significantly misshape duodenal and ductal anatomy, leading to intestinal obstruction. In this case, a less invasive procedure was warranted based on the literature review recorded in discussion. Appropriate surgical techniques are necessary to reduce morbidity, mortality and the need for secondary operations. Because of the malignant potential of PJS polyps, we emphasize on prophylactic surveillance in atypical pediatric cases. These include screening at-risk individuals (by history, examination and genetic testing) and cancer surveillance of affected individuals (by lifelong monitoring and follow-up) [2]. The argument whether a solitary PJS-type duodenal polyp is another existing type of clinical entity related to PJS, requires further genetic analyses of more affected individuals in the general population. We aim to discover the genomic background of the disease and its unknown cancer risk in further published studies worldwide [10].
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All the authors state that there are no commercial financial incentives related with publishing current work.

References