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A rare case of adult ileo-colic intussusception: Hamartomatous polyp as a lead point with concurrent appendiceal neurofibroma in Neurofibromatosis Type 1

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ABSTRACT

Intussusception is a rare cause of adult intestinal obstruction and unlike in children, adult intussusception is commonly caused by a lead point, requiring surgical intervention in most cases. Hamartomatous polyp is a non-neoplastic growth of tissue containing mature cells, distributed in an abnormal manner. It is often associated with intestinal polyposis syndromes such as Peutz-Jeghers syndrome and Juvenile polyposis. In the current case, we report an extremely rare case of ileo-colic intussusception secondary to a lead point of an isolated ileal hamartomatous polyp in an elderly gentleman with Neurofibromatosis type-1. Patient was successfully treated with ileo-colic resection involving the intussuscepted segment of bowel. There was an incidental finding of a nodule in the appendix and the histology confirmed this as a neurofibroma. Post-operative recovery of the patient was unremarkable.

Keywords: Intussusception; Hamartomatous polyp; Neurofibromatosis; Appendiceal neurofibroma

Abbreviations

CRP: C-reactive protein CT: Computed tomography
GI: Gastrointestinal HP: Hamartomatous polyp
JLIHMP: Juvenile-like inflammatory/hyperplastic mucosal polyp
NF1: Neurofibromatosis Type 1
PJS: Peutz-Jeghers syndrome

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Background

Intussusception is a condition where there is a telescoping of one segment of bowel into the adjacent one [1]. It is a rare cause of adult intestinal obstruction accounting for only 1%-5% of cases [2]. Unlike in children where the aetiology is idiopathic, adult intussusception is commonly caused by a pathologic lead point, requiring surgical intervention [3]. Hamartomatous polyp (HP) is a non-neoplastic growth of tissue containing normal mature cells, but in an abnormal distribution [4]. Previously, several reports described cases of intussusception secondary to a HP in patients with polyposis syndrome [5]. However, ileo-colic intussusception secondary to a HP in patients with Neurofibromatosis Type 1 (NF1) is a rare occurrence. The current case describes an extremely rare but unique presentation of an ileo-colic intussusception secondary to a lead point of an ileal HP and concurrent appendiceal neurofibroma in a patient with NF1 and its surgical management.

Case

A 76-year-old gentleman presented to our emergency department with 12-hour history of worsening right sided abdominal pain. The pain started acutely and was sharp in nature. It was associated with nausea and multiple episodes of diarrhoea. He denied similar experience in the past. His medical background included NF1 with café au-lait skin lesions and neurofibromas on his torso and upper back with no known other organ involvement. He also had ischaemic heart disease requiring one coronary stent and hypertension. There was family history of NF1 in his first-degree relatives including his mother and sister. His regular medications included aspirin, metoprolol and atorvastatin.

On physical examination, he was afebrile and his blood pressure was 140/60mmHg with a pulse rate of 61 beats per minute. His abdomen was distended and there were signs of local peritonism on the right side with guarding and percussive tenderness. Left-sided abdomen was not tender. His left-sided abdomen was soft with minimal tenderness. Multiple Café-au-lait lesions and neurofibromas on his chest and abdomen were noted (Figure 2). On laboratory evaluation, he had haemoglobin of 123g/L [135-180g/L], white cell count (WCC) of 17.2 [4.0-11.0] x 10^9/L, creatinine of 63umol/L [60-110umol/L] and C-reactive protein (CRP) of 1.1 mg/L [<6.0mg/L]. Venous blood gas showed pH of 7.38 with base excess of -2.6 mmol/L [-2.0/+3.0] and lactate of 1.9 mmol/L [<2.0 mmol/L]. His computed tomography (CT) scan showed abnormal cluster of distal small bowel loops centred in the right lower quadrant, with twisting of the bowel loops and mesenteric vessels and the involved bowels had mural thickening (Figure 1).

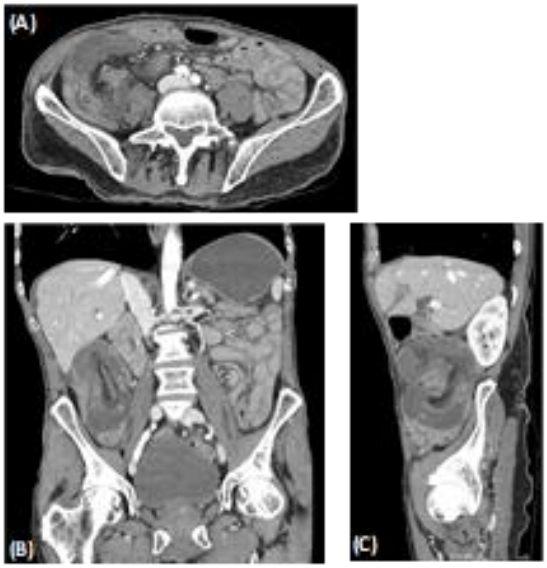

Figure 1 Abdominal computed tomography (CT) scans. A: Axial slide; B: Coronal slide; C: Sagittal slide.

Given the clinical and radiological findings, closed bowel loop obstruction due to volvulus or intussusception was suspected. Patient was taken to the operating theatre promptly. A mid-line laparotomy was performed and revealed ileo-colic intussusception with an entry point at around 10cm from the ileo-caecal junction and a lead point sitting in the mid ascending colon (Figure 2). There was moderate amount of free fluid and the proximal small bowel was distended. A small nodule in the tip of the appendix was also noted (Figure 3), but no other palpable masses were found during exploration of the rest of the bowel. Gentle reduction was initially attempted considering the extent of the intussusception without success. Limited right hemi-colectomy was performed following mobilization of the right colon. The proximal resection margin was located proximal to the ileal entry point and the distal resection margin was in the mid-ascending colon. Side-to-side staple anastomosis was performed using 75mm linear stapler. On further dissection of the resected specimen, the lead point was identified to be an ileal polyp (Figure 3). Histopathology result showed intussusception of terminal ileum with a polypoid nodule at the lead point into the caecum. The nodule showed reactive epithelial changes, goblet cell hyperplasia and focally serrated architecture, most consistent with a HP. The nodule at the tip of appendix was found to be a plexiform neurofibroma. There were no other lesions found in the specimen.

The patient was transferred to the ward and remained stable post-operatively. Diet was slowly progressed and he was discharged on the 7th day after the operation to the care of his family. He was followed up in the general surgery clinic one month after the discharge and had an unremarkable recovery. Referral for colonoscopy was made at the time of review.

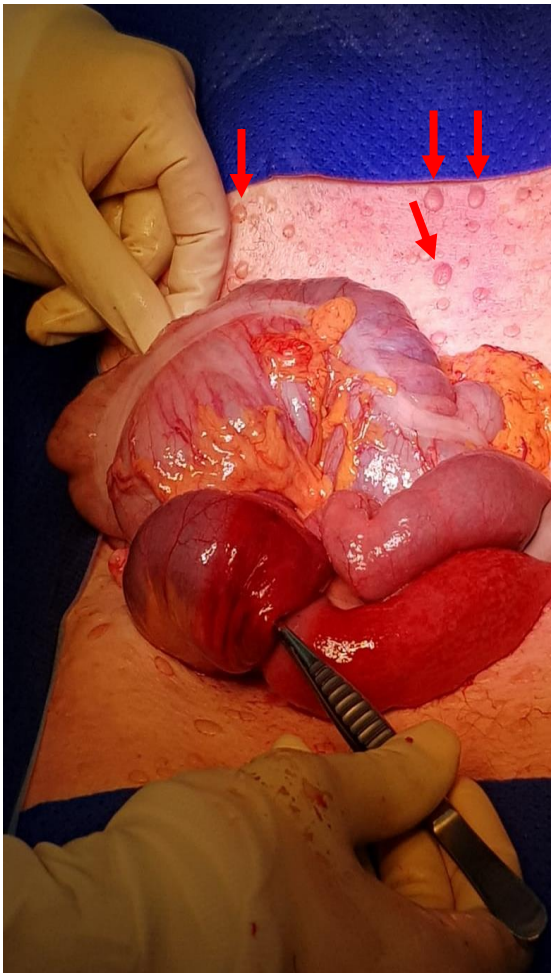

Figure 2 Intra-operative picture of ileo-colic intussusception prior to resection. The tip of the Debakey forceps is located at the entry point of intussusception. Red arrows indicate the café-au-lait lesions of the patient's abdomen.

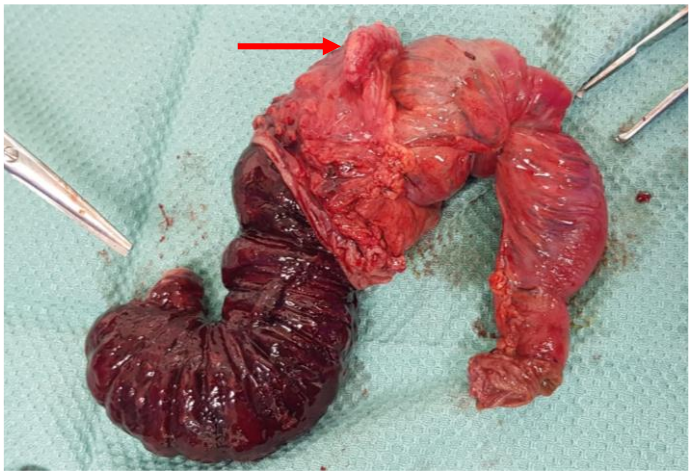

Figure 3 Intra-operative picture of ileo-colic intussusception after resection. The tip of the Metzenbaum scissors indicates the lead point of intussusception on the left. Red arrow indicates a nodule on appendiceal tip representing appendiceal neurofibroma.

Discussion

Adult intussusception differs from paediatric intussusception in many aspects including aetiology, symptoms and management [6]. In adults, intussusception is rarely idiopathic as a pathologic cause is identified in 70% to 90% of cases [7]. However, the non-specific clinical presentation and a wide variety of symptoms and signs make an early diagnosis difficult. Common symptoms and signs include gastrointestinal bleeding, altered bowel habits, nausea, vomiting and abdominal distension [8]. In our case, the patient presented with symptoms of abdominal pain and diarrhoea.

Imaging is a critical tool in diagnosing intussusception in adults due to its elusive presentation. A systemic review by Hong *et al*, 2019, showed the most accurate pre-operative diagnostic modality was CT scan, when compared with colonoscopy, barium enema and ultrasonography [9]. CT scan helps clinicians detect abdominal emergencies including intussusception and avoid unnecessary surgery [10]. The CT findings of our patient suggested intussusception, prompting the surgical team to perform emergency abdominal operation in a timely manner.

Unlike in children where the treatment is mainly non-operative, surgical resection is almost always required in adults to remove the lead point and ischaemic portion of bowel [6]. One of the causes of the lead point is a gastrointestinal (GI) polyp, which is defined as a mass or a nodule that protrudes above the level of the surrounding mucosa [4]. Hamartomatous polyp (HP) is a non-malignant growth of tissue consisting of well-differentiated, but different types of cells in an abnormal distribution [6]. Only a few cases of intussusception secondary to a solitary HP have been reported, most of which were found in the paediatric population and the cases are also scarce in the adult patients [11].

The patient has been diagnosed with NF1 from birth and had typical cutaneous manifestations. NF1 is a rare autosomal dominant hereditary condition with the most common presentations being cutaneous lesions including neurofibromas, café-au-lait lesions, axillary and inguinal freckling and Lisch nodules [12]. Gastrointestinal manifestations of NF1 are found in 5% to 25% of patients, most of which are neurofibroma and neuroendocrine tumours [13]. Gastrointestinal Juvenile-like inflammatory/hyperplastic mucosal polyps (JLIHMPs) have been proposed as a specific gastrointestinal manifestation associated with NF1 [14]. While isolated case reports previously suggested potential association between NF-1 and Juvenile polyposis [11,15], other data showed no relationship between the two [16]. The histopathology result of our patient showed a HP in ileum as a lead point of the intussusception without background history of any multiple hamartomatous polyposis syndromes such as Peutz-Jeghers syndrome (PJS) and Juvenile polyposis syndrome, highlighting the rarity of the c-

urrent presentation.

On histopathology result, there was an incidental finding of a concurrent appendiceal neurofibroma. Appendiceal neurofibroma is extremely rare where only a few cases are reported in the literature [17]. The standard treatment for patients with appendiceal neurofibroma is surgical resection due to the risk of malignant transformation [18]. Appendiceal neurofibroma was included in the right hemi-colectomy performed in our case. Although most physicians agree that surgical intervention is warranted for adult intussusception, the necessity for intra-operative reduction of intussusception and the extent of bowel resection remain controversial [19]. The potential risks associated with manipulation and reduction of intussuscepted bowel include transperitoneal seeding of tumour, perforation of bowel with seeding of micro-organisms and increased risk of anastomotic complications due to oedema of the bowel [8]. If there are signs of ischaemia or inflammation of the involved bowel, reduction should not be attempted [20]. In the current case, a gentle manipulation to reduce the intussuscepted bowel was attempted once without success. We subsequently proceeded to the limited right hemi-colectomy involving the intussuscepted ileo-caecal area and primary anastomosis between the distal ilium and mid-ascending colon. Ileo-colic intussusception secondary to an ileal HP with a concurrent finding of appendiceal neurofibroma in an adult patient with NF1 has not been described in the literature, emphasising the novelty of the current case report.

Conclusion

The current case describes an extremely rare but unique presentation of ileo-colic intussusception secondary to a harmatomatous polyp with concurrent finding of appendiceal neurofibroma in an adult patient with NF1. Due to its rarity and non-specific presentation, pre-operative diagnosis remains challenging. Abdominal CT remains as the most reliable diagnostic tool in adult intussusception. Surgical resection of the intussuscepted bowel is the recommended treatment, especially for the elderly patients as pathologic lead point is almost always present.

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