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Primary Small Bowel Neurofibroma Induced Intussusception Complicated with Small Bowel Obstruction and Perforation: A Case Report

***Abstract*—** Neurofibromatosis (NF) is a genetic neurological disorder which can lead to abnormal tumour suppression. Neurofibromatosis associated neurological tumour is usually benign in nature, only occasionally can become malignant. Gastrointestinal involvement is reported to be infrequent with only up to 25% of patients with neurofibromatosis showing involvement. Solitary gastrointestinal neurofibroma is very rare, with most cases involving the stomach or small bowel. We report here a case of solitary small intestinal neurofibroma with no other associated systemic signs, causing intussusception and intestinal obstruction.

***Keywords* - Neurofibromatosis, gastrointestinal neurofibroma, intussusception, intestinal obstruction, small intestine**

1 INTRODUCTION

Neurofibromatosis (NF) is a group of genetic disorder in which causes tumours to grow from the nervous tissue, which falls under the classification of phacomatoses. There are three genetically distinct neurofibromatoses which include neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2) and Schwannomatosis. NF1, also known as Von Recklinghausen's disease, is the commonest neurofibromatosis, occurring in about I in 3000 births [1]. NF2 occurs less frequently in approximately I in 50000 births [2]. Schwannomatosis or neurolemmomatosis is an extremely rare type of tumour syndrome characterised by the presence of multiple schwannomas in the absence of typical NF1 and NF2 syndromes [3].

Neurofibroma is the cardinal lesion of NF1 and it can be solitary, multiple, or plexiform [4]. Solitary gastrointestinal neurofibroma is rare and the clinical manifestations vary depending on the location and extent of mucosal involvement [5]. The final diagnosis of NF often relies on histopathological examination.

2 PRESENTATION

A 69-year-old man with no known medical illness, presented to the hospital with complaints of loss of appetite and generalized abdominal pain for four days. In the last two days prior to admission, the symptoms worsened, and he started to have vomiting and not passing flatus and stool for two days. On physical examination, the abdomen was distended, with generalized tenderness and involuntary guarding. Digital rectal examination revealed normal brownish stool.

Plain supine abdominal radiograph showed dilated air-filled small bowel (Figure 1). Contrast-enhanced Computed Tomography (CT) abdomen was then performed, revealing intussuscepted distal small bowel with an intraluminal mass, causing proximal small bowel dilatation (Figure 2). Presence of pneumoperitoneum, together with mesenteric inflammation and pelvic fluid collection, suggest perforated intussuscepted small bowel (image not shown).

The patient underwent surgery and small bowel resection with a double-barrel stoma. The resected small bowel was then sent for histopathological examination and the findings came back as neurofibroma. The patient continued to be followed up in the outpatient clinic and has no active complaint after the surgery.

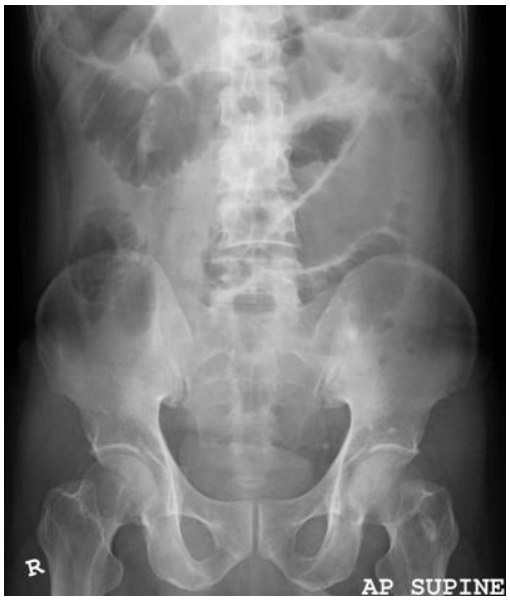

Figure 1: Plain supine abdominal radiograph showed dilated air-filled small bowel with thumb printing sign indicating small bowel thickening.

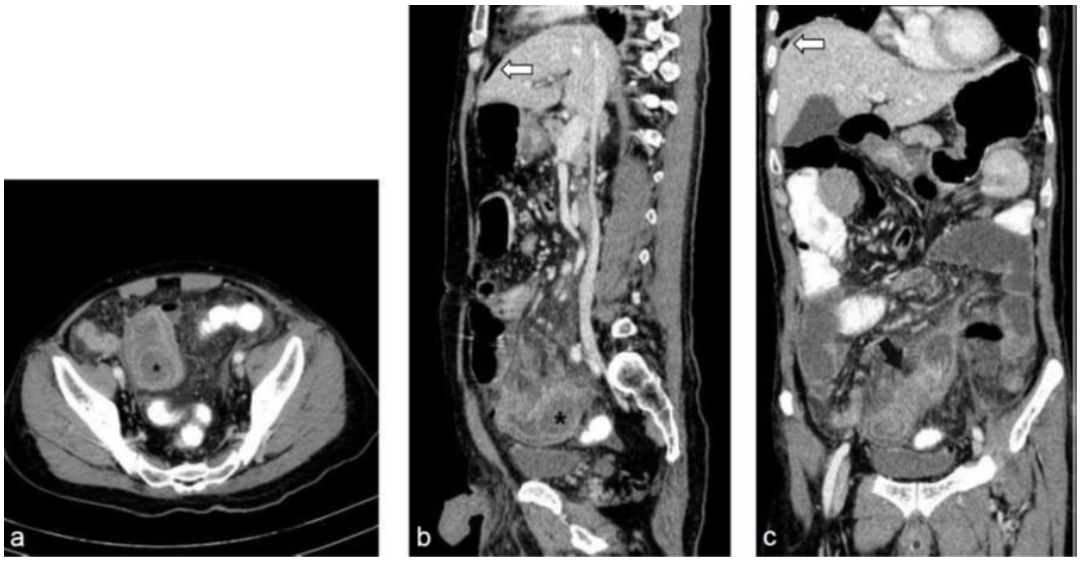

Figure 2: Axial (a), sagittal (b) and coronal (c) views of contrast-enhanced CT scan of the abdomen and pelvis. There is an intussuscepted distal small bowel (black arrow) with an intraluminal mass (asterisk), causing proximal obstruction and dilatation. Note the presence of air under diaphragm representing pneumoperitoneum (white arrow).

3 DISCUSSION

Small bowel tumours in NF1 consist of neuroendocrine tumours, gastrointestinal stromal tumour and mesenchymal tumours including neurofibroma, ganglioneuroma and fibrosarcoma [6].

Neurofibroma is the cardinal lesion of NF1 [4]. They are usually multiple upon presentation and manifestations may involve the skin, nervous system, eyes, bones, gastrointestinal tract, and other body parts. Gastrointestinal tract involvement is an uncommon entity, it has been

reported in 25% of patients with NF1, with most cases involving the stomach or small bowel [7].

Gastrointestinal manifestation of NF1 usually arise during middle or later life; generally later than the appearance of the cutaneous manifestation of the disorder [8]. Clinical presentations of gastrointestinal lesion are variable and depend on the location and extent of mucosal involvement. Mucosal involvement may lead to occult or profound gastrointestinal bleed. Intestinal obstruction from intussusception secondary to gastrointestinal neurofibroma may manifest themselves with vomiting and abdominal distension [5].

Intussusception is rarely occurred in adult (5% of all intussusceptions) and if it occurs, differentiating small bowel and large bowel intussusception is very important as in 63% of small bowel intussusceptions, a benign underlying lesion can be found, wherein 58% of case of large bowel intussusceptions, a malignant aetiology has to be expected [9][10].

Due to the nonspecific clinical presentation of gastrointestinal lesion manifestations and limitations of endoscopic techniques, CT enterography has been suggested to detect the gastrointestinal lesions. With the improved contrast and special resolution of multidetector CT, CT enterography may detect even the smaller lesions [5].

However, NF1 patients may have multiple tumours that show similar features of CT enterography so that differentiation is not achievable. NF1 associated small bowel tumours are usually well-circumscribed and show mild to marked enhancement on CT enterography [6]. CT can show focal or diffuse mural thickening, which may have a low-attenuation ringlike or rope-like pattern that corresponds to the gross morphology of plexiform neurofibromas [11].

Magnetic Resonance Imaging (MRI) may be considered as an alternative to CT enterography due to its high sensitivity for detection of small bowel tumours [5]. MR imaging features of neurofibromas are often characteristic and can be helpful in confusing cases and in the evaluation of a mass in a patient with known NF1 [12]. Neurofibromas are characteristically low signal intensity on T1-weighted images and heterogeneous high signal intensity on T2-weighted images. The high T2 signal corresponds pathologically to areas of cystic degeneration or myxoid matrix, and the low T2 signal represents collagen and fibrous tissue [13].

Surgical excision is the treatment of choice for all symptomatic tumours occurring in patients with NF1. As the benign tumours occurring in these patients always carry some risk of malignant degeneration, surgical excision of such tumours is advocated [14].

4 CONCLUSION

Gastrointestinal neurofibromatosis can be seen in isolation without other features suggestive of NF. In an adult with intussusception, a thorough evaluation of the CT findings along with clinical history, examination and histopathology examination is warranted.

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CONSENT FOR PUBLICATION

Written consent was granted from the patient for publication of this case report and the accompanying images. Institutional Review Board approval is not required at the authors' institutions for the presentation of a single case report.

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