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CASE REPORT

A RARE CASE OF INTESTINAL NEUROFIBROMA PRESENTING AS INTUSSUSCEPTIONS

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ABSTRACT: Neurofibroma is a benign nerve sheath tumour in the peripheral nervous system. They are benign slow growing nerve tumours similar to schwannomas. Neurogenic tumours of the small intestine are rare. The isolated presence of neurofibromatous lesions in the gastrointestinal tract, with no associated systemic syndromes, is a rarely reported clinical entity. We report a rare case of middle aged female, with no history of neurofibromatosis or other systemic disease, presented with small bowel obstruction secondary to an ileo-ileal intussusception induced by an isolated ileal neurofibromatous mass. The patient underwent a regional resection anastomosis and uneventful post op recovery.

KEYWORDS: Neurofibroma, Intussusception, ileo-ileal.

INTRODUCTION: Neurofibroma of small intestine are usually accompanied by Von Recklinghausen's disease (NF1) and usually originate in the intramuscular plexus of Auerbach. Neurofibromatous proliferations in the gastrointestinal tract, in general, and the small intestines, in particular, have well been described in neurofibromatosis type 1. However, the isolated presence of such lesions in the intestines with no evidence of systemic disease is a rarely reported clinical entity. Herein, we report the case of an isolated ileal neurofibroma in a middle-aged female, presenting as an ileo-ileal intussusception.

CASE REPORT: A 40 year old female presented to emergency department with complains of pain abdomen, distension of abdomen, bilious vomiting and constipation since 3days.

On examination pulse rate 94 beats/min, Bp 100/70 mmHg. Per abdomen distended with diffuse tenderness, guarding and rigidity present with increased bowel sounds.

Erect X ray abdomen showed multiple air fluid levels s/o small bowel obstruction. Usg abdomen showed dilated bowel loops with ileoileal intussusception (Target sign appearance).

Patient was subjected for emergency laparotomy and ileoileal intussception was reduced and regional resection anastomosis was done. Resected bowel showed a large polyp obstructing the lumen and the specimen sent to histopathology confirmed it as neurofibroma of ileum.

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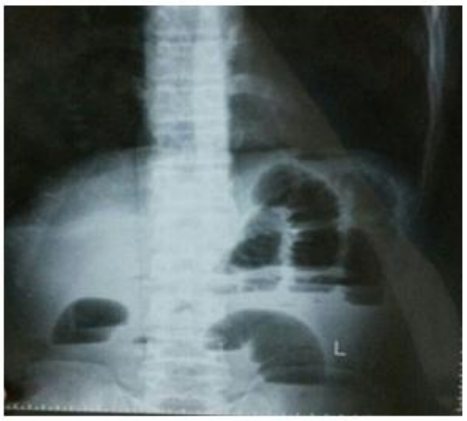

Fig. 1: X ray erect abdomen showing multiple air fluid levels

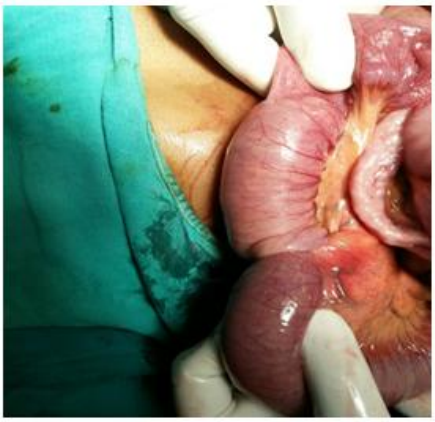

Fig. 2: Intraoperative picture of ileoileal intussusception

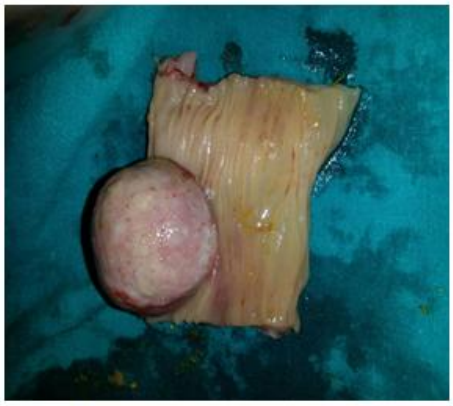

Fig. 3: Ileal polyp as lead point of intussusception

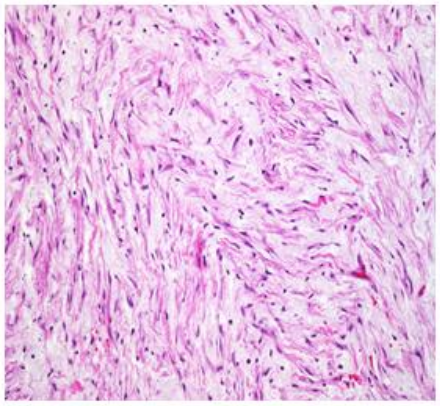

Fig. 4: Histopathological evidence of neurofibroma

DISCUSSION: Neurofibromas are benign neoplasms consisting of proliferations of all the elements in the peripheral nerves, including neurites and fibroblasts, and the predominance of elongated, serpentine Schwann cells, with their slender, spindle-shaped nuclei. Typically, these components are dispersed in a disorderly pattern, often in a loose, myxoid stroma.[1,2] Neurofibromas are usually multiple upon presentation and are usually part of two autosomal dominant disorders with variable penetrance: neurofibromatosis type 1 (NF1, von Recklinghausen's disease) and neurofibromatosis type 2 (NF2, central or bilateral acoustic neurofibromatosis).[3]

However, approximately half the cases of NF1 and NF2 arise from new mutations.[4] These disease entities have variable clinical expressions with manifestations involving the skin, nervous system, eyes, bones, gastrointestinal tract (GIT), vascular system and other body parts.[3] While NF1 is more common than NF2, NF1 is characterised by cutaneous manifestations as café-au-lait spots and axillary freckling along with a large number of nervous system tumours. On the other hand, the hallmark of NF2, and as its name suggests, is bilateral vestibular schwannomas in over 90% of patients, in addition to other nervous system tumours.[4]

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NF-1 patients may present with a diffuse neurofibromatous proliferation expanding the lamina propria mucosae and the submucosa of the gastrointestinal tract. Peripheral nerve sheath tumors of different histological subtypes may develop in the gastrointestinal organs of NF-1 patients. However, compared to the cutaneous manifestations, neurogenic tumors are relatively uncommon in the gastrointestinal tract. They may occur at any site from the esophagus to the anorectum and in the associated peritoneal and mesenteric soft tissues[5-7] Gastrointestinal involvement in neurofibromatosis is an uncommon entity.[8] While the neurofibromas do not typically affect the gastrointestinal tract in NF2,[8] these lesions are the most common abdominal neoplasms encountered in NF1, affecting the GIT in 10–25% of patients.[9]

Ganglioneuromatosis and neurofibromatosis are the pathologic forms of gastrointestinal involvement.[10] Neurofibromas of the GIT are usually originating from either the plexus of Meissner in the submucosa or the plexus of Auerbach in the muscularis propria or even from the serosa.[11,12] These lesions are often sessile and wide-based but also pedunculated polyps have been observed.[11]

Characteristic neurofibromas have been found in the GIT in 11% of patients with NF1,[13] according to some reports. Multiple neurofibromas are more often discovered in the jejunum, stomach, ileum, duodenum and colon according to the frequency of the their appearance.[3,13]

Consequently, the presence of gastrointestinal neurofibromatosis in association with NF1, and probably other syndromes, is not a rare clinical entity. However, it is rarely encountered as a separate pathologic entity[11] and reports of isolated findings of neurofibromatous proliferations in patients with no additional clinical evidence of neurocutaneous, intestinal polyposis or multiple endocrine neoplasia syndromes have been rarely documented.[14] In these settings, isolated intestinal neurofibromatous proliferations may be the initial manifestation of NF1 or MEN 2b.[15]

Intussusception is an unusual cause of bowel obstruction in adults. It is more frequent in boys under the age of two, although it can be encountered at any age. So, the primary therapeutic option of isolated neurofibromatous proliferations of the intestines is surgical,[9] depending on the location and size of the lesions. For asymptomatic, incidental findings during endoscopy, no further treatment may be required.[15] Otherwise, resection of the lesions is dictated by patient's symptoms and operability. In all cases, a correct diagnosis has considerable implications for further management as the bowel involvement could be the first manifestation of neurofibromatosis.

A similar case of an isolated ileal neurofibroma has been reported by Watanuki et al.[16] in 1995. The patient presented, however, with an ileocolic intussusception. Moreover, single or multiple neurofibromas have rarely been reported in literature. These lesions were found in the soft palate, oesophagus, stomach, gallbladder, common bile duct, small bowel and the mesentery, colon and the anal canal with no evidence of associated systemic disease.[17]

CONCLUSION: An isolated ileal neurofibroma with no associated signs of neurofibromatosis or other relevant systemic disease is a rare pathological entity. The clinical significance of such a diagnosis lies mainly in the need of further follow up of these patients as the bowel involvement could be the first manifestation of neurofibromatosis type 1 or multiple endocrine neoplasia type 2b.

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