Solitary intestinal neurofibroma with no associated systemic syndromes causing intussusception: Case report and literature review

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ABSTRACT

INTRODUCTION: The isolated presence of neurofibromatous lesions in the gastrointestinal tract, with no associated systemic syndromes, is a rarely reported clinical entity.

PRESENTATION OF CASE: A 48-year-old lady, 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 segmental enterectomy and after a smooth recovery, she was put on a long-term follow-up schedule.

DISCUSSION: This article presents a review of the literature of this area clinical entity. Very few reports of gastrointestinal isolated neurofibromas could be found. Similarly, extra-digestive isolated lesions have been rarely reported.

CONCLUSION: Isolated ileal neurofibroma 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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1. Introduction

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 lady, presenting as an ileo-ileal intussusception. A review of the medical literature on this rare entity follows.

2. Report of a case

2.1. History

A 48-year-old lady presented to our hospital with a 10-day history of intermittent, colicky, epigastric abdominal pain radiating occasionally to the whole abdomen, exacerbated by food intake. She is known to suffer from peptic ulcer disease (PUD) with a small, sliding-type, hiatal hernia. She has undergone laparoscopic cholecystectomy and appendicectomy several years before and, upon presentation, was on no medications. After normal chest X-ray, abdominal computed tomography (CT) scan and laboratory studies, an upper gastrointestinal endoscopic exploration showed severe, diffuse gastritis with antral and prepyloric erosions. A gastric biopsy showed non-atrophic glands and moderate chronic inflammation with few crypts showing Helicobacter pylori (H. pylori) organisms, suggesting type B gastritis.

So, the patient was diagnosed as suffering from severe gastritis and was started on H. pylori eradication therapy; however, she showed no symptomatic improvement. Three days later, while still at the hospital, the patient’s abdominal pain worsened and she started to develop global abdominal distension with clinical deterioration suggesting a distal gastrointestinal obstruction. An erect abdominal X-ray showed multiple air/fluid levels. Feculent material drained upon inserting a naso-gastric tube. A repeat, urgent abdominal CT scan (Fig. 1) showed severe distension of the stomach, duodenum, jejunum and ileum with fluid stasis due to intestinal intussusception at the level of the terminal ileum caused by an ileal tumour resulting in this obstructive syndrome.

Thence, the surgery team was consulted and surgical exploration was decided in view of the patient’s presentation and worsening clinical condition. During the laparotomy, around 500 cc of clear ascitic fluid was drained and largely distended jejunal and ileal loops were encountered. Upon running the small bowels, an ileo-ileal intussusception (Fig. 2) at some 60 cm from the ileocecal valve was identified and reduced and an intra-luminal mass could easily be palpated, the mesentery looked normal with no evidence of vascular compromise. No lymph nodes or other palpable intra-luminal masses could be discovered. A segmental enterectomy, including about 10 cm of the ileum from either side of the mass along with the associated mesentery was performed. Continuity of

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the bowels was re-established by a hand-sewn, side-to-side anastomosis.

2.2. Pathology

Macroscopically, the ileal segment measured 24 cm × 4 cm × 5 cm. When the bowel loop was incised, a well-delineated, intra-luminal mass arising from the bowel wall was identified measuring about 3.5 cm × 3 cm × 3 cm, this mass was spherical and pedunculated, having a smooth surface with a hard consistency; grossly, no hard or infiltrative areas related to the mass could be identified (Fig. 3). The pathology report revealed a normal intestinal wall except for a central, well-delineated, fibrous, spindle-cell neoplasm in different planes consistent with a neurofibroma.

2.3. Outcome

Following the pathological diagnosis of an isolated small bowel neurofibroma, a thorough physical examination of the patient failed to reveal any evidence of neurofibromatosis, multiple endocrine neoplasia or other syndromes. She had a smooth post-operative course. Food intake was initiated on the fourth post-operative day and the patient left the hospital on the seventh post-operative day. After discussing the condition with the patient, a follow-up schedule, consisting of 6-monthly visits, was planned with her to diagnose any emerging signs of neurofibromatosis or multiple endocrine neoplasia in the future. Three weeks post-operatively, a colonoscopy was performed that showed to be normal, ruling out any colonic polyps.

3. 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.5 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

Gastrointestinal involvement in neurofibromatosis is an uncommon entity.5 While the neurofibromas do not typically affect the gastrointestinal tract in NF2,5 these lesions are the most common abdominal neoplasms encountered in NF1, affecting the GIT in 10–25% of patients.6 Ganglioeuromatosis and neurofibromatosis are the pathologic forms of gastrointestinal involvement.7 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. These lesions are often sessile and wide-based but also pedunculated polyps have been observed. Ganglioneuromatosis, on the other hand, refers to extended hyperplasia and hypertrophy of the nerve plexuses and ganglion cells in the mucosa or throughout the bowel wall. This may lead to mural thickening and eventually stricture formation. Similar lesions could be found within the bowel mesentery, these lesions may be grossly mistaken with lymph nodes. Characteristic neurofibromas have been found in the GIT in 11% of patients with NF1, according to some reports. Multiple neurofibromas are more often documented in the jejenum, stomach, ileum, duodenum and colon according to the frequency of the their appearance. To note, further, that NF1 is also associated with the occurrence of other neoplasms that involve the GIT, including carcinoids, somatostatinomas, leiomyomas, sarcomas and pancreatic adenocarcinomas. Moreover, neurofibromas and ganglioneuromas of the GIT have also been reported in multiple endocrine neoplasia type 2b (MEN 2b), juvenile and adenomatous colonic polyposis.

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 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. In these settings, isolated intestinal neurofibromatous proliferations may be the initial manifestation of NF1 or MEN 2b.

The clinical presentation of isolated neurofibromatous lesions of the intestines are myriad and are dependent upon the focal or diffuse nature of the lesions, their location, their effect on gastrointestinal motility and their possible impingement on adjacent structures, resulting in abdominal pain, palpable masses, bleeding due to necrosis or ulceration of the mucosa, obstruction due to intussusception or extra-luminal pressure, diarrhoea, perforation, obstructive jaundice and obstruction of the pancreatic duct, among others.

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. The aetiology and pathology of invagination in children and adults are different. Infantile invaginations constitute more than 80% of infantile bowel obstruction, and 90% of the cases do not indicate any aetiologic cause or thought to be caused by enlarged nodes associated with an adenoviral infection. In contrast, adult invaginations are rare, and constitute 5% of all invaginations and less than 1% of all mechanical bowel obstruction in addition, a demonstrable aetiology is found in nearly 90% of cases in the adult population of which could be isolated intestinal neurofibromas.

Radiologically, the differential diagnosis of single or multiple nodular neurofibromatous lesions is wide and includes many epithelial and stromal neoplasms as well as nodular lymphomas. Yet, diffuse intestinal ganglioneuromatosis may mimic, radiologically, Crohn’s disease, intestinal lymphoma or carcinoid tumour with diffuse infiltration of the bowel wall.

The endoscopic appearance of these lesions depends on the focal or diffuse nature of the lesions. Most lesions can be approached endoscopically, and endoscopic biopsies are a mainstay of the diagnosis. But as neurofibromas arise deep to the epithelium, the biopsies may yield only unaffected overlying bowel mucosa or minimally diagnostic superficial lesional tissue mainly when lesions are small in size. Macroscopically, these tumours are firm and solid in consistency, and white to tan in colour. Haemorrhage and necrosis are exceptional, but superficial ulceration has been reported in both focal and diffuse entities.

In view of the uncertain aetiology and diagnosis and high incidence of malignancy (approaching 50%), the treatment of intussusception in adults is invariably surgical resection. However, the extent of bowel resection and the manipulation of the intussuscipied bowel during reduction remain controversial. In contrast to paediatric patients, where intussusception is primary and benign, preoperative reduction with barium or air is not suggested as a definite treatment for adults. The theoretical risks of preliminary manipulation and reduction of an intussuscepted bowel include (1) intraluminal seeding and venous tumour disemination, (2) perforation and seeding of microorganisms and tumour cells to the peritoneal cavity, and (3) increased risk of anastomotic complications of the manipulated friable and edematous bowel tissue. Moreover, reduction should not be attempted if there are signs of inflammation or ischaemia of the bowel wall and at age above 60 years. However, several others believe that the risks are theoretical, and gentle reduction should be attempted in selected cases to avoid unnecessary resection of healthy bowel. Endoscopic resection of colonic lipomatous polyps and laparoscopic resection of benign bowel tumours causing ileal and/or ileocolic intussusception has a role in very selected settings, care must be taken during trocars insertion and insufflation specially in cases of intestinal dilatation in order to avoid bowel injury.

So, the primary therapeutic option of isolated neurofibromatous proliferations of the intestines is surgical, depending on the location and size of the lesions. For asymptomatic, incidental findings during endoscopy, no further treatment may be required. 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. 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.

The presence of isolated neurofibromas outside the GIT has also been documented. These rare cases involved the kidney, spermatic cord, nasal cavity, palateonial tumour, paraffaryngeal space, larynx, humerus, submandibular salivary gland, conjunctiva, retroperitoneal space, cranial ventricles and chest wall.

4. 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.

Conflict of interest statement

No conflicts of interest.

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Ethical approval

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contributions

Ali Al-Harake has operated and managed the patient. Mohomad Chour has written the case and contributed to study design and data collection. Osama S. Al Beteddini has written the article and collected relevant literature.

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