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Solitary and complicated neurofibroma of small Bowel: A case report

Hassane Ait Ali *, Brahim Zeriouh, Usman Egyir Ebo, Badr Serji, Tijani Elharroudi

Department of Oncological Surgery, Mohamed VI University Hospital Center, Oujda, Morocco

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ABSTRACT

INTRODUCTION: Neurofibromatosis is a genetic disorder characterized by tumors and pigmentary changes on the skin, such as spots that color leans to 'White Coffee'.

Neurofibromas of the gastrointestinal tract are commonly associated with neurofibromatosis type I (NF1). Although, digestive involvement can be the single manifestation of the disease and may consequently; represent the only diagnostic element.

PRESENTATION OF CASE: We report here; a case of a patient admitted to the emergency department with a bowel obstruction, for which radiological investigations revealed the presence of intussusception due to an intestinal tumor.

The patient underwent a bowel resection with anastomosis, and then, after being examined histologically, the result has identified an intestinal neurofibroma without evidence of malignancy. Then and on the fourth day following the surgery, the patient was discharged with good clinical improvement.

DISCUSSION: The intestinal neurofibroma may be the first and the only manifestation of neurofibromatosis type I. Also, it's uncommon to present a neurofibroma isolated from the small bowel with an intussusception, which makes the pre-surgical diagnosis very difficult. And until now, only a few case reports of these conditions have been reported.

CONCLUSION: We report this uncommon clinical case of an isolated neurofibroma from the small bowel to raise awareness among the medical team about this exceptional pathology. Nevertheless, its risk of developing serious complications and malignant transformation led us to opt for earlier surgical treatment.

Furthermore, it requires a close clinical follow-up to eliminate the neurofibromatosis type I or the multiple endocrine neoplasia type II.

1. Introduction

Neurofibromas are peripheral nerve sheath tumors that consist of Schwan cells, perineural cells, and myofibroblasts [1–3]. They are also a characteristic lesion of Neurofibromatosis type 1. However, multiple neurofibromas have been found to involve almost all parts of the body: including the gastrointestinal tract, but the small bowel neurofibroma can be the only manifestation of the disease, and therefore, represents the element that allows us to diagnose neurofibromatosis. As is with small bowel tumors, bowel Neurofibroma is a rare entity, and the diagnosis is usually late after the onset of complications.

We describe a rare case of a solitary neurofibroma of the small bowel, complicated by an ileo-ileal intussusception resulting in an acute intestinal obstruction, in a patient without features of neurofibromatosis type 1 (NF1) and successfully managed by operative intervention in a university hospital center.

This work has been reported in line with the SCARE 2018 criteria [4].

2. Presentation of case

The patient is an 84-year old-man, married, without profession, with BMI 24 kg/m2 referred by a family physician to the emergency department for the management of an acute intestinal obstruction (absence of flatus and stool) which had begun four days prior, associated with vomiting, without fever and without any other associated signs.

The patient had no significant family history, no previous medical or surgical history, no regular medications, no allergies, and no relevant genetic information. There was neither smoking history nor other special habits of medical importance.

On physical examination, the Temperature was 37 °C, heart rate was 75 beats per minute, respiratory rate was 18 breaths per minute, and blood pressure was 129/72 mmHg.

His abdomen was undistended as well as abdominal palpation revealed diffuse tenderness. The digital rectal examination found an empty rectal vault.

* Corresponding author.
E-mail address: dr.hassaneaitali@gmail.com (H. Ait Ali).

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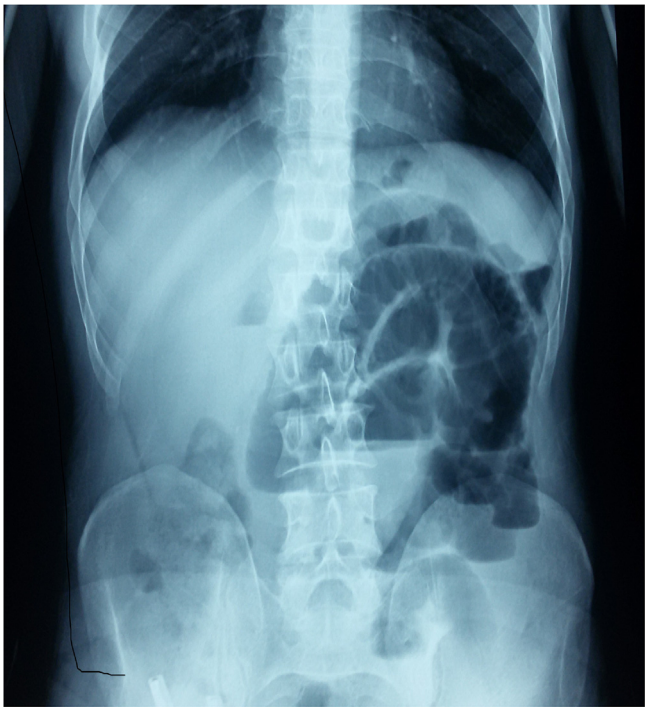

Fig. 1. The abdominal radiography (X-ray) demonstrated an intestinal obstruction with Gaz fluid levels.

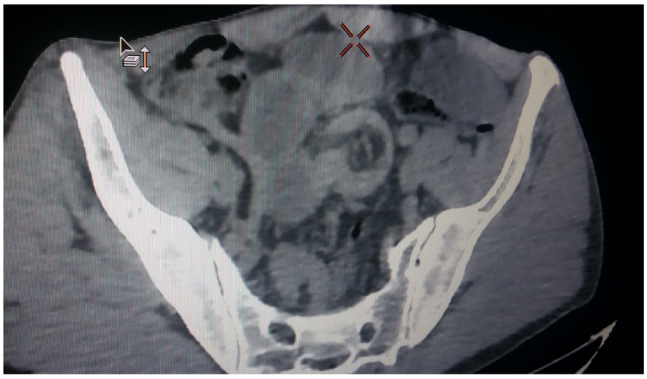

Fig. 2. The abdominal CT scan showing an intestinal obstruction with the ileal site of intussusception (Target sign).

Clinically the patient had no café-au-lait spots, axillary freckling, peripheral neurofibromas, nor pigmented hamartomas of the iris. There was no family history of neurofibromatosis. An abdominal X-ray showed air/fluid levels (Fig. 1). Routine blood investigations and tumor markers were normal.

An emergency abdominal CT scan (Fig. 2) showed distension of jejunum and ileum with fluid stasis upstream of intestinal intussusception in the small bowel caused by an ileal tumor resulting in this obstructive syndrome.

Therefore the patient underwent emergency exploratory laparotomy. Thus, surgery was performed with midline laparotomy, and the exploration found an intestinal intussusception located at some 100 cm from the ileocecal valve on a bowel tumor (Fig. 3). As well as, no lymph nodes or other palpable intra-luminal mass could be discovered. The rest of the exploration did not find any ascites, carcinoma, or liver metastasis. Thereafter, he had a segmental resection of the small bowel (Fig. 4), and the surgical specimen was sent for histological examination.

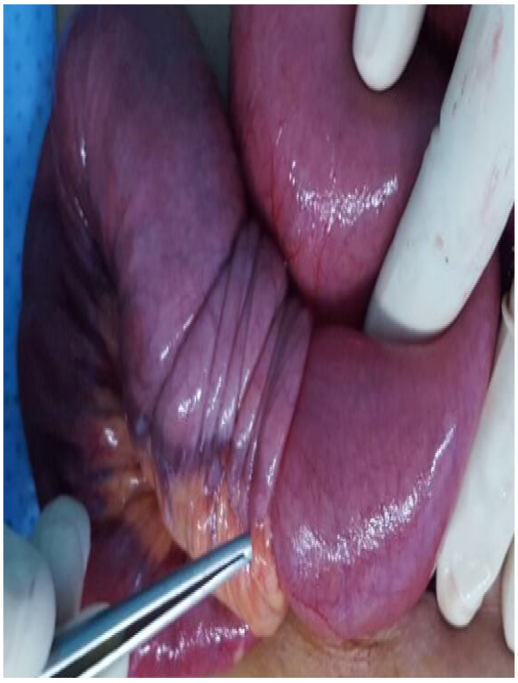

Fig. 3. Intra-operative image showing the ileo-ileal site of the intussusception at some 100 cm from the ileocecal valve.

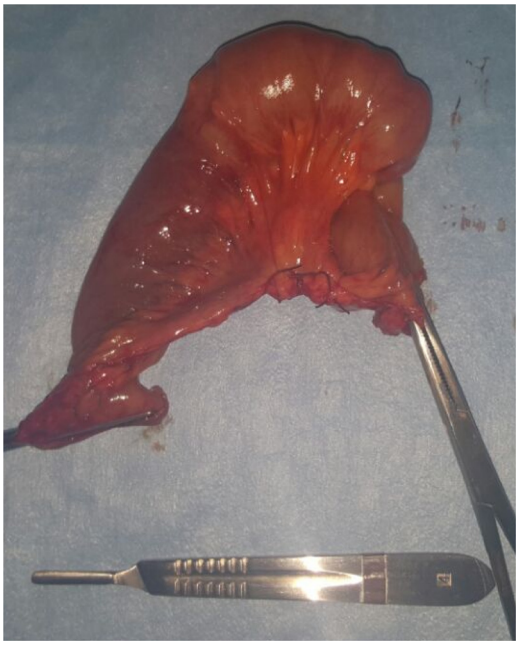

Fig. 4. Image of the surgical specimen showing the tumor responsible for the intussusception.

The bowels were re-established, at the time of bowel resection, by a hand-sewn, with ileo-ileal end to end anastomosis, and the abdomen was closed.

This operation was performed by a general surgeon with 4 years of experience at the university hospital center.

Intestinal transit was returned on the second day after surgery, and the patient was discharged on the fourth day with a follow-up in the outpatient clinic.

The Postoperative follow-up was simple without any immediate or late complications.

The histological examination of the surgical specimen was returned in favor of a neurofibroma 3.5 cm long axis, histologically benign and the excision limits are intact.

SCARE 2018 paper was used for the construction of this paper [4].

Following the unexpected result of the pathological examination, which was returned in favor of a neurofibroma of the small bowel, an exhaustive search for other neurofibromatosis locations was performed, in association with the dermatology and ophthalmology departments, which were normal.

There was no history of neurofibromatosis in any other member of the family.

The procedure and follow-up were done according to what is generally accepted in such cases.

A colonoscopy was performed 3 months after the surgery and, no lesions were found.

After a year of surveillance, the patient did not have any tumor recurrence or other neurofibromatosis signs.

3. Discussion

Neurofibromatosis is a neuroectodermal disorder [5] characterized by pigmentary changes of the skin (café-au-lait) associated with skin and visceral tumors (neurofibroma) as well as systemic abnormalities. These tumors are usually noncancerous (benign) but sometimes can become cancerous (malignant). Then, they are two distinct entities of neurofibromatosis; neurofibromatosis type 1 (von Recklinghausen disease or NF1) is caused by alterations in the NF-1 gene [6], and neurofibromatosis type 2 (NF2), which is considered as the central nervous system neurofibroma.

The clinical diagnosis of NF1 is based on clinical criteria established by the National Institutes of Health, which requires the presence of two or more of the following criteria: Six or more café au lait patches, two or more neurofibromas, one plexiform neurofibroma, freckling in the axillary or groin region, Lisch nodules, optic glioma, a first degree relative with NF1 and/or the presence of osseous dysplasia [3].

However, gastrointestinal involvement in neurofibromatosis 1 represents 10%–25% of all patients [7], and these involvements are often asymptomatic, thus may be underestimated. It is associated with NF1 but not NF2 [8]. Neurofibromas are the most common type of gastrointestinal manifestations, and they are located mostly in the small bowel. They are benign nerve sheath tumors arising from Schwann cells, and the Auerbach plexus is the origin site [9].

It may present as a solitary mass with smooth, well-defined margins or a plexiform presentation [10]. Solitary neurofibroma (SNF) means a localized neurofibroma without any other manifestation of neurofibromatosis type 1 [11]. On the other hand, localization in the small bowel, in general, is less described in the literature [12], and even rarer as an isolated neurofibroma without any other systemic signs of neurofibromatosis and maybe the initial sign of NF1 in patients without any other clinical manifestations of the disease. One possibility is described; the f̈amilial intestinal neurofibromatosisïs a rare dominant autosomique disorder, phenotypically distinct from NF1, and characterized by multiple intestinal neurofibromas restricted only to the intestine [13].

All ages can be affected, but a higher incidence is observed between 40 and 60 years of age [14]. In our case, the extremely high age of our patient suggested initially, rather a malignant pathology. Many authors point out that the ileum is most often involved [15], but all gastrointestinal tract locations can be affected.

The clinical symptomatology may depend on the macroscopic aspect of the lesion and its location. It is Asymptomatic in 65%, and sometimes the gastrointestinal tract neurofibromas are incidentally detected during surgery for associated tumors [16]. Although there is no specific sign of gastrointestinal neurofibromatosis, clinical manifestations include abdominal pain, constipation, anemia, melena, and an abdominal mass. Serious complications that have been reported include intestinal obstruction, ischemic bowel, perforation, intestinal obstruction on intussusception, and megacolon. Whereas the skin lesions are easily recognizable, the diagnosis of small bowel neurofibroma is difficult and often discovered in late stages or revealed by a complication such as a hemorrhagic or intestinal obstruction. However, the obstruction may be the result of invagination or obstruction by tumor thickening.

Our patient had no clinical symptoms prior to the intestinal obstruction.

There is no recommendation about modalities to diagnose the small bowel neurofibromas because of the nonspecific clinical presentation of these lesions and the limitation of endoscopic techniques. However, endoscopically, neurofibromas are often sessile and wide-based or pedunculated polyps [17].

On CT scan, they appear as homogeneously hypoattenuating round or tubular mass. This characteristic reflects components of myxoid stroma, Schwann cells, adipocytes, and cystic degeneration [10].

MR enterography has a high sensitivity for detecting small bowel lesions and is more preferable to computed tomography (CT) scan enterography for the follow-up.

On MRI, they present low signal intensity on T1-weighted images and variable signal on T2-weighted images [16]. However, the histological examination of a biopsy fragment or operative specimen remains the gold standard for the confirmatory diagnosis [18].

Our patient was diagnosed with a solitary small bowel neurofibroma, without any clinical signs of Neurofibromatosis 1, and the diagnosis was made after histological examination of the surgical specimen.

The presence of such lesions in the intestinal tract without associated systemic manifestations is a rare finding. However, we searched for published articles in PubMed on solitary small bowel neurofibromas, but only a few cases were reported in these conditions. Moreover, we find two similar cases of an isolated ileal neurofibroma that have been reported by Watanuki et al. in 1995 [19] and by Ali Al-Harakea et al. in 2013 [20] as well as these patients presented with acute intussusception.

In all cases, a diagnosis of bowel neurofibroma requires looking for other signs of neurofibromatosis and also looking for another location of the tumor in the bowel. Nevertheless, in rare cases, no other tumor localization or extra-digestive manifestations are found, as our observation proves.

The differential diagnosis for bowel neurofibroma includes; schwannoma, myxoma, inflammatory fibroid polyp, and GIST [2].

The natural history of these lesions is typically benign, but the malignant transformation has been reported between 5 and 15%, especially in patients over 40 years of age with NF1. The lifetime malignancy risk of these tumors in patients with Neurofibromatosis 1 is 8%–13% [3]. This transformation is often clinically silent and is often diagnosed at an advanced stage. Therefore the primary treatment of isolated neurofibromatosis is surgical resection because of the danger of malignant degeneration, obstruction, intussusception, and bleeding. But it always depends on the site, size of the tumor, and the patient's operability.

Furthermore, the intussusception treatment in children is generally conservative but often is surgical in adult's patient. However, the manipulation of the tumor during the operation before resection must be avoided, given its dissemination risk.

The prognosis is difficult to establish due to the risk of local recurrence and malignant transformation, however, a follow-up of these patients is important because the prognosis remains poor and depends on the tumor size and location, resection mar-

gins, adjuvant chemotherapy, distant metastasis, stage and site [21].

4. Conclusion

Solitary neurofibromas of the small bowel are rare. Even rarer are isolated neurofibromas without any other systemic signs of neurofibromatosis. While the natural history of these lesions is typically benign, but the malignant transformation has been reported. However, early diagnosis of these gastrointestinal manifestations is important because of the danger of malignant degeneration, obstruction, intussusception, and bleeding. Therefore, surgical resection prevents these risks, and a careful examination is required to find out unrecognized neurofibroma during operation.

Close clinical follow-up of these patients is thus important to exclude Neurofibromatosis.

In the end, this tumor should be considered in the preoperative and intra-operative differential diagnosis of bowel tumors in patients without any signs of NF I.

Declaration of Competing Interest

The authors report no declarations of interest.

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Ethical approval

No ethical approval necessary.

Consent

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's contribution

Ait Ali Hassane: Writing the manuscript, literature review, final approval of the manuscript and follow up.

Zeriouh Brahim, Egyir Ebo Usman: participants in the writing of the article.

Serji Badr, Elharroudi Tijani: Editing the review and final approval of the manuscript.

Registration of research studies

Not applicable.

Guarantor

Hassane Ait Ali.

Provenance and peer review

Not commissioned, externally peer-reviewed.

Patient perspective

The procedure of intervention was explained to the patient with all findings and possible complications. He accepted the procedure and informed consent was given.

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