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**Neurofibroma discharged from anus with stool: A case report and review of literature**

Miao Y *et al.* Neurofibroma discharged from anus with stool

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**Abstract**

Isolated neurofibromas that affect the gastrointestinal tract are rare and almost always manifest as neurofibromatosis type 1 or multiple endocrine neoplasia type 2b. In this paper, we present a case of a 24-year-old female with abdominal pain who discharged a neurofibroma in her stool without any blood on it. A colonoscopy showed multiple small polyps in the sigmoid colon and a nodule in the ileocecus. The pathology results and the immunohistochemical stains of the removed neoplasm from the ileocecus confirmed the diagnosis was a bowel neurofibroma. We report a rare case of ileocecal neurofibroma due to the patient’s affected gastrointestinal tract, without any associated systemic syndrome other than a neurofibroma discharged in the stool.

**Key words:** Neurofibroma; Isolated; Gastrointestinal tract; Ileocecus; Clinical presentation

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**Core tip:** The neurofibroma of the gastrointestinal tract are rare, various types of manifest have been reported previously, but a neurofibroma discharged from the patient’s intestine with stool without any other associated systemic syndromes is rare and may the first time to be reported.

Miao Y, Wang JJ, Chen ZM, Zhu JL, Wang MB, Cai SQ. Neurofibroma discharged from anus with stool: A case report and review of literature. *World J Clin Cases* 2018; In press

**INTRODUCTION**

Neurofibroma of the bowel tract can occur with multiple symptoms and has several different names, including intestinal neurofibromatosis, ganglioneuromatosis, diffuse plexiform neurofibromatosis, neuronal intestinal dysplasia, and diffuse colonic ganglioneuromatous polyposis. About [a quarter](http://www.baidu.com/link?url=DfsUSrnXB4O0KrajK1yYmlFblBZfbNjkFMSZf0D1cSQ5nQwgcSFoNm4ccK6ANrh4TEbWO9s_cw8rA--d4tIYxuyu934fFZEaL8Vo_dABe_K) of neurofibromatosis type-1 (NF1) and multiple endocrine-neoplasia type-2b (MEN2b) cases were reported to be associated with gastrointestinal neurofibromatosis[[1](#_ENREF_1)], while the occurrence of gastrointestinal neurofibromatosis alone was reported to be extremely rare[[2](#_ENREF_2),[3](#_ENREF_3)]. Currently, whether neurofibromatosis of the bowel tract without any systemic syndrome is a totally distinct condition or simply a phenotypic manifestation of NF1 or MEN2b remains an open question.

**CASE REPORT**

A previously healthy 24-year-old female came to our hospital complaining of a month-long history of abdominal pain after meals. She stated that she had never developed dysphagia, diarrhea, nausea, vomiting, or fever during the month of abdominal pain. Forty days prior to her symptoms, she found an approximately 8 cm × 5 cm × 5 cm lump in her stool without any blood on it (Figure 1). The lump was sent to another hospital for biopsy, and the results showed a submucosal spindle-cell tumor with surface-tissue necrosis that was inclined toward leiomyoma (Figure 2). Slices of the lump were taken to our institution for immunohistochemical analysis, which indicated that it came from a submucosal neurofibroma. Three days later, the female was given a colonoscopy, which showed a neoplasm with a smooth surface at the ileocecus (Figure 3A) with polyps at the sigmoid colon (Figure 3B). After admitting the female as an inpatient, a computed tomography (CT) scan of the abdomen was performed, revealing a hypoattenuating tumor of the ascending colon (Figure 4).

The patient then underwent an exploratory laparotomy, with primary anastomosis, after optimization for removing the tumor. We found the 5 cm × 6 cm tumor on the ileocecus at surgery but did not find anything else on the affected bowel. Pathologic examination of the resected specimen revealed it was a submucosal spindle-cell tumor of the ileocecus (Figure 5A). Immunohistochemical stains of the resected specimen showed CD117(-), CD34(-), Ki67(+) 1%, Actin(-), S100(+++), Desmin(-), CD10(-), and Dog(-) (Figure 5B). The pathology results confirmed the tumor to be a neurofibroma. The patient did well initially; however, on the 10th postoperative day, the female had an anastomotic fistula (Clavien-Dindo Class I), and finally recovery well.

**DISCUSSION**

Isolated colonic neurofibromatosis is a benign neural tumor of the lower gastrointestinal tract. It can originate from the plexus of Meissner, the plexus of Auerbach, or even the serosa[[4](#_ENREF_4)]. It is can be the onset manifestation of generalized systemic NF1 or MEN2b. Histologically, although isolated colonic neurofibromatosis manifests as a single or multiple high-degree of histologic differentiation of neoplasms or as a diffuse neuronal hyperplasia, it is commonly termed ganglioneuromatosis. In this case, the histology reports alone cannot specify whether it is NF1 or MEN2b because these conditions share some identical features[[5](#_ENREF_5)].

The differing clinical symptoms found in neurofibromatosis of the hindgut tract depend on the lesion characteristics, such as the location, motility, and adjacent structures of the affected tract. Clinical presentation of the lesions can be abdominal pain[[4](#_ENREF_4)], gut obstruction[[6](#_ENREF_6),[7](#_ENREF_7)], palpable masses[[8](#_ENREF_8)], constipation[[9](#_ENREF_9)], or diarrhea[[10](#_ENREF_10)].

In MEN2b, the development of medullary thyroid carcinoma, pheovhromocytoma, and medullary carcinoma[[11](#_ENREF_11)] is a clinical indicator besides what can be seen on the histological exam. In NF1, the development of classic dermal neurofibromas, café-au-lait macules or Lisch nodules[[12](#_ENREF_12)] is an additional clinical indicator. In the current case, our patient showed none of the additional clinical features.

The diffuse form of colonic neurofibromatosis and Crohn’s disease can mimic each other radiographically. Both colonic neurofibromatosis and Crohn’s disease can appear as single or multiple thickened portions of the gastrointestinal tract on CT scans. In one report, a patient was clinically suspected of having Crohn’s disease based on a CT scan, but the diagnosis was corrected to neurofibromatosis after histological exam of the resected bowel[[9](#_ENREF_9)].

The etiology of isolated neurofibromatosis is still not clarified, but circulating nerve factors[[13](#_ENREF_13)] and a neurofibromatosis gene mutation[[14](#_ENREF_14)] were reported to be involved in hyperplasia of the nerve plexus. The primary treatment of isolated neurofibromatosis is surgical removal. Whether further therapy is required depends on the endoscopic findings and the histological exam.

In summary, we report a case of isolated neurofibromatosis with the onset of a lump in the patient’s stool and without any other additional clinical features. Despite its rarity, the neurofibromatosis is the only clinical indicator in this case. We suspect that part or all of the neurofibroma underwent necrosis and fell into the stool.

**Article Highlights**

***Case characteristics***

The unique character and only clinical symptom of this particular case is the patient presents a month-long history of abdominal pain after meals and a lump discharged from intestine with stool without any blood on it. And there is no other clinical presentation.

***Clinical diagnosis***

Ileocecal neoplasia.

***Differential diagnosis***

Appendicitis, cholecystitis, gastroenteritis, colon cancer, and bowel obstruction.

***Imaging diagnosis***

Ileocecal neoplasia.

***Pathological diagnosis***

Ileocecal neurofibroma.

***Treatment***

Ileocecustomy with primary anastomosis.

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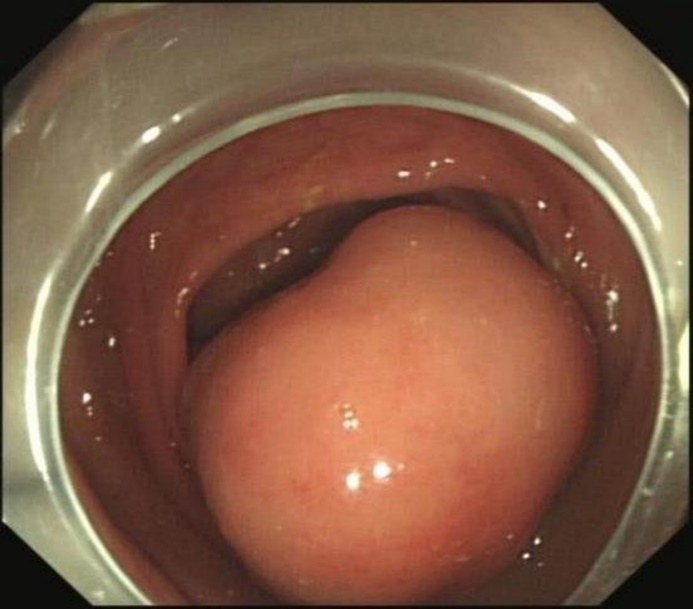


**Figure 1 Photograph of the lump.**

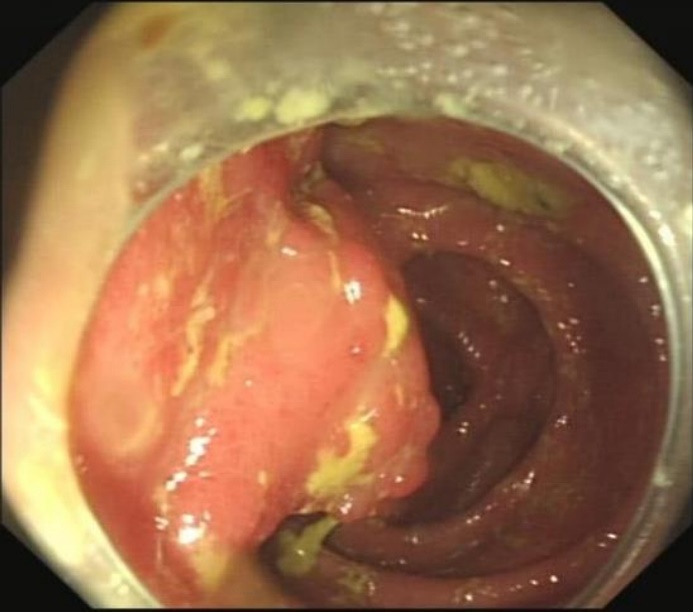


**Figure 2 Proliferation of spindle cells in the lump (HE, × 200).**

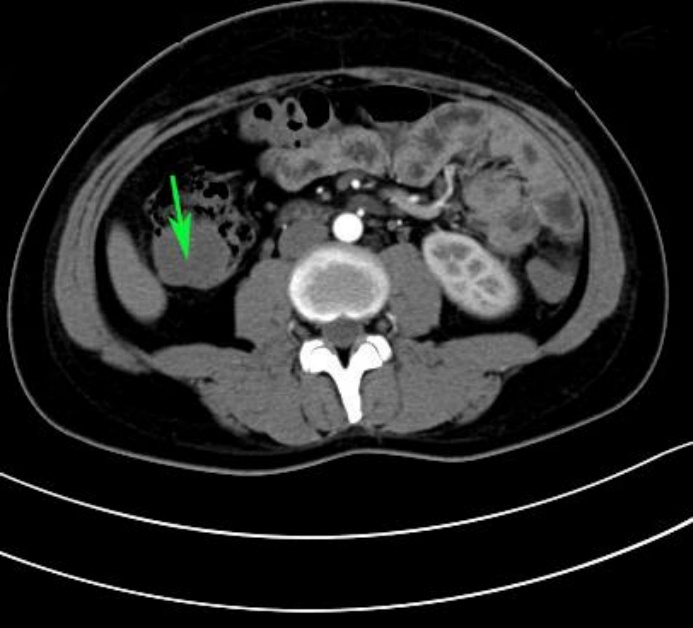
A



B

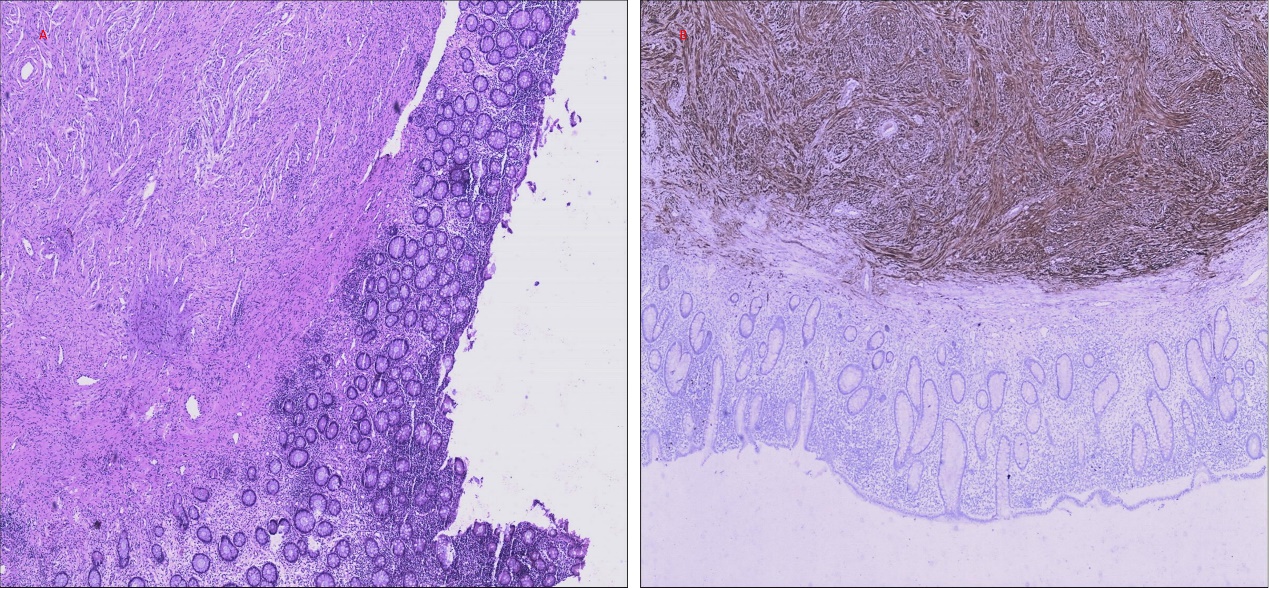


**Figure 3 Endoscopic images of isolated nodularity of the ileocecus (A) and polyps of sigmoid colon (B).**



**Figure 4 Photograph of computed tomography-scan abdomen shows hypoattenuating tumor of the ascending colon (green arrow).**

A B



**Figure 5 Photograph of resected colon (HE, × 200) (A) and immunohistochemical stain for S-100 protein (× 200) (B).**