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Single Case

Distal Duodenal Stricture Secondary to Mesenteric Fibromatosis (Intra-Abdominal Desmoid Tumor) of the Jejunum

Sarah Huang[a] Jamil Mohammad Shah[b] Eduardo Quintero[c] Philip Xiao[d] Armand Asarian[e] Madhavi Reddy[c]

[a]Department of Medicine, New York-Presbyterian Brooklyn Methodist Hospital, Clinical Affiliate of Weill Cornell Medicine, Brooklyn, NY, USA; [b]Division of Gastroenterology and Hepatology, Rutgers New Jersey Medical School, Newark, NJ, USA; [c]Division of Gastroenterology, Hepatology, and Advanced Endoscopy, The Brooklyn Hospital Center, Academic Affiliate of The Icahn School of Medicine at Mount Sinai, Clinical Affiliate of The Mount Sinai Hospital, Brooklyn, NY, USA; [d]Department of Anatomic Pathology, The Brooklyn Hospital Center, Clinical Affiliate of The Mount Sinai Hospital, Academic Affiliate of The Icahn School of Medicine at Mount Sinai, Brooklyn, NY, USA; [e]Department of Surgery, The Brooklyn Hospital Center, Clinical Affiliate of The Mount Sinai Hospital, Academic Affiliate of The Icahn School of Medicine at Mount Sinai, Brooklyn, NY, USA

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Desmoid tumor · Hematochezia · Abdominal bloating · Duodenal stricture

Abstract
Introduction: Mesenteric fibromatosis (intra-abdominal desmoid tumor) is rare, with only a few cases reported in the literature. Clinical symptoms range from asymptomatic, nausea, early satiety, abdominal pain, and gastrointestinal bleeding. Although histologically benign, such a tumor may become locally invasive, and aggressive forms contribute to significant morbidity and mortality. ***Case Presentation:*** We report the case of a 52-year-old West African male with a 1-year history of intermittent hematochezia and intermittent bloating. Colonoscopy revealed a 4-mm rectal polyp and internal hemorrhoids. Esophagogastroduodenoscopy revealed a severe duodenal stricture 4–5 cm distal to the ampulla. Further work-up with contrast-enhanced computed tomography of the abdomen and pelvis revealed a 5.0 × 3.7 × 4.3-cm mass within the mesentery, encasing the distal portion of the duodenum. Exploratory laparotomy was performed, and the mass was excised from the jejunum. Histopathology findings and immunohistochemical analysis revealed the diagnosis to be mesenteric fibromatosis (desmoid tumor), positive for nuclear β-catenin and SMA, and negative expression of STAT6, desmin,

Correspondence to:
Sarah Huang, sarah.huang132@gmail.com

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caldesmon, pan-cytokeratin, or c-KIT. The Ki67 index is <1%. ***Conclusion:*** This case report highlights the diagnostic challenges of mesenteric fibromatosis due to its nonspecific clinical presentation. Recognizing uncommon presentations of mesenteric fibromatosis and risk factors aids in early diagnosis, management, and treatment. Importantly, this also aids in the prevention of complications such as intestinal obstruction, bowel ischemia, and fistula formation.

Introduction

Of all aggressive fibromatosis, also known as desmoid tumors, the intra-abdominal type is the rarest and has the worst prognosis [1]. Mesenteric fibromatoses (intra-abdominal desmoid tumors) are low-grade, monoclonal, fibrous mesenchymal tumors that may range from benign proliferations to high-grade fibrosarcomas [2]. Despite an unpredictable course [3], these tumors tend to infiltrate and recur locally, with no metastasis [1, 4], and may compress nearby organs, nerves, and vessels.

Desmoid tumors are extremely rare, comprising 0.03% of all neoplasms and less than 3% of all soft tissue tumors [5]. The incidence of desmoid tumors occurring intra-abdominally is the rarest of desmoid tumors [1]. Among the general population, the estimated incidence of desmoid tumors is two to four per million per year [6]. Desmoid tumors most commonly affect individuals in the third and fourth decades of life [7] and have a predominance of women in their reproductive age [2, 3] in and around pregnancy or with oral contraceptive usage [8] but can affect anyone and all ages [2]. There is no significant predilection by race or ethnicity [9].

Most cases of desmoid tumors are sporadic and are associated with surgery, trauma, or estrogen levels [1]. Hereditary cases are associated with familial adenomatous polyposis (FAP) or adenomatous polyposis coli (APC) mutations. Common manifestations of intra-abdominal desmoid tumors include the presence of a palpable solid abdominal mass, abdominal pain, and in select cases, no discomfort [1].

Here, we highlight the variable clinical features of intra-abdominal desmoid tumors, thus making clinical diagnosis challenging. The CARE Checklist has been completed by the authors for this case report, attached as supplementary material (for all online suppl. material, see https://doi.org/10.1159/000538489).

Case Presentation

A 52-year-old obese West African male patient with type II diabetes presents to the gastroenterology clinic with a 1-year history of intermittent hematochezia and intermittent postprandial bloating. He denies any current or recent history of anorexia, anemia, weight loss, fevers, chills, abdominal pain, constipation, nausea, vomiting, or diarrhea. He had no prior history of esophagogastroduodenoscopy (EGD) nor colonoscopy. The patient denies any family history of cancer, previous history of surgery or trauma, or smoking, alcohol use, or illicit drug use.

On physical examination, the patient had no signs of acute distress. On abdominal inspection, there were no scars, striae, nor bruising. Normal bowel sounds were auscultated. There were no tenderness on superficial or deep palpation, and no palpable masses. No organomegaly was noted.

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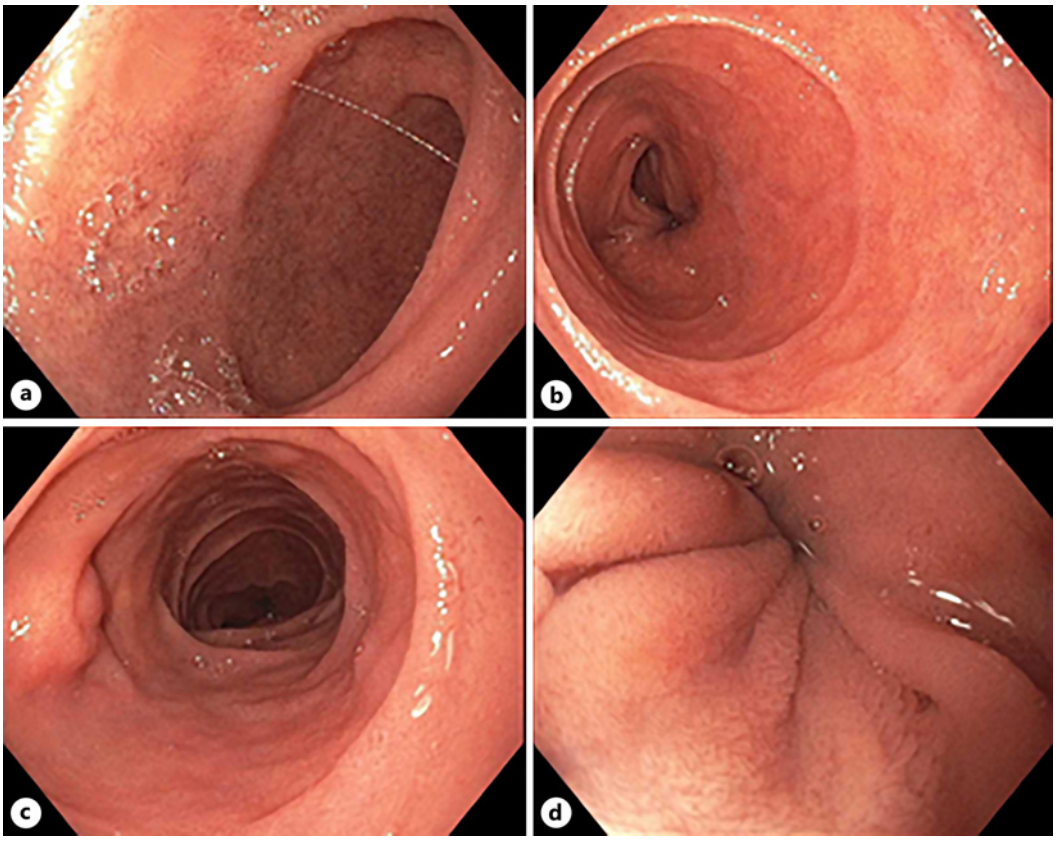

Fig. 1. EGD findings. **a** Duodenal bulb. **b** 2nd portion of duodenum, area of stenosis seen distally. **c** 2nd portion of duodenum; ampulla is at the 9 o' clock position. **d** 3rd portion of duodenum (area of stenosis).

The patient underwent an elective EGD and colonoscopy to evaluate his symptoms. Colonoscopy revealed one 4-mm rectal polyp, which was resected, as well as internal hemorrhoids. EGD revealed a severe duodenal stricture 4–5 cm distal to the ampulla (shown in Fig. 1), suspicious for extrinsic compression of the duodenum. The patient was recommended admission to the hospital for further imaging evaluation of the tight stricture at the distal duodenum on EGD. His vital signs remained stable. Complete blood count and iron profile returned within normal range. Blood urea nitrogen, creatinine, and electrolytes were within normal limits. Tumor markers CA 19-9, CEA, and AFP remained within range. Liver enzymes (AST, ALT, and alkaline phosphatase) and pancreatic enzymes (amylase and lipase) were within normal limits.

Abdominal and pelvic contrast-enhanced computed tomography (CT) scan revealed a 5.0 × 3.7 × 4.3-cm mass within the mesentery, encasing the distal portion of the duodenum (shown in Fig. 2), with no signs of obstruction, possibly representing a neoplasm of mesenteric or small bowel origin. The patient underwent exploratory laparotomy and en bloc resection of mesenteric mass, jejunum (with primary anastomosis), and antimesenteric part of 4th portion of duodenum (Heineke-Mikulicz strictureplasty). After surgical excision, the specimen was sent for histopathological analysis.

Microscopic examination revealed the jejunal mesenteric mass to be fibroblastic/myofibroblastic, with uniform spindle cells arranged in sweeping fascicles (shown in Fig. 3). Immunohistochemistry analysis stained positive for nuclear β-catenin and smooth muscle actin, and stained negative for STAT6, desmin, caldesmon, pan-cytokeratin, or c-KIT.

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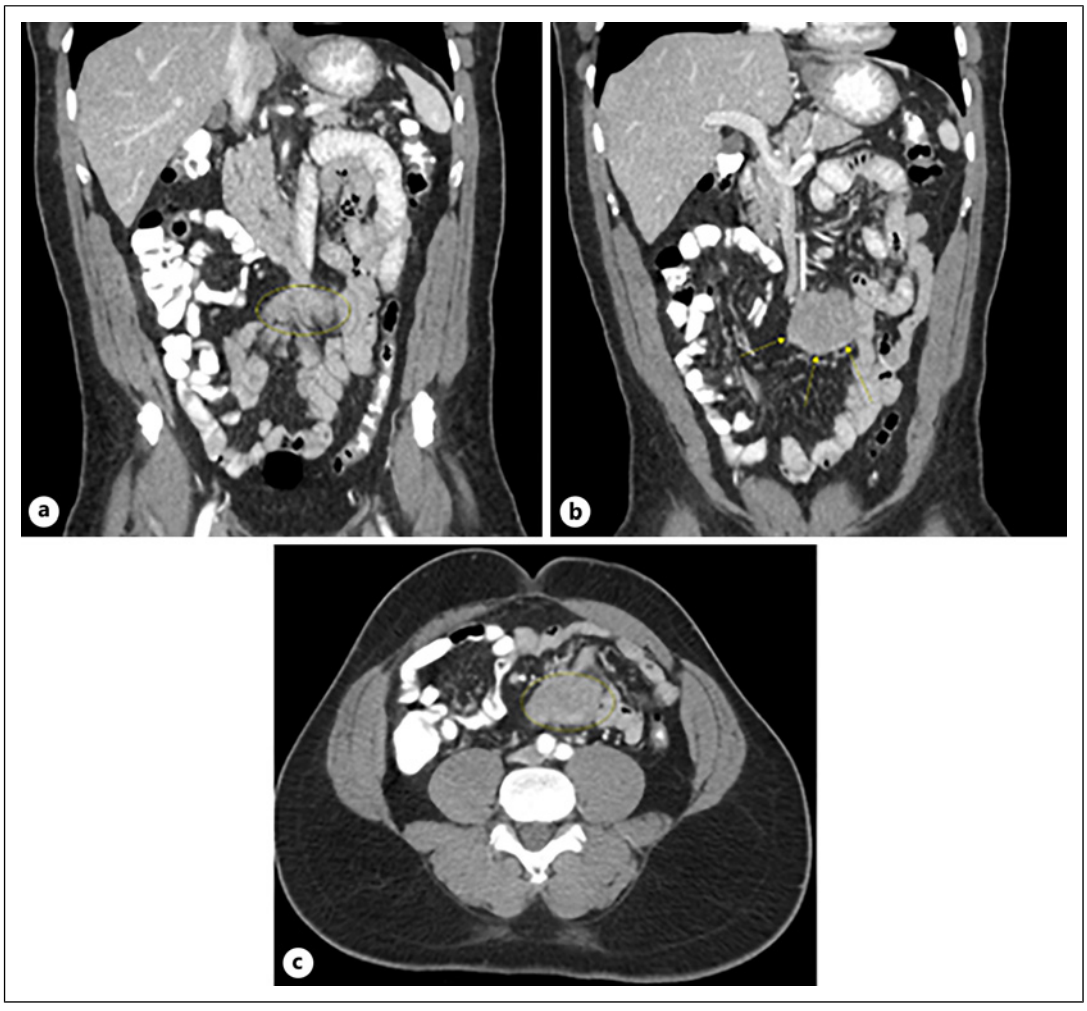

Fig. 2. Contrast-enhanced CT of the abdomen and pelvis. **a** Coronal view of the abdomen: encircled (in yellow) is the mesenteric mass, encasing the distal portion of the duodenum, which also pulls down and tethers the duodenum. **b** Coronal view of the abdomen: yellow arrows point at the jejunum, exhibiting the mass which is inseparable from the jejunum, with likely jejunal collapse. **c** Axial view of the abdomen: encircled is the mesenteric mass at the axial cross section.

The Ki67 index is <1%. The microscopic findings and immunophenotype are consistent with mesenteric fibromatosis (desmoid tumor). The patient was informed and educated of his diagnosis and was recommended regular follow-up in the gastroenterology and surgery clinic.

Discussion

Desmoid tumors, also known as aggressive fibromatosis, are locally infiltrative tumors with no known capacity for distant metastases. They are mesenchymal tumors with a fibrous consistency. Of note, even after total surgical resection, desmoid tumors have an elevated rate of recurrence. Local spread can result in significant morbidity and mortality through destruction of adjacent vital organs and structures.

There is a well-known association between desmoid tumors and FAP – a variant known as Gardner syndrome. While most desmoid tumors occur sporadically, 5 to 15 percent of cases

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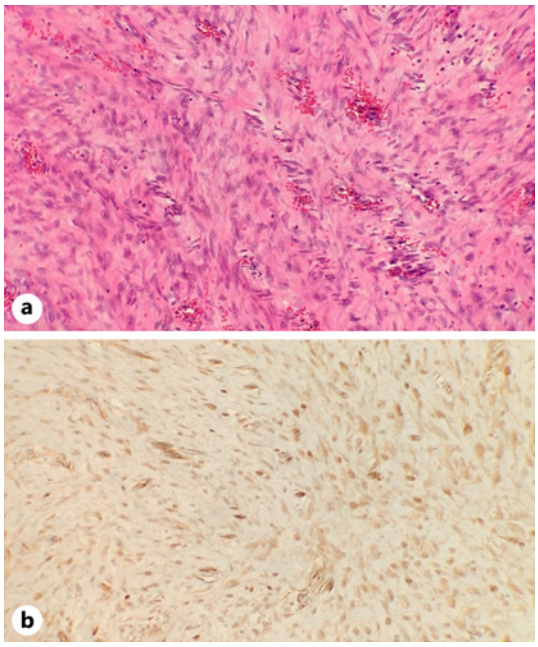

Fig. 3. Histopathology. **a** Microscopic examination reveals spindled cells of uniform appearance and pale cytoplasm in a collagenous stroma with fascicles to herringbone pattern (×20). **b** Immunohistochemical stain reveals beta-catenin nuclear staining in fibroblasts (×20).

are associated with FAP [10, 11]. Conversely, among all individuals with FAP, between 10 and 20 percent are affected by desmoid tumors [12, 13].

The etiology of desmoid tumor formation is only partially understood. However, growing evidence points toward involvement of the APC gene and beta-catenin, both components of the Wnt signaling pathway, in the molecular pathogenesis of desmoid tumor formation – both in Gardner syndrome and in the development of sporadic desmoid tumors [14]. However, in Gardner syndrome, desmoid tumors result from inactivation of the APC gene and ensuing proliferation of beta-catenin in the cells [15]. In contrast, such loss-of-function mutations in the APC gene are rare in sporadic desmoid tumors, which usually result from mutations in the gene for beta-catenin, CTNNB1 [14, 16].

Furthermore, desmoid tumors originating from the small bowel are exceedingly rare [7]. There are only a few such cases reported in the literature [17, 18]. Many individuals with intra-abdominal desmoid tumors – either in the bowel or, more commonly, the mesentery – are asymptomatic. However, particularly later in the disease course, patients can present with such signs and symptoms as palpable mass, nausea, early satiety, abdominal pain, and gastrointestinal bleeding [18]. Further complications include intestinal obstruction, bowel ischemia, ureteric obstruction, functional deterioration in ileoanal anastomosis (characteristically seen in patients who have undergone colectomy for FAP), abscess, and hepatic pneumatosis [19, 20]. In a few cases, the desmoid tumor has invaded the intestinal wall, causing a fistula [8, 17, 21].

Contrast-enhanced CT scan or magnetic resonance imaging is necessary to define the relationship of the desmoid tumor to adjacent structures. This way, resectability and the need for therapy can be properly evaluated. On CT scan, the desmoid tumor typically appears as a well-circumscribed homogeneous mass. It may be isodense or hyperdense with respect to the muscle. Also seen, at times, are cases of a heterogeneous mass with an infiltrative outer margin. Unfortunately, there are no radiographic features that can reliably differentiate between desmoid tumors and malignant soft tissue tumors. Thus, histological examination of a biopsy specimen and immunohistochemistry are useful to establish the diagnosis. Staining for beta-catenin is essential to confirm a desmoid tumor. The spindle cells commonly stain for

vimentin, smooth muscle actin, and nuclear beta-catenin but are usually negative for desmin, cytokeratins, and S-100 [4, 8].

For intra-abdominal desmoid tumors, surgical resection with a wide negative margin is the preferred treatment when feasible [22]. After resection, surveillance and active follow-up are crucial as desmoid tumors have a high rate of recurrence.

For patients with unresectable disease or who are not good candidates for surgery, or in the setting of disease recurrence or progression, radiation therapy and systemic therapy are available treatment options. Radiation therapy is indicated for the treatment of positive margins and large unresectable tumors. However, due to its toxicity to the surrounding structures, it is seldom used in the treatment of intra-abdominal desmoid tumors. While there are several options for neoadjuvant systemic therapy, including antihormonal therapy, tyrosine kinase inhibitors, and chemotherapy, the response rates are generally low [23]. Systemic therapy is available as a palliative therapeutic option as well.

Conclusion

The occurrence of aggressive fibromatosis (desmoid tumor) is a rare finding, and even more so, their occurrence in the small or large bowel occurs infrequently. It is important for clinicians to recognize uncommon presentations as well as risk factors for early diagnosis, management, and treatment of desmoid tumors, given their aggressive potential. Additionally, due to its high local recurrence rate, it is important to continue close outpatient follow-up and regular surveillance.

Statement of Ethics

The authors have no ethical conflicts to disclose. Informed written consent was obtained from the patient of the case report including use of details of medical case and publication of accompanying images. The case report was reviewed, and the need for approval was waived by The Brooklyn Hospital Center, Institutional Review Board.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

Conception and design for the manuscript by S.H. and J.S. S.H., J.S., E.Q., A.A., and M.R. contributed to the drafting and critical revision of the article for important intellectual content and approval for the final version of the manuscript. P.X. contributed pathology slides and captions for the article.

Data Availability Statement

The data are not publicly available due to privacy or ethical restrictions. All clinically relevant information is included in the article. Further inquiries can be directed to the corresponding author.

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