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Case Report
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Plexiform Fibromyxoma of the Colon: A Case Report

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Abstract

Introduction: Plexiform fibromyxoma is a rare mesenchymal tumor found in the gastrointestinal tract, most commonly in the gastric antrum. Limited data exists regarding a plexiform fibromyxoma arising primarily from the colon.

Case Report: We report the first case of a plexiform fibromyxoma arising from the colon in a 43 year-old male with an unremarkable past medical and surgical history. After presenting to his primary care provider with vague abdominal pain, the patient was found to have a right upper quadrant mass on imaging that was surgically resected with a laparoscopic-assisted right colectomy for presumed adenocarcinoma of the colon. However, pathology revealed that the mass was a benign plexiform fibromyxoma.

Discussion: The differential diagnosis of plexiform fibromyxoma in the colon includes more common colon masses like adenocarcinoma as well as spindle cell lesions including Gastrointestinal Stromal Tumor (GIST), schwannoma, plexiform neurofibroma, leiomyoma, leiomyosarcoma, and inflammatory myofibroblastic tumor. Immunohistochemical staining and genetic testing can help differentiate these etiologies and better characterize management and prognosis.

Conclusion: In a patient presenting with vague abdominal pain with imaging findings of a colonic mass, it is crucial to maintain a broad differential diagnosis. In this paper, we describe the first case of plexiform fibromyxoma arising primarily from the colon.

Keywords: Plexiform fibromyxoma; Mesenchymal tumor; Spindle cells; Abnormal abdominal imaging

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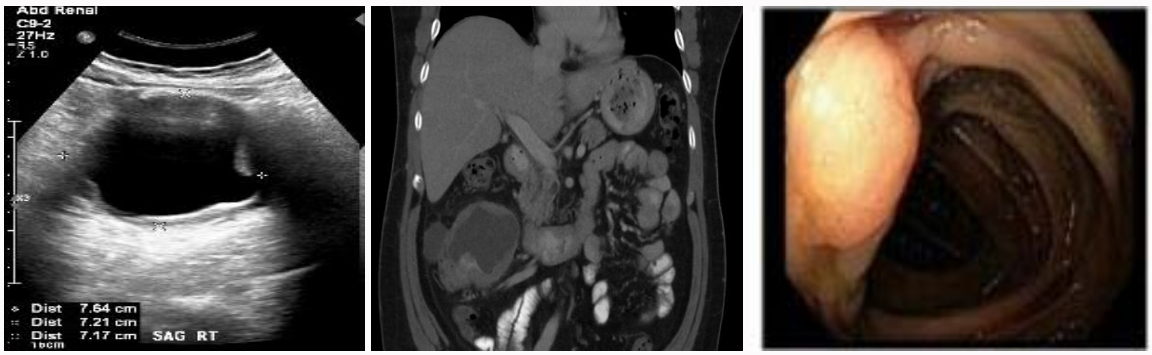
Introduction

First described by Takahashi and colleagues in 2007 as plexiform angiomyxoid myofibroblastic tumor, plexiform fibromyxoma is a rare mesenchymal tumor found in the gastrointestinal tract [1,2]. Nearly all cases of plexiform fibromyxoma have been reported in the antrum of the stomach [3], with scattered rare cases reported in the duodenum [4], esophagus [5], jejunum [6] and ileocecal region [7]. We here report the first case of plexiform fibromyxoma arising in the colon. This work is compliant with the SCARE Guidelines [8].

Case Presentation

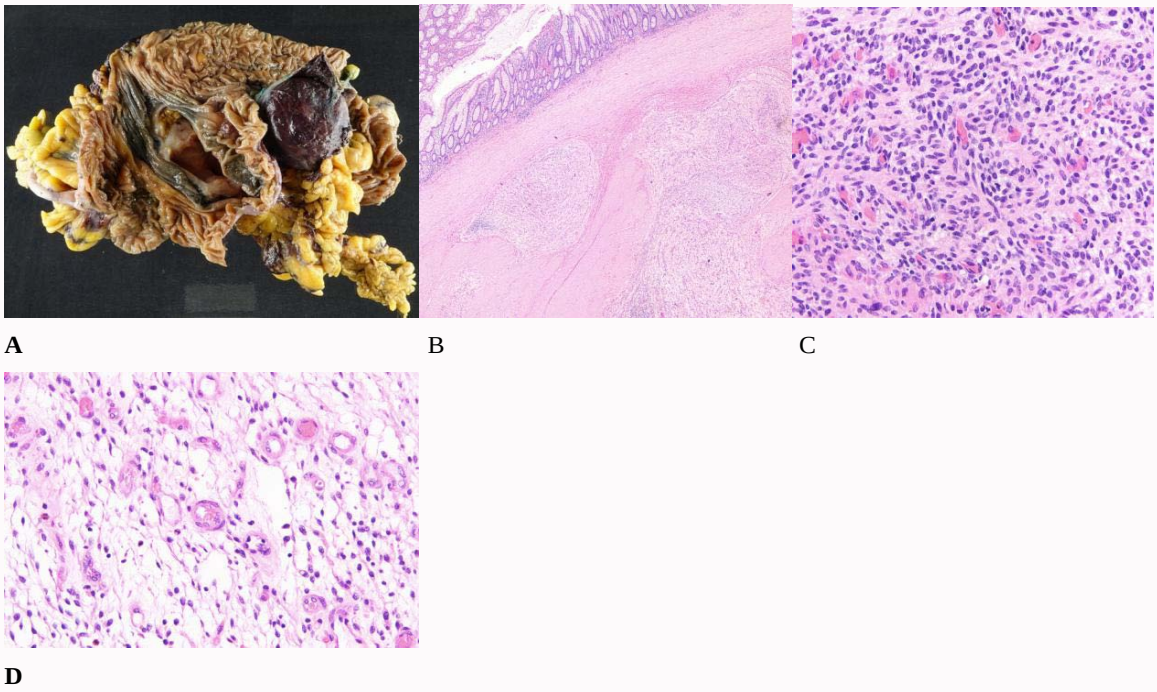
This 43-year-old male presented to Beth Israel Deaconess Medical Center (Boston, MA, USA) for evaluation of right-sided abdominal pain. He did not report any fevers, chills, night sweats, unintentional weight loss, changes in bowel habits, nausea, vomiting or rectal bleeding. His past medical history is notable for hypertension, gastroesophageal reflux disease and obstructive sleep apnea. He had no history of previous surgeries. He is a never user of alcohol or recreational drugs. He is a current smoker of less than 10 cigarettes per day. The patient has no known family history of Crohn's disease, colitis or colorectal cancer. The patient's general physical exam did not demonstrate any abnormalities. There was no palpable mass found in the abdomen. The patient had a previous abdominal ultrasound 7 years prior for RUQ abdominal pain that was normal.

Work up began with an ultrasound demonstrating a cystic mass with a thickened irregular wall seen in the right abdomen of unclear origin (Figure 1A). Follow up CT demonstrated a bi-lobed cystic tumor that measured 9 cm × 5.1 cm with a thick enhancing wall and localized



A B C

Figure 1: Ultrasound of right upper quadrant (A), coronal CT scans of the abdomen (B), and laparascopic view of the proximal ascending colon. C) There is a well-defined cystic mass arising from the proximal ascending colon.



A B C

D

Figure 2: (A) Gross specimen (B) reveals a 6.4cm mass arising from the proximal ascending colon and histology demonstrates a plexiform, multinodular spindle cell lesion (C) with areas of hypercellularity (D) and hypocellularity.

lymphadenopathy (Figure 1B). Interestingly, at this point in his work-up the patient's symptoms had resolved entirely and evaluation was continued because of the initial radiographic abnormality. The patient then underwent a colonoscopy which revealed a semi-circumferential non-bleeding 6 cm mass in the proximal ascending colon that was biopsied (Figure 1C). Pathology revealed focal low-grade dysplasia and colonic fragments with reactive changes, but no invasive carcinoma. The patient underwent an additional CT torso for staging of a presumed malignancy, which demonstrated no new thoracic findings but worsening lymphadenopathy near the mass.

Based on concern for malignancy given his imaging, the patient underwent a laparoscopic-assisted right colectomy. The gross pathology revealed a tumor that consisted of two conjoined masses coming from the wall of the colon. The first was an 8.6 cm firm, white intramural cystic mass that protruded through the mesentery. The second mass, a hemorrhagic cystic, measured 6.4 cm and similarly protruded through the mesentery (Figure 2A). We theorize this self-limited bleed into the wall of the colon is what caused the initial symptoms that lead to identifying the mass.

Histology demonstrated a plexiform, multinodular spindle cell lesion (Figure 2B) infiltrating the wall of the colon, with areas showing extension through and ulceration of the colonic mucosa. The nodules were composed of tumoral spindle cells within a fibromyxoid background that demonstrated a prominent vascular component. Cellularity was variable, with some regions appearing hypercellular (Figure 2C) and others appearing hypocellular (Figure 2D), in keeping with most reported examples of this neoplasm. The spindle cells themselves were bland and uniform, with rare enlarged nuclei and inconspicuous nucleoli. Mitotic activity and areas of necrosis were absent. The tumor stained diffusely positive for smooth muscle actin, with focal desmin positivity. Immunohistochemical stains for c-kit/CD117, DOG1, cytokeratin MNF116, and CD34 were all

negative within the tumor cells. Fluorescence *in situ* hybridization was negative for the t(11;12)(q11;q13) *MALAT1-GLI1* fusion reported in occasional cases of plexiform fibromyxoma, though it did demonstrate MALAT1 nuclear amplification, of uncertain significance.

The patient underwent a primary ileocolic anastomosis, recovered uneventfully and given the minimal risk of recurrence or malignancy will be followed expectantly.

Discussion

Plexiform fibromyxoma is a rare and benign mesenchymal tumor characterized by a bland and uniform pattern of spindle cells arranged in a characteristic plexiform/multinodular architecture [2]. Given their predilection for the gastric antrum in close proximity to the pylorus, some plexiform fibromyxoma scan present with pyloric obstruction and associated weight loss; however, gastrointestinal bleeding is described as the most common presenting symptom [3]. For this patient, the presenting complaint was abdominal pain, with no evidence of gastrointestinal bleed or weight loss. We theorize that the patient's presenting symptoms were due to a self-contained bleed into the mass or the wall of the colon, but not into the lumen. The initial findings of a cystic mass on abdominal ultrasound and the follow up colonoscopy with biopsy that revealed low grade dysplasia warranted a CT scan for presumed malignancy. The initial colonoscopic biopsy obtained too superficial a sample to demonstrate the final pathology. The finding of a cystic tumor with wall enhancement on CT has been reported in other cases of plexiform fibromyxoma of the antrum [8]. However, similar CT findings may be seen in patients with colonic malignancies. Given their benign features on pathology, surgical resection has an excellent prognosis and is the standard treatment for patients with plexiform fibromyxoma [9].

The differential diagnosis of plexiform fibromyxoma in the colon includes more common colon masses like adenocarcinoma, as well as other spindle cell lesions in the colon including Gastrointestinal Stromal Tumor (GIST), schwannoma, plexiform neurofibroma, leiomyoma, leiomyosarcoma, and inflammatory myofibroblastic tumor. A key step in narrowing the differential for these masses is immunohistochemical staining and molecular testing.

Adenocarcinoma is the most common primary colon malignancy. Gland formation is usually evident, and cells would stain with keratin antibodies, such as MNF116, a cytokeratin monoclonal antibody [10]. GIST is rare in the colon but common in other parts of the GI tract; it is also a mesenchymal neoplasm composed of spindled cells that stain positive for CD117, DOG1, and CD34; most cases harbor a *KIT* or *PDGFRA* mutation [11,12]. Schwanomma is a mesenchymal nerve sheath tumor that can arise in the colon; it stains positive for S-100 as well as GFAP [13,14]. Similarly, plexiform neurofibroma, a structurally complex mass arising in large nerve trunks, will also stain positive for S-100, but it is usually found in patients with underlying neurofibromatosis type 1 [15]. Leiomyoma and leiomyosarcoma are smooth muscle neoplasms that stain positive for both SMA and desmin [16]. Finally, inflammatory myofibroblastic tumor, a tumor characterized by a lymphoplasmacytic inflammatory infiltrate in addition to spindle cells, tends to stain positive for ALK1 and can demonstrate *ALK* abnormalities [17].

In addition to immunohistochemical staining, molecular testing provides addition diagnostic value. A subset of plexiform fibromyxomas have been shown to have a *MALAT1-GLI1* translocation that results in overexpression in GLI1 protein, which is a component of the Sonic Hedgehog signaling pathway and has control of oncogenic genes [18]. While this patient's tumor was negative for this translocation, there was nuclear amplification of uncertain significance of *MALAT1*.

Conclusion

Overall, the diagnosis of plexiform fibromyxoma in this patient's colon was an incidental diagnosis found on imaging and confirmed with pathology in this patient who initially had a suspected colonic malignancy. This rare, benign mesenchymal neoplasm typically arises in the stomach; we report the first case arising in the colon, demonstrating that it should be considered in differential diagnoses in this location.

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.

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