

Unusual presentation of desmoid tumor in the small intestine: a case report

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Abstract Desmoid tumor originating from the small intestine is extremely rare. We report a 50-year-old man who presented with the sudden onset of severe abdominal pain. Computerized tomography (CT) demonstrated a huge homogeneous tumor in the lower abdomen that appeared to be in continuity with the distal ileum. The mass adherent to the ileum was resected and proved to be a desmoid tumor. The patient has remained recurrence free on follow-up.

Keywords Desmoid tumor · Aggressive fibromatosis · Small intestine · Peritonitis

Introduction

Desmoid tumors (desmoid-type fibromatosis) are rare, deep-seated, usually unencapsulated, slowly growing fibroblastic neoplasms that arise from fascial or musculoaponeurotic structures [1]. Desmoids account for about

0.03% of all neoplasms and less than 3% percent of all soft tissue tumors. The estimated incidence in the general population is two to four per million populations per year [1]. Although desmoids are considered benign because they do not metastasize, they may infiltrate adjacent tissues, causing organ disruption that may in some cases be fatal. Desmoid tumors are generally classified as intra-abdominal or extra-abdominal. Most intra-abdominal desmoids arise in the mesentery, particularly of the small bowel, and are in fact the most common primary tumor of the mesentery [1–3]. We report an intra-abdominal desmoid arising in an unusual location.

Case report

A 50-year-old man presented with the sudden onset of sharp, cramping, peri-umbilical, non-radiating pain. The pain was associated with diaphoresis, but the patient denied nausea, vomiting, diarrhea, melena, hematochezia, a change in bowel habits, or weight loss. The pain was so intense that he immediately went to the emergency room. His past medical history was unremarkable. In particular, he had no history of previous abdominal surgery.

On physical examination, he appeared to be in pain. His blood pressure was 89/60 mmHg, temperature 40.5°C, and pulse 120 beats per minute. The abdomen was distended with peri-umbilical and rebound tenderness and muscle guarding. All four extremities were cool. His leukocyte count was 3,200/mm³ with 2% bands and 78% neutrophils.

Decubitus plain abdominal radiography revealed a soft tissue density in the lower abdomen. Computerized tomography (CT) demonstrated a huge homogeneous tumor in the same area that appeared to be in continuity with the distal small intestine. There was a small loop of

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Fig. 1 Contrast-enhanced abdominal CT showing a huge homogeneous tumor abutting a small bowel loop and surrounding a distal loop of bowel that contains an intraluminal air-fluid level (arrow)



Fig. 2 Intra-operative photograph of a huge mass on the anti-mesenteric side of the small intestine; the resected mass is tan-to-white

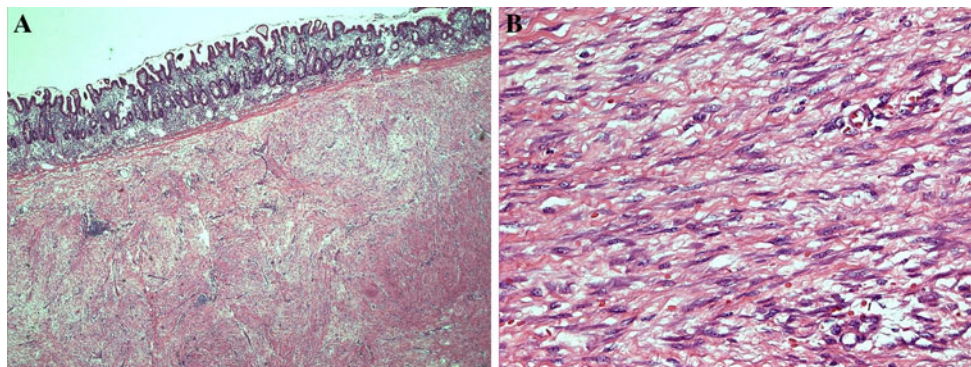


Fig. 3 Photomicrograph showing **a** infiltration of the muscularis propria and submucosal layer of the small intestine without penetration of the mucosa (HE stain, $\times 40$) and **b** bland spindle and stellate

bowel with an air-fluid level that seemed to be encased by the tumor, suggesting possible strangulation (Fig. 1). At laparotomy, the tumor was seen to arise from the anti-mesenteric side of the ileum 60–80 cm proximal to the ileocecal valve. The tumor itself and the involved small bowel were diffusely hyperemic. The tumor and a segment of small bowel were resected. Ascites and blood culture all grew *Escherichia coli*.

Grossly, the resected tumor was a white-to-tan, firm, encapsulated intramural tumor (Fig. 2). Microscopically, it involved the muscularis propria and submucosal layers of the bowel wall without penetrating the overlying mucosa or invading the mesentery (Fig. 3a). It was composed of bland spindle and stellate cells arranged in sweeping bundles deposited in a dense collagenous background (Fig. 3b). The appearance was consistent with a desmoid tumor but was also reminiscent of other mesenchymal tumors. However, on immunohistochemical study, the tissue was negative for CD117, smooth muscle actin, desmin, and S-100 protein. There was also no *KIT* or *PDGFR* gene mutation, ruling out gastrointestinal stromal tumor. Mutation of the beta-catenin gene was present, supporting the diagnosis of desmoid tumor.

The patient recovered and had no evidence of tumor recurrence 24 months after resection.

Discussion

The most unusual feature of this patient's desmoid tumor was its origin from the anti-mesenteric side of the ileum, unlike most reported abdominal desmoids that arise in the mesentery [3]. Rarely, desmoids have been reported in the esophagogastric junction [4], pancreas [5], appendix [6], and ileum [7]. Though we cannot prove it, we speculate that this patient's tumor may have arisen from the wall of the small bowel and grown out from there.

The etiology of desmoids has not been well defined, but several factors have been implicated, including in some cases mutations in the APC and beta-catenin genes, and trisomy of chromosomes 20 and 8 [8–11]. This is why the beta-catenin gene mutation in our patient was helpful for diagnosis.

Most patients with intra-abdominal desmoids are asymptomatic for long periods. Only late in the course are there signs or symptoms such as a palpable mass, abdominal pain, or gastrointestinal bleeding [3]. Reported complications of intra-abdominal desmoids include ureteric obstruction, intestinal obstruction, intestinal ischemia, deterioration in an ileoanal anastomoses, abscess, and hepatic pneumatosis [8, 12–16]. In a few cases, desmoids have invaded the bowel wall, resulting in a fistula and translocation of intestinal bacteria into the tumor [17–19]. Our patient's acute abdominal pain and peritonitis might have been caused by strangulation or micro-perforation of a loop of bowel by this huge tumor.

The diagnosis of a desmoid tumor can only be established histologically, and our patient's findings were characteristic. However, various mesenchymal tumors in the gastrointestinal tract or abdominal cavity may have partially overlap of the gross appearance, histology, and immunophenotype [20]. In our patient, negative results of staining for desmin, smooth muscle actin, and S-100 excluded other mesenchymal tumors such as fibrosarcoma, leiomyoma, leiomyosarcoma, and malignant peripheral nerve sheath tumor. The clinical, radiologic, and gross appearance of the specimen was most suggestive of gastrointestinal stromal tumor or desmoid. But as noted in the case report, the lack of staining for CD117 by immunohistochemistry and the absence of a *KIT* or *PDGFR* gene mutation were helpful in ruling out the former [20, 21], while the beta-catenin gene mutation was consistent with the diagnosis of a desmoid tumor.

Despite complete resection with negative microscopic margins, desmoid tumors have recurrence rate of about 40% [1, 22, 23]. It is therefore suggested that surgery should be performed only when absolutely necessary [9]. This was the case with our patient who had a potentially life-threatening complication. He has remained recurrence free for 2 years. There is no standard protocol for follow-up of patients with desmoid tumors, but we have followed him clinically and performed a CT in the 1 year after surgery.

In conclusion, we report a case of a desmoid tumor arising from the anti-mesenteric side of the small bowel. Despite the unusual location, both histology and genetic study helped confirm the diagnosis.

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