An unexpected intrabdominal discovery: a mesenteric desmoid tumor with small bowel obstruction. Case report and literature review

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Abstract

This case report describes a 54-year-old man who presented with abdominal discomfort, nausea, early satiety, and constipation. A CT scan revealed a large mass infiltrating the mesentery and close to bowel loops, raising suspicion of intestinal sub-occlusion. Urgent surgery was performed, resulting in a segmental jejunum resection to remove the identified mesenteric mass. Histological examination revealed a diagnosis of desmoid fibromatosis which is a rare type of soft tissue tumor with borderline malignancy that can cause bowel obstruction when it develops intra-abdominally. It is often associated with genetic predispositions and risk factors such as previous surgery and pregnancy. The case highlights the importance of considering a patient's surgical history when encountering desmoid fibromatosis to determine the most appropriate treatment approach.

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Key words: desmoid fibromatosis; soft tissue tumor; bowel obstruction; mesenteric mass; surgery trauma.

Conflict of interest: the authors declare no potential conflict of interest, and all authors confirm accuracy.

Ethics approval and consent to participate: no ethical committee approval was required for this case report by the Department because this article does not contain any studies with human participants or animals. Informed consent was obtained from the patient included in this study.

Patient consent for publication: the patient gave his written consent to use his data for the publication of this case report and any accompanying images.

Availability of data and materials: all data underlying the findings are fully available.

Received: 12 May 2023.
Accepted: 11 July 2023.

Case Reports in Emergency Surgery and Trauma 2023; 1:13
doi:10.4081/crest.2023.13

Introduction

In the United States and Western Europe, the third common cause of mechanical small bowel obstruction is tumors,[1] contest of which we can distinguish external to the bowel (extrinsic) neoplasm and within the wall of the bowel (intrinsic) neoplasm.

Desmoid fibromatosis is a rare type of soft tissue tumor that can develop in the abdomen, more precisely in the mesentery. It is a benign lesion but characterized by the tendency to aggressive local invasion and local recurrence,[2] and it can cause bowel obstruction. Treatment is justified in case of symptomatic or ingrowing mass and surgery can be the first line of therapy even if intra-abdominal desmoids are less resectable compared to the extra-abdominal desmoids. Post-operative follow-up is important because desmoid tumors are characterized by a high rate of local recurrence even after complete resection.[2]

Case Report

The patient was a 54-year-old man with no significant medical history except for an appendectomy performed a year earlier due to acute appendicitis (histological examination confirmed the diagnosis of appendix inflammation). He presented with abdominal tension, nausea, early satiety, constipation, and general discomfort.

During the clinical evaluation, his abdomen was distended and slightly painful, but there were no signs of peritoneal irritation. Notably, Murphy's and Blumberg's signs were negative. His biochemical profile showed normal results, including negative inflammation indices, normal electrolyte values, liver and renal indices, and negative oncological markers.

The abdominal CT scan revealed a large rounded mass with irregular profiles located within the mesenteric adipose tissues in the right iliac fossa, measuring 65x61x78mm. The mass was near adjacent bowel loops but without infiltration or encasement of any vascular vessels (Figures 1 and 2).

Chest CT scan findings were negative for lymphadenopathy and focal lesions. These radiological images raised suspicion of an intestinal GIST, lymphoma, or mesenteric primary tumor causing intestinal sub-occlusion, which could potentially progress to com-

plete intestinal occlusion. Due to the mass's proximity to the small bowel loops and limitations in obtaining a definitive diagnosis through biopsy due to a poor US window, urgent surgical intervention was deemed necessary.

During exploratory laparoscopy, a solid 7cm mass infiltrating the root of the jejunal mesentery and adjacent to the corresponding jejunal loop, approximately 40cm from Treitz, was identified. Another smaller mass with similar characteristics was found nearby. Minimal intraperitoneal fluid was observed in the Douglas pouch and collected for cytology, while no evidence of peritoneal or hepatic metastasis was found.

Considering the large size and position of the identified mass, the procedure was converted from laparoscopic to open intervention to ensure safe completion. A mini-laparotomy was performed, and approximately 70cm of the jejunum and corresponding mesentery were resected. A mechanical L-L antiperistaltic anastomosis was performed, and cholecystectomy was also conducted due to adenomyosis identified during the CT scan.

Histological examination of the surgical specimen confirmed the diagnosis of desmoid fibromatosis with negative microscopic resection margins (R0 resection). Genetic analysis revealed a T41A mutation in the CTNNB1 gene. The patient was scheduled for follow-up evaluations, including clinical assessment and abdominal CT scans at three and six months post-surgery, which revealed no significant findings. During the latest clinical evaluation, the patient reported feeling well and did not experience any clinical disturbances, except for mild constipation. The next follow-up is planned for six months.

Discussion

Desmoid fibromatosis, also known as desmoid tumors, are rare connective tissue tumors that arise from fibroblastic proliferations in fascial and aponeurotic tissues. These tumors are benign, slow-growing neoplasms with a tendency for aggressive local invasion and local recurrence, even after surgical resection. However, they do not metastasize to distant sites.[3] Desmoid tumors account for approximately 0.03% of all neoplasms and fewer than 3% of all soft tissue tumors[4] and are most commonly affecting individuals between the ages of 15 and 60, with a slightly higher incidence in females.[5] They typically occur in the extremities, neck, thoracic and abdominal wall, and abdomen, including the mesentery and small bowel. Intra-abdominal desmoid tumors are even rarer, representing 8% of all desmoid tumors (8% of all desmoid tumors).

Most desmoid tumors arise sporadically, but approximately 5% to 15% are associated with familial adenomatous polyposis (FAP).[6] In patients with intra-abdominal desmoid tumors, screening for FAP, such as colonoscopy and genetic screening, is recommended due to the genetic predisposition; however, the chance of discovering FAP through screening is relatively low. Screening may be more useful for patients with a higher risk of having FAP, such as those under 40 years of age, with intra-abdominal or retroperitoneal tumors, multifocal disease, or a family history of FAP.[2]

Other factors that contribute to the development of desmoid tumors include local trauma, particularly surgery. In patients with FAP, there is a 75% chance of developing desmoid tumors after surgery.[3]

Pregnancy is also considered a risk factor due to exposure to elevated hormone levels and pregnancy-related trauma.[7]

The etiology of desmoid tumors is still unknown, but emerging evidence suggests a dysregulated wound-healing process involving the APC and beta-catenin genes in the pathogenesis of these tumors and other fibroblastic lesio10ns. In normal conditions, beta-catenin protein levels are elevated in fibroblasts only during the proliferative phase of wound healing (Wnt/beta-catenin pathway). Mutations in the APC or beta-catenin genes can lead to a persistent accumulation of beta-catenin in the nucleus, promoting proliferation and enhanced survival of fibroblast.[2]

In the majority of sporadic desmoids, we can find somatic APC mutations or activating mutations of CTNNB1:[8] the three most common mutations in CTNNB1 gene are T41A, S45F, and S45P[9] instead mutation in any location of APC gene may occur in sporadic desmoid. Increasing evidence points to the involvement of both APC and beta-catenin genes in the molecular pathogenesis of inherited desmoids and these case mutations in the 3' end of APC gene seem to correlate with higher risk of tumor occurrence in patients with FAP.[10]

Cross-sectional imaging, such as CT or MRI, is necessary to define the relationship of the tumor to adjacent structures, assess

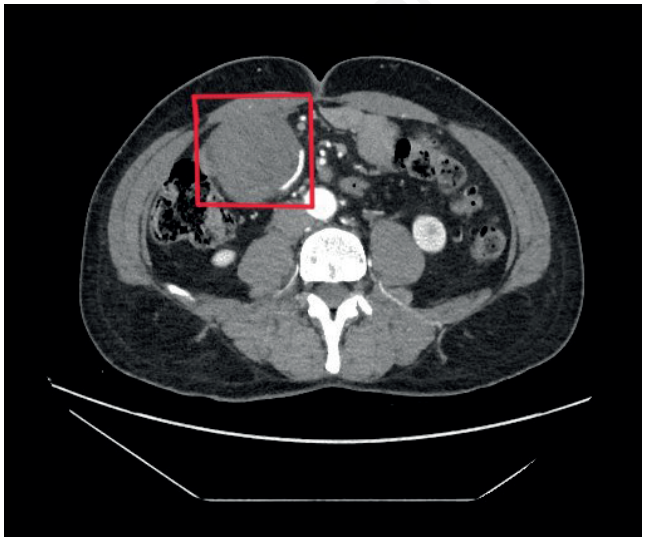

Figure 1. Arterial phase of desmoid tumor at CT scan: a mass with an heterogeneous density and a small arterial vessel passing trough.

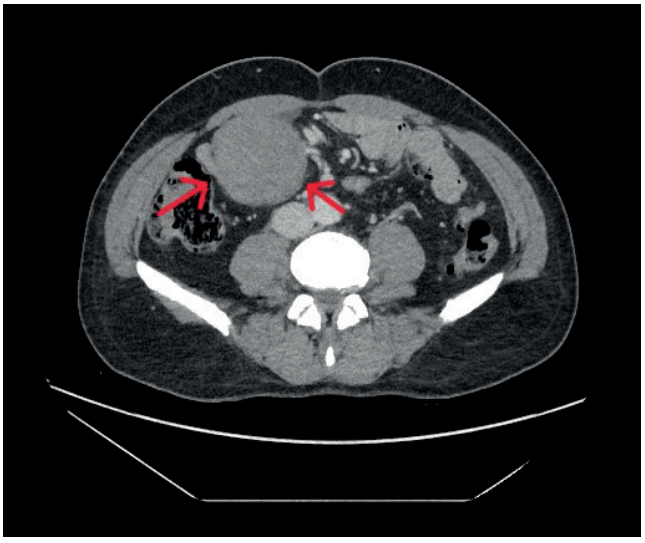

Figure 2. Portal phase of desmoid tumor at CT scan.

resectability, and determine the need for treatment. There are no radiographic characteristics that can reliably distinguish desmoids from malignant soft tissue tumors. CT imaging typically reveals a well-circumscribed, non-encapsulated soft tissue mass that slightly enhances, often heterogeneously, with intravenous contrast. MRI characteristics of desmoid tumors are related to their cellularity and fibrous content, and they can help assess the aggressiveness of the lesion. A hyperintense T2 signal is associated with hypercellularity,[11] and this T2 hyperintensity may diminish over time as tumor cellularity decreases and collagen deposition increases.

The diagnosis of a desmoid tumor can only be established through histological examination of a biopsy specimen. However, performing a biopsy for pathological diagnosis is controversial, as it may accelerate tumor progression, and incisional biopsy also carries the risk of being non-diagnostic.[11]

Desmoid tumors do not have a propensity for regional or distant spread, so staging radiographic studies of other sites is not necessary. As a result, there is no commonly agreed-upon staging system for desmoid tumors.[2]

However, certain types of desmoid tumors, particularly those located in the abdomen, can be life-threatening due to the destruction of adjacent vital structures and organs. Consequently, intra-abdominal desmoid tumors are more likely to be fatal compared to tumors in other locations.[2] Factors that negatively influence prognosis and recurrence rates include distal and intra-abdominal tumor location, multiple tumors, mesentery infiltration or invasion, ureter encasement, tumor size larger than 7 cm, female gender, and age under 37 years.[12]

Consensus-based guidelines from the National Comprehensive Cancer Network (NCCN)[13] suggest observation as the primary therapeutic option for patients with desmoid tumors that are potentially resectable but asymptomatic, non-life-threatening, and not causing significant impairment. Observation is also an option for unresectable tumors or cases where surgery would result in excessive morbidity.[2]

Treatment is indicated for symptomatic patients or those with progressively enlarging tumors, regardless of symptoms, when there is an imminent risk to adjacent structures or when the tumor causes cosmetic concerns.[2] Treatment options include systemic therapy, radiotherapy, and surgery.

When surgery is indicated, the standard goal is complete resection of the tumor with negative microscopic margins (R0).

However, positive margins are acceptable if necessary to preserve function and minimize major morbidity. That's because desmoid tumors have a high rate of recurrence even after complete surgical removal, and the contribution of incomplete resection to local recurrence rates is still unclear.[2]

While some series support role of surgical resection as a valid treatment option when negative margins can be obtained,[14] many series have reported that even patients who undergo aggressive resection with widely negative margins have recurrence rates of 16 to 39 percent.[15] These data have led some to conclude that aggressive attempts to achieve negative resection margins are not warranted if they result in excessive morbidity.

For patients who have large intra-abdominal desmoid tumors, surgery is still a standard approach for resectable tumors. However, the infiltrative nature of the desmoid in this situation often precludes radical surgery with an R0 resection. Medical therapy instead of surgery is often the preferred option for patients with more difficult tumors, such as those involving the mesentery, major vessels, or other critical structures.[16]

Radiation therapy (RT) and systemic therapy are effective primary treatment options for patients who are not suitable candidates for surgery, decline surgery, or would experience excessive surgical morbidity.[17] Systemic therapy can be broadly classified into cytotoxic chemotherapy (such as pegylated liposomal doxorubicin and doxorubicin), tyrosine kinase inhibitors (such as sorafenib and sunitinib), hormonal therapy (such as tamoxifen), and nonsteroidal anti-inflammatory drugs (NSAIDs).[18]

There is no evidence-based protocol for surveillance following treatment. Consensus-based guidelines from the NCCN recommend a history and physical examination, along with appropriate imaging every three to six months for two to three years, followed by annual evaluations.[13]

In cases of recurrent disease, a multimodality approach is crucial, considering systemic therapy, surgery, and radiation therapy as potential treatment options.[2,18]

Conclusions

In conclusion, desmoid fibromatosis is a tumor that can develop as a result of previous trauma, including previous surgery, so patients with this pathology should be evaluated for a history of surgical trauma or, in the case of women, previous pregnancies. Screening for familial adenomatous polyposis (colonoscopy, genetic screening) may be warranted in certain cases. Treatment decisions should be based on the individual patient and tumor characteristics, with options including systemic therapy, surgery, and radiation therapy. Surgery, when indicated, can be performed with a laparoscopic approach first even in emergencies, depending on the surgeon's experience; in addition, laparoscopy has double the advantage of a diagnostic and therapeutic role. Close postoperative follow-up is crucial due to the high rate of local recurrence, and further research is needed to establish standardized surveillance protocols and improve treatment outcomes for desmoid fibromatosis patients.

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