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Case Report

Aggressive Fibromatosis of the Left Mesocolon Mimicking a Gastrointestinal Stromal Tumor: A Case Report

Mohammad Abu-Jeyyab[a] Hanan Al-Asbahi[b] Mohammad Al-Jafari[a]
Bushra Khalaf Al-Tarawneh[c] Abdulqadir J. Nashwan[d]

[a]School of Medicine, Mutah University, Al-Karak, Jordan; [b]General Surgery Department, Al-Basheer Hospital, Amman, Jordan; [c]Pathology and Microbiology Department, School of Medicine, Mutah University, Al-Karak, Jordan; [d]Hamad Medical Corporation, Doha, Qatar

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Fibromatosis · Gastrointestinal stromal tumor · Mesocolon · Preoperative diagnosis

Abstract
Mesenteric fibromatosis (MF) is a proliferative fibroblastic lesion of the intestinal mesentery. It constitutes 8% of all desmoid tumors, representing 0.03% of all neoplasms. It is benign histologically, although it could infiltrate locally and recur following excision; however, it is free from the potential to metastasize. It is spontaneous or associated with familial adenomatous polyposis (FAP]) mutation as a part of Gardner's syndrome. This case report discusses the radiological, intraoperative, and histopathological findings from a 45-year-old male patient who presented with abdominal pain and a palpable mass in the left hemiabdomen. The pain was dull and aching, extending to the back and unrelated to any other gastrointestinal symptoms. There was no history of severe weight reduction. Furthermore, he is not a smoker. There were no comorbidities, severe medical diseases, or prior surgical procedures. Computerized tomography revealed a well-defined, lobulated, heterogeneously enhancing altered signal intensity mass at the mesocolon. Ultrasonography of the abdomen showed an intra-abdominal mass.
Macroscopic mass characteristics include a well-defined mass measuring 22 × 14 × 11 cm connected to a small intestine segment measuring 21 × 2 × 2 cm. Histopathological and immunohistochemical examinations of the resected tumor, including positive nuclear immunostaining for beta-catenin, confirmed a postoperative diagnosis of desmoid-type fibromatosis. Based on its clinical presentation and computed tomography results, this case demonstrated how desmoid-type fibromatosis of the colon might mimic gastrointestinal

Correspondence to:
Abdulqadir J. Nashwan, anashwan@hamad.qa

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stromal tumors (GISTs). Due to the varied therapies and follow-up methods used for these lesions, the differential diagnosis between desmoid-type fibromatosis and GIST is clinically significant.

Introduction

Aggressive fibromatosis or desmoid fibromatosis (DF) is a rare, aggressive entity of abdominal wall tumors. This type of tumor is characterized by the fact that it is aggressive but usually localized [1]. The reported incidence is 2–4 cases per million people yearly [2]. DF can be subdivided into two main categories: abdominal and extra-abdominal types. The more common type is the abdominal one, which arises from the abdominal wall. It is thought to have a higher rate in women who had a recent pregnancy. However, it can also be classified according to the genetic biases of sporadic and familial desmoid fibromatosis [1–3]. Here, we present a case of a case of 45-year-old man diagnosed as having abdominal desmoid fibromatosis by the histopathology examination. We will present the case, go through the investigations, and discuss the treatment plan for this patient. The CARE Checklist has been completed by the authors for this case report, attached as online supplementary material (for all online suppl. material, see https://doi.org/10.1159/000534038).

Case Presentation

A 45-year-old man presented with a 6-month history of mild to moderate left-side abdominal pain. The pain was dull aching, radiating to the back without being associated with any other abdominal symptoms, including nausea, vomiting, or change in bowel habits. There was no history of any significant loss of weight or previous episodes of the same symptoms in his life. The patient denied any traumatic event. Additionally, he is not a smoker. There were no comorbidities or chronic medical illnesses, or previous surgeries.

The remaining patient's history is unremarkable. By examining the patient, he is conscious, oriented, and alert, looking well with normal vital signs, no neck lymphadenopathy, and a normal chest. On abdominal examination, a freely mobile mass approximately 20 × 20 cm in size, smooth, firm consistency, and well-defined margins was present on the left side of the abdomen.

Investigations

Ultrasonography (USG) of the whole abdomen showed it as an intra-abdominal mass (mesenteric mass), and there is no hydronephrosis or free fluid collection. Computerized tomography for the abdomen revealed well-defined, lobulated, heterogeneously enhancing altered signal intensity mass at the mesocolon (Fig. 1).

The Laboratory Tests Were Normal

The patient was counseled for the management plan, all the treatment options were discussed clearly; however, he refused chemotherapy – imatinib. As a result, the surgeon decided to do a subtotal mass excision and the patient agreed. The surgical procedure findings

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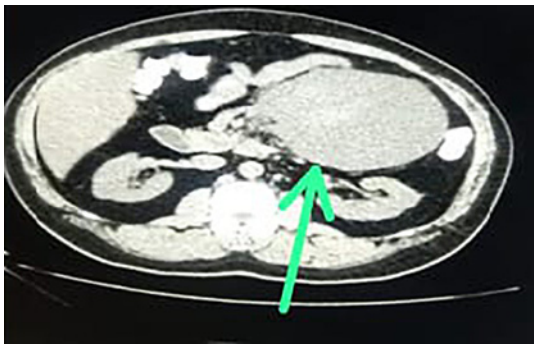

Fig. 1. Abdominal CT scan showing the abdominal mass.

were that a solid mass originated near the posterior abdominal wall at the upper para midline area wrapped by an adherent small intestine. Because of the unfavorable circumstances, a near-total mass excision was done after restricting a severely adherent small bowel segment.

Histopathology Examination

Macroscopic Features

Specimen fixed in formalin consists of a well-defined mass measuring 22 × 14 × 11 cm attached to a small bowel fragment measuring 21 × 2 × 2 cm (Fig. 2). Step sectioning of the mass revealed a whitish-whorled cut surface.

Microscopic Description

The section revealed a cellular proliferation arranged in fascicles and focal areas of storiform architecture associated with prominent and thin-walled blood vessels (Fig. 3). The tumor extends through the bowel to the muscularis mucosa (Fig. 4). The cells were spindle, elongated, and bland with wavy nuclei, pale pink to gray cytoplasm, and inconspicuous nucleoli (Fig. 5). Keloid-like collagen bands were also seen. Rare mitotic figures were seen. No nuclear hyperchromasia or necrosis is seen. The mass was incompletely excised. Immunohistochemical stains were done and showed the following:

- β-catenin: nuclear positive staining
- SMA: focally positive
- Dog1: noncontributory
- CD117: negative
- CD34: not available.

Based on clinical history, physical examination, and imaging, the preoperative differential diagnosis was either DF or gastrointestinal stromal tumor. However, after mass resection and histopathology examination results, particularly immunohistochemical stains with β-catenin positive nuclear staining, the gastrointestinal stromal tumor was excluded, and the diagnosis of DF was confirmed. Figure 6 shows the patient's case starting from the first presentation.

Discussion

Desmoid tumors are interestingly uncommon well-differentiated musculoaponeurotic neoplasms, representing 0.03% of all neoplasms and 3% less than all soft tissue tumors [4] and with almost 3.7 new cases occurring per one million individuals on an annual basis [5].

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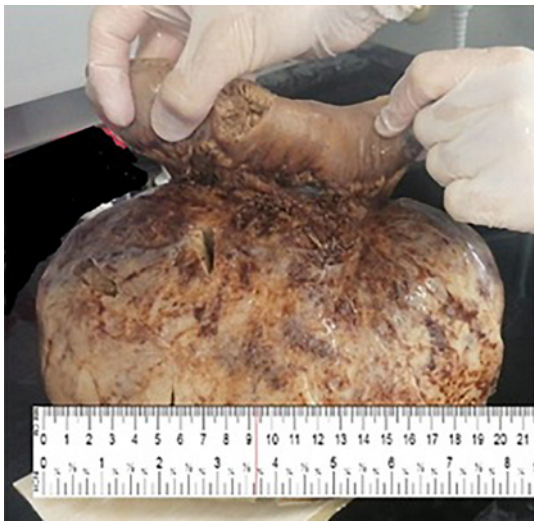

Fig. 2. Gross appearance of the mass.

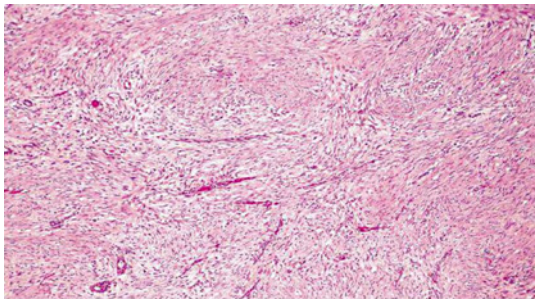

Fig. 3. Cellular proliferation arranged in fascicles and focal areas of storiform architecture associated with prominent and thin-walled blood vessels.

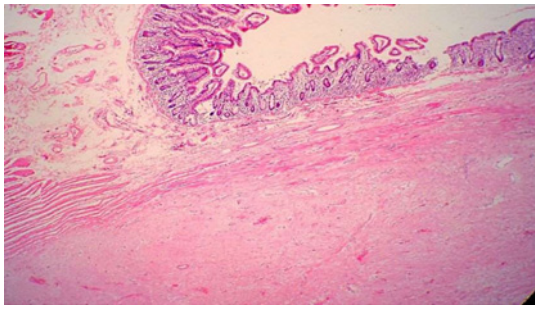

Fig. 4. Tumor extension through the bowel to the muscularis mucosa.

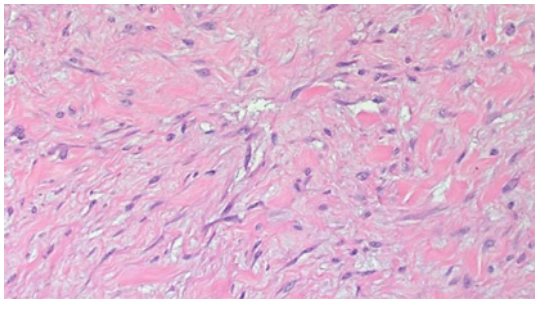

Fig. 5. Cellular characteristics of the tumor.

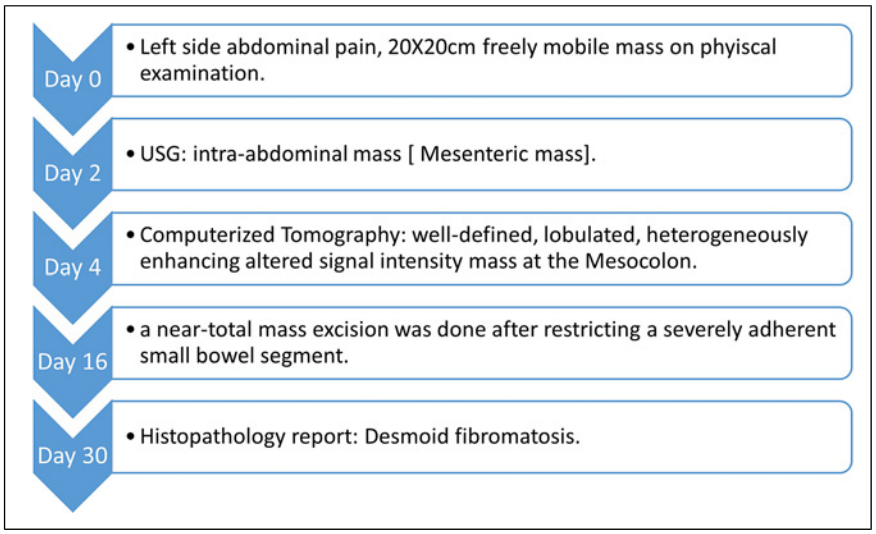

Fig. 6. Diagram showing the patient's case starting from the first presentation.

It is widely known that desmoid fibromatosis is not cancerous per se; however, the infiltrative proliferation of this type of neoplasm might have fatal consequences [6]; in that, they have a high rate of recurrence; recurrence is present in ≤45% of cases, attributed to difficult access to surgical resection. Besides, the effect of compression of neighboring organs may lead to fatalities [5].

Desmoid tumors might occur in any part of the body. However, it is primarily located either abdominally or extra-abdominally. For further elaboration, many studies suggest that approximately 37–50% of DFs are initiated in the abdominal region [5].

Abdominal DFs may occur sporadically or be associated with familial adenomatous polyposis (FAP). FAP with DF is known as Gardner syndrome. An inactivating mutation of the APC gene causes this syndrome [5, 6].

Extra-abdominal desmoids arise from the muscular or nonmuscular connective tissue such as fascia or intramuscular septa. Muscles of the shoulder, pelvic girdle, and thigh of young adults are mainly affected. The head, neck, pelvis, and mesentery desmoids are seen in clinical practice [6, 7]. Several studies revealed that DFs are correlated with trauma and females, with more prevalence in those who have experienced pregnancy but are not common in menopause.

Also, there is no predominant age group as these tumors might occur at any age, from 15 to 60 years old [5, 7]. In this presented case, the patient is a male in his forties and has no factors that might have contributed to having a desmid tumor such as traumatic history, wound scars, or family history. Clinically, there are no specific symptoms.

Mostly, desmoids present as non-painful slow-growing masses; however, this presentation depends, in the first place, upon the growth's location. For instance, mesenteric fibromatosis arises from the small bowel mesentery yet might originate from the gastrocolic ligament, ileocolic mesentery, and omentum. Furthermore, intra-abdominal desmoids ordinarily have no symptoms [4], though ureteral obstruction, intestinal perforation, fistula, or functional impairment of ileoanal anastomosis following colectomy in FAP cases might be seen [8].

On the other hand, should this type of tumor size cause significant compression of adjacent viscera, intestinal obstruction, bowel ischemia, and hydronephrosis can occur as a result of vascular compression and ureteric compression, respectively [4].

In this reported case, the patient's main complaint was the dull aching abdominal pain rather than the enormous abdominal mass, which has gradually increased in size. Undoubtedly, recently advanced imaging modalities have played a significant role in establishing the diagnosis of fibromatosis. Ultrasound, computed tomography scan, and magnetic radiology imaging are used for evaluation in such cases.

Ultrasonography has a variable echogenicity with smooth, well-defined margins [5]. In addition, through CAT scans, most of these tumors appear as well-circumscribed homogeneous masses that could be hyperdense or isodense concerning muscular density [4].

In the imaging workup of our case, abdominal-pelvic USG of the presence of a mesenteric mass with no hydronephrosis or free fluid collection while a CAT scan revealed a well-defined, lobulated, heterogeneously enhancing altered signal intensity mass at the mesocolon.

In the discussed case, MRI shows low signal intensity relative to muscle on T1-weighted images and variable signal intensity on T2-weighted images. MRI scans indicate how the tumors are likely to behave, with a bright signal indicating a high-water content, which has been correlated with rapid growth though MRI scans are also used in diagnosing DFs, it was not used as a workup modality for our patient [5].

A definitive diagnosis must be established with histopathological analysis. As microscopic detection of nuclear localization of beta-catenin protein by immunohistochemistry study confirms the diagnosis of DFs [6]. Moreover, the immunohistochemical response for actin might be partially positive, and immunohistochemical muscle cell markers may differentiate desmoids from fibrosarcoma [5]. In our case, the histopathological study showed the presence of spindle, elongated cellular proliferation with positive nuclear staining of β-catenin.

Several means of treatment include surgical-wide excision, antiestrogen therapy, radiotherapy, and chemotherapy [8]. Nevertheless, the efficacy of these means is unpredictable, still, Complete resection of the tumor with negative microscopic margins is the standard surgical goal but is often constrained by anatomic boundaries [4]. Though it was suggested that abdominal wall DF is more accessible to detach from the underlying tissues with lower recurrence rates than that mesenteric or retroperitoneal fibromatosis. There is a 25–50 percent recurrence regardless of radical surgery and adjuvant radiotherapy. On the other hand, neoadjuvant radiotherapy was inefficient as it has been tried for local control [8]. Our patient has been managed by performing a subtotal excisional surgery.

Gastrointestinal stromal (GISTs) and fibromatoses are both rare mesenchymal neoplasms that can develop in the gastrointestinal system. They do, however, have unique clinical, radiological, and pathological characteristics that must be distinguished for appropriate diagnosis and therapy. GISTs are more frequent in elderly people, but fibromatoses can develop at any age [9]. GISTs often manifest as subepithelial masses that can cause bleeding, blockage, or perforation, whereas fibromatoses are infiltrative lesions that can compress or shift neighboring tissues [10, 11]. GISTs are well-defined, heterogeneous masses with contrast enhancement and necrosis or cystic degeneration on radiology, whereas fibromatoses are homogenous, low-attenuation masses with smooth borders and no necrosis or cystic degeneration [10]. GISTs are pathologically composed of spindle or epithelioid cells with variable mitotic activity and immunohistochemical expression of CD117, CD34, and DOG1, whereas fibromatoses are composed of bland spindle cells with low mitotic activity and immunohistochemical expression of beta-catenin, smooth muscle actin, and desmin. As a result, differential diagnosis of GISTs and fibromatoses is critical to avoid misdiagnosis and incorrect therapy [11].

The majority of GISTs are caused by mutations in the KIT or PDGFRA genes, which can be inhibited by tyrosine kinase inhibitors such imatinib, sunitinib, regorafenib, and ripretinib [12]. Some GISTs, however, do not have these mutations and are designated as wild-type GISTs. Succinate dehydrogenase (SDH), an enzyme involved in the Krebs cycle and oxidative

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phosphorylation, is defective in around 50% of them [13]. When compared to other GIST subtypes, SDH-deficient GISTs show different clinical, pathological, and molecular characteristics. They are more common in younger individuals and are typically associated with genetic disorders such as Carney triad or Carney-Stratakis syndrome [14]. They also exhibit a distinct gene expression profile, which is distinguished by the activation of hypoxia-inducible factor 2 and its downstream targets [15]. Tyrosine kinase inhibitors' significance in SDH-deficient GISTs is debatable, as they appear to have limited effectiveness and may promote resistance mechanisms [16]. As a result, there is an unmet need for innovative therapeutic solutions for this difficult patient population. Targeting alternative pathways implicated in SDH-deficient GISTs, such as fibroblast growth factor receptor, mammalian target of rapamycin, or immune checkpoint inhibitors, has recently been investigated [17, 18]. These novel systemic therapies have the potential to improve the outcomes of SDH-deficient GISTs and need further exploration. Furthermore, identifying predictive biomarkers for these therapies is critical for selecting the best patients and optimizing their clinical benefit.

In our case, we report the case of a 45-year-old man with mesenteric fibromatosis. The gold standard for the treatment of mesenteric fibromatosis is complete resection of the tumor with negative microscopic margins, but it is often limited by anatomic boundaries [4]. So, the patient was treated with subtotal mass excision after restricting a severely adherent small bowel segment. Postoperative histopathology confirmed the initial diagnosis of mesenteric fibromatosis.

Conclusion

Mesenteric fibromatosis is a rare clinical entity. Therefore, it should be considered in middle-aged patients with unspecific symptoms or unexplained abdominal mass presentation. Constant follow-up for those diagnosed with DFs is recommended to detect recurrence should it occur.

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Statement of Ethics

The article describes a case report. The Ethics Committee at Al-Karak governmental hospital waived ethical approval. Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author contributions

Mohammad Abu-Jeyyab, Hanna Al-Asbahi, Mohammad Al-Jafari, and Abdulqadir J. Nashwan have contributed to writing and reviewing the manuscript. Bushra Khalaf Al-Tarawneh did the histopathological examination and revised the final manuscript. All authors have read and approved the manuscript.

Data Availability Statement

All data generated or analyzed during this study are included in this article and its online supplementary material. Further inquiries can be directed to the corresponding author.

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