Case Report

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Retroperitoneal desmoid-type fibromatosis: a case report

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Introduction and importance: Desmoid-type fibromatosis (DF) is a rare subtype of soft tissue sarcomas that most commonly occurs in the anterior abdominal wall. When occurring in the retroperitoneum, DF is usually part of familial syndromes while only rarely sporadic. This makes it imperative to report any instance of experience with DF and the oncological outcomes of the different approaches to management. We report two cases of sporadic and severe DF occurring in the retroperitoneum at our institution.
Case presentation: The first case is a male that presented with urinary obstruction symptoms and underwent surgical resection of the tumor that extended into the left kidney. The second case is a female with a history of recurrent desmoid tumors of the thigh and was incidentally diagnosed with retroperitoneal DF on imaging. She underwent tumor resection and radiotherapy; however, the tumor recurred with urinary obstruction symptoms that required another surgical resection. Histopathological characteristics and radiological imaging of both cases are described below.
Clinical discussion: Desmoid tumors often recur, thus significantly influencing the quality of life which is reflected in one of our cases. Surgery remains a mainstay treatment, and both cases presented in this report required surgical resection of the tumors as symptomatic and curative measures.
Conclusion: Retroperitoneal DF is a rare entity, and our cases add to the scarce literature available on the topic, which may well contribute to the formulation of practice-changing recommendations and guidelines focused on this rare variant of DF.

Keywords: aggressive fibromatosis, case report, desmoid, desmoid-type fibromatosis, soft tissue sarcoma

Introduction

Desmoid-type fibromatosis (DF), also known as desmoid tumor or aggressive fibromatosis (AF), is a rare entity on the soft tissue sarcomas (STS) spectrum of tumors accounting for only 3% of all STSs and 0.03% of all neoplasms. Desmoid tumors are mesenchymal in origin and tend to occur in deep soft tissues[1]. DF most commonly occurs in the anterior abdominal wall and shoulder girdle, while a sporadic occurrence in the retroperitoneum is quite rare[2].

Given this information about DF, we are reporting our experience with two cases of retroperitoneal desmoid tumors at the American University of Beirut Medical Center (AUBMC).

HIGHLIGHTS

- Sporadic desmoid-type fibromatosis (DF) occurrence in the retroperitoneum is rare.
- DF has a high recurrence rate affecting the quality of life.
- Surgery remains the only definitive treatment for DF.
- Future research on the value of immunotherapy for managing DF may improve prognosis.

Methods

Ethics approval for this study (EC Reference Number BIO-2017-0412) was provided by the Institutional Review Board (IRB) at AUBMC. Informed consent was waived, as per the IRB, as no identifying patient information was used, and the patient data was retrieved from the SARCOMA database, which was created by retrospective chart review at AUBMC.

The work reported in this manuscript is in line with the Surgical CAse REport (SCARE) 2020 guidelines[3].

Case presentation 1

The first case is a 53-year-old male with no significant past medical, genetic, or relevant familial history who presented with a complaint of urinary urgency and left leg edema and weakness. Upon presentation, a computed tomography (CT) scan with intravenous contrast was done that revealed a retroperitoneal mass and left hydro-uretero-nephrosis secondary to the obstruction by the tumor. The biopsy done was nonrevealing. The mass effect of the tumor, in addition to the finding of

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infiltration into surrounding tissues such as the psoas muscle, made STS the highest on the differential. A blood workup done revealed normal values with a hemoglobin level of 13.5, total protein level of 72, and albumin/globulin ratio of 39 : 33. The patient underwent surgery for resection of the left kidney, the retroperitoneal mass and an attached part of the left psoas, and left internal iliac vessels were resected. Surgery was performed by a general surgeon specializing in surgical oncology. The surgical pathology revealed an irregular nonhomogenous mass, dark blue to brown, with a smooth and rough outer surface and focal lobulation measuring $13 \times 9 \times 4.7$ cm in size and 280 g in weight. In addition, the specimen showed a firm white gray-tan fibrous mass with whorled bands infiltrating focally the surrounding skeletal muscles, mainly psoas, and soft tissues, primarily iliac vessels. The tumor's sections were composed of fascicles and bundles of elongated spindle-shaped cells surrounded by a densely collagenous stroma in a vascular background. These pathological findings were consistent with a diagnosis of DF. The kidney parenchyma revealed patchy interstitial inflammation with focal cortical scarring. The patient's hospital stay was uneventful. The patient was lost to follow-up after the surgery. No data is available on further treatments that he received or recurrence.

Case presentation 2

The second case is of a 22-year-old female with a history of recurrent extra-abdominal desmoid fibromatosis of the left thigh since age 16, for which she underwent surgical resection each time and a family history of breast cancer that presented with no symptoms. Upon follow-up imaging by MRI of the pelvis and lower extremities, a mass was noted in the left retroperitoneal pelvic area. Knowing the patient's history, the differential diagnosis of the pelvic infiltrating mass was narrowed down to a probable DF. The patient underwent surgical excision by a general surgeon with extensive expertise in retroperitoneal surgery. The operation yielded a $10.5 \times 6.5 \times 3.5$ cm irregularly shaped tumor. On histopathological examination, the tissue had an inner surface with a whorl-like appearance intermixed with some adipose tissue consistent with a diagnosis of DF. No areas of hemorrhage, necrosis, or calcifications were noted. The patient had an uneventful hospital stay. Follow-up CT scan (Fig. 1) of the abdomen and pelvis done at 5 months postoperation revealed a soft tissue structure in the anterior aspect of the left pelvis. The differential diagnosis was narrowed down to either a displaced ovary from postsurgical adhesions or a residual tumor.

After this point, the patient was lost to follow-up until 2 years later, in September 2008, when she presented for gait problems, and imaging confirmed that the tumor recurred in her thigh, and she presented for wide excision of the mass. It is imperative to note that the loss to follow-up was mainly because of the patient's psychological state, which she described as a loss of hope and depression. At this time, she also underwent 27 cycles of radiotherapy to prevent a recurrence. However, by 2010, the patient started having urinary obstruction symptoms, and a CT scan (Fig. 2) of the abdomen pelvis revealed locoregional recurrence and progression of the pelvic mass along the left external and internal iliac chains, extending to involve the obturator internus and paravaginal region while impinging on the left ureter thus causing severe hydronephrosis.

A repeat surgical excision procedure was done, and pathology confirmed that it was a DF recurrence. The patient tolerated all operative surgical resections, had uneventful hospital stays and was discharged in stable status. The last follow-up for the patient was 8 years later when a scan of the left lower extremity revealed a recurrence of the tumor in the thigh but not the pelvis. The patient underwent surgical resection for the thigh tumor with good outcomes and did not present for follow-up later.

Discussion

STS encompass a wide spectrum of tumors that can occur at any anatomic location of the body. The most common locations for soft tissue sarcomas are the thigh and buttocks, followed by the torso and then the retroperitoneum and upper extremity. A rare subtype of STS is DF, also referred to as desmoid tumor or AF. DF most commonly localizes to the abdominal wall, intestinal mesentery, or shoulder girdle[4]. In the rare occasions when DF occurs in retroperitoneum, it is usually within the clinical context of a syndrome such as familial polyposis coli/familial adenomatous polyposis (FAP) or Gardner's syndrome, especially if the patient has a history of abdominal surgery[1]. It usually arises

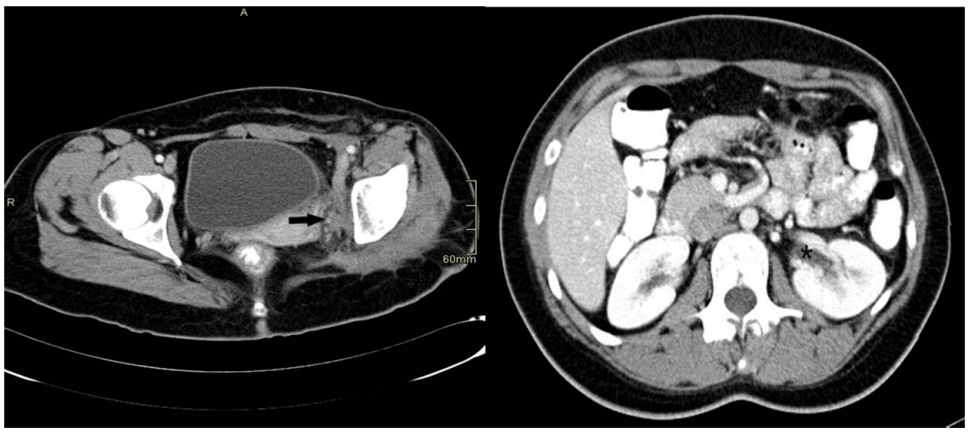

Figure 1. Computed tomography of 2007, axial plane, and contrast-enhanced: there is a small soft tissue mass (black arrow) along the left pelvic sidewall contacting the left external iliac vessels and left obturator internus muscle. The mass does not obstruct the left ureter, and there is no hydronephrosis (asterisk).

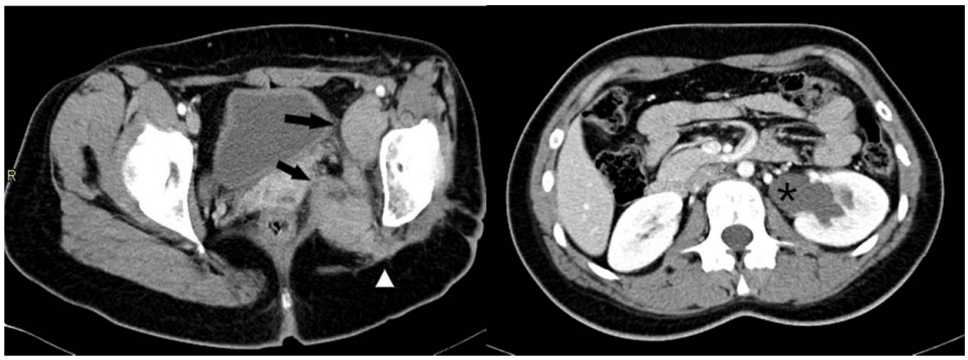

Figure 2. Computed tomography of 2010, axial plane, and contrast-enhanced: the lobulated soft tissue mass (black arrows) along the left pelvic sidewall is increased in size, now encasing the left external iliac vessels, infiltrating the left obturator internus and levator ani muscles, and extending through the sciatic notch (white arrowhead). The mass obstructs the left ureter resulting in moderate hydronephrosis (asterisk).

from the deep musculoaponeurotic planes such as muscle fascia, tendons, and aponeurosis[2]. It is a fibroproliferative process that consists of bands of fibroblasts and myofibroblasts that proliferate and infiltrate in a fascicle-like pattern that disrupts different adjacent structures and anatomical planes[5]. Most desmoid tumors are sporadic in nature, but some (~20%) are associated with familial syndromes such as FAP. This association is suspected in atypical cases such as occurrence in the retroperitoneum or intra-abdominal[6]. These tumors are rarely symptomatic, but when they are, it is mainly due to the mass effect on adjacent vasculature and organs; mostly, they are incidental findings on imaging studies. The pathogenesis of desmoid tumors remains a controversial topic; however, certain factors have been proven to play a role, such as abnormal signaling pathways, genetic mutations, and environmental insults[7]. Diagnosing DF relies primarily on the histological findings of biopsy and may be assisted by imaging [CT, MRI, and positron emission tomography (PET)] features[5]. Unlike other STS subtypes, desmoid tumors do not require staging because it does not exhibit a tendency to spread, neither locally nor distantly. Observation is an acceptable approach to asymptomatic DF, while surgical resection with negative margins remains the mainstay treatment for symptomatic tumors[8,9]. Other treatment strategies include surgery with radiotherapy or radiation therapy alone. Even in cases of complete tumor resection with negative margins, DF has the propensity to recur in up to 39% of cases[10].

Several case reports are reported in the literature on sporadic and nonsporadic retroperitoneal DF, mainly postoperative or in association with FAP[11]. We report two cases of sporadic DF that is not associated with previous abdominal surgeries or Gardner's syndrome. Most cases reported in literature identify positive family history of colorectal cancer in cases of sporadic DF and even syndromic DF[1,11]. However, in both cases, we report they had a positive family history of cancer other than colorectal, pancreatic, and breast. Moreover, both had the retroperitoneal mass presenting with urinary obstruction symptoms upon impingement of the tumor on the ureters, which is also reported in the literature. As in other case reports[2], the primary modality of management is surgical resection. However, reports on the remarkable response of desmoid tumors to tamoxifen are found. This is best explained by the fact that most DF is hormone-sensitive tumors with abundant hormone receptors[12].

Conclusion

Knowing that it is rare for DF to occur in the retroperitoneum, it is paramount to report any encountered cases. In this manuscript, we shed light on the importance of keeping DF on the differential of indolent retroperitoneal growths. Moreover, the cases' progression shows the recurrence potential of DF and the need for surgery for symptomatic management as well as improved quality of life.

Ethical approval

Ethics approval for this study (EC Reference Number BIO-2017-0412) was provided by the Institutional Review Board (IRB) at the American University of Beirut Medical Center (AUBMC).

Consent

Informed consent was waived, as per the IRB, as no identifying patient information was used, and the patient data was retrieved from the SARCOMA database, which was created by retrospective chart review at AUBMC. The authors of this case report recognize the patient's right to privacy and confidentiality, and as such, it was imperative to make sure that no identifying information is mentioned in this report, including, but not limited to, names, initials, dates of birth, medical records numbers, identity-revealing images or any other statement or material. As such, the requirement for the patient's consent for publication was waived as permissible in the HHS Regulations and Policies for retrospective chart reviews that do not reveal patient-identifying information.

Patient consent

The two cases reported in this case report manuscript provided verbal consent allowing the use of their medical history and information for this paper.

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None.

Author contribution

M.H.E.-C.: collected data and wrote the manuscript; D.D.: contributed to data collection; H.K.: contributed by choosing radiological images and coining figure legends; H.T., H.A.H., G.A.S., M.K., and E.S.: each contributed by reviewing and editing the manuscript.

Conflicts of interest disclosure

The authors declare that they have no conflicts of interest.

Research registration unique identifying number (UIN)

1. Name of the registry: not applicable.
2. Unique identifying number or registration ID: not applicable.
3. Hyperlink to your specific registration (must be publicly accessible and will be checked): not applicable.

Guarantor

Dr Mohamad Hadi El Charif and Dr Eman Sbaity are the guarantors of this case report.

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