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Diagnostic and therapeutic dilemmas in intra-abdominal desmoid tumors: A case report and literature review



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ABSTRACT

INTRODUCTION: Intra-abdominal desmoid tumors (DTs) are a rare and anatomically diverse group of locally-aggressive, benign neoplasms. They are often difficult to diagnose, even in patients who possess risk factors for the disease. Even after a diagnosis has been reached, the optimal therapy is often not well-defined.

PRESENTATION OF CASE: The case discussed of a 33-year old male with a giant intra-abdominal desmoid is an example of both the diagnostic and therapeutic dilemmas that arise when confronted with a patient with a DT. Initial confusion over diagnosis led to ineffective therapy, but once the correct diagnosis was made, the patient went on to definitive surgical resection.

DISCUSSION: The differential diagnosis of DTs is broad, and the diagnosis is often delayed due to nonspecific presentations. Immunohistochemistry is crucial in the accurate histological diagnosis, which guides treatment. Chemotherapy and radiation have a role in the management of both primary and recurrent lesions, but surgical resection remains the cornerstone of treatment.

CONCLUSION: DTs present a clinical challenge in their diagnosis and management, and despite providing standard medical and surgical treatment, recurrence rates are high and continued surveillance is crucial.

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1. Introduction

Intra-abdominal desmoid tumors (DTs), are benign locally-aggressive mesenchymal neoplasms that lack the potential for metastasis. DTs are rare as they account for only 0.03% of all neoplasms and less than 3% of all soft tissue tumors [1,2]. Approximately 80% of DTs are sporadic and can occur anywhere in the body, but the balance are associated with pregnancy, trauma and genetic syndromes such as familial adenomatous polyposis (FAP) and are more commonly intra-abdominal [3]. In the absence of these risk factors, intra-abdominal DTs present a diagnostic and therapeutic dilemma for the surgeon.

We present the case of a 33-year-old otherwise healthy male with a giant intra-abdominal DT that masqueraded as a gastrointestinal stromal tumor (GIST) until ultimate diagnosis at the time of surgical resection.

2. Case report

A 33-year-old male without medical, surgical or significant family history presented with a complaint of several months of vague, diffuse abdominal pain that was initially attributed to constipation and treated in the outpatient setting. The patient was referred to a surgeon after a computed tomography (CT) scan was obtained due to continued symptoms. A large soft tissue mass in the mid abdomen extending to the upper pelvis was discovered (Fig. 1). The mass, which consisted of hypodense regions and multiple vessels, measured 15.7 × 28.6 × 24.2 cm. The mass was displacing bowel loops, but was not causing obstruction or other mass effect.

In order to inform initial treatment, an ultrasound-guided core needle biopsy was performed. Histologically, neoplastic spindle cells were identified that were moderately positive on immunohistochemical staining for muscle specific actin, S-100 and CD117 and negative for cytokeratin AE1/AE3, desmin and CD34. Cytology revealed no clonal proliferation of lymphocytes.

Given the patient's clinical presentation and histopathology, there was high suspicion that the lesion represented a large GIST. The patient began neoadjuvant treatment with imatinib. After four months, the patient's pain, anorexia and weight loss did not resolve. A repeat CT scan revealed that the mass had increased significantly

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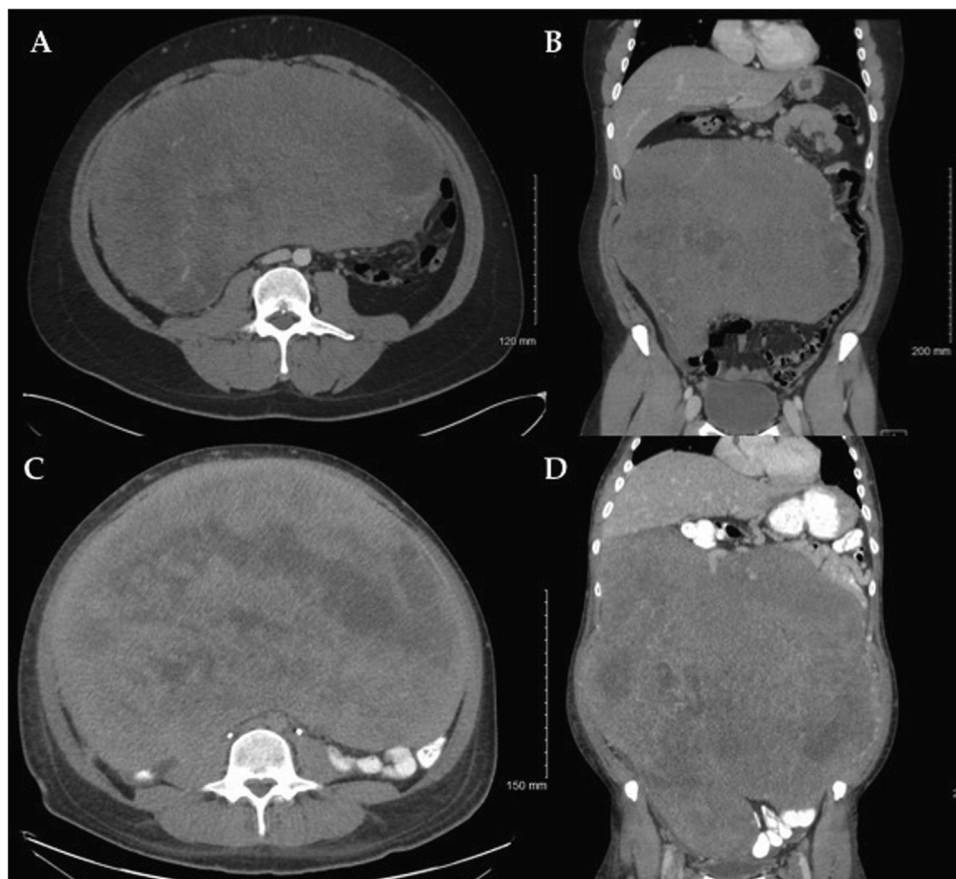


Fig. 1. Computed tomography (CT) scans. At initial presentation (A and B), the soft tissue mass was 15.7 × 28.6 × 24.2 cm. After a trial of four months of imatinib, the mass had grown to a size of 20 × 37 × 32 cm and demonstrated mass effect on surround organs (C and D).



Fig. 2. Gross images. The patient's abdominal distension improved when compared preoperatively (A) and postoperatively (B). The resection specimen (C) was 45 × 33 × 23 cm, and it is pictured here immediately after resection with a 15 cm ruler.

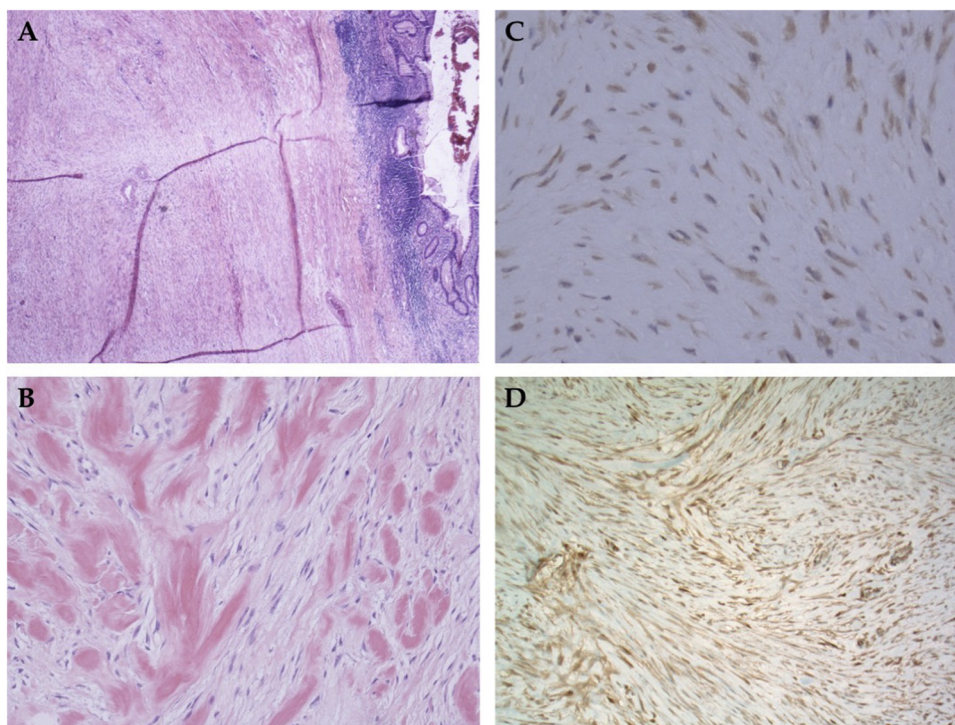


Fig. 3. Histology and immunohistochemistry of resection specimen. Hematoxylin and eosin staining of the specimen confirmed a spindle cell morphology and focal invasion of the appendix on low power (A – 2X, B – 100X). Immunohistochemistry was positive for CD117 (C – 100X) and nuclear β -catenin (D – 100X).

to greater than $20 \times 37 \times 32$ cm (Fig. 1) and was causing compression of the small bowel, ureters, and kidneys.

Due to a progression of the mass on medical therapy, the patient was taken to the operating room for resection. The patient underwent cystoscopy and ureteral stent placement followed by an exploratory laparotomy. The tumor was found to be tethered to the omentum and the bowel near the appendix; an omentectomy and ileocecectomy were performed in order to remove the tumor en bloc.

Pathologic analysis of the tumor revealed that it measured $45 \times 33 \times 23$ cm and had a mass of 16.8 kg (Fig. 2). There were negative resection margins, and with the exception of focal involvement of the appendiceal wall, there was no other involvement of the bowel. Immunohistochemistry was performed and the mass was positive for beta-catenin, weakly positive for CD117 and negative for desmin, smooth muscle actin, CD34, S100 and cytokeratin (Fig. 3). These features are consistent with a diagnosis of desmoid tumor. The patient did well post-operatively and was discharged home one week after surgery. He continues to undergo surveillance for recurrence.

3. Discussion

Intra-abdominal desmoid tumors (DTs) are rare with an incidence of 2–4 cases per million per year [1]. Like the index patient who had no family history of colon cancer and no personal history of abdominal trauma, many intra-abdominal DTs are diagnosed in asymptomatic patients on routine imaging. These lesions tend to be slow-growing and can invade structures without initial symptoms. As in this case, some patients have vague abdominal complaints that are initially dismissed until cross-sectional imaging is performed and reveals a locally advanced tumor. Because of the lack of confinement of DTs, other patients present with intestinal obstruction, organ ischemia and genitourinary complaints depending on the location into which the tumor has invaded [4,5].

This varied location and presentation often results in diagnostic difficulty as the differential diagnosis is so broad and includes more common diseases such as inflammatory lesions, liposarcoma, fibrosarcoma, and GISTs [4]. Histology and immunohistochemistry on biopsy specimens is helpful in developing a treatment strategy since these diseases differ greatly in their initial approaches to management. Relating to this case, both GISTs and DTs are recognizable in their proliferation of spindle cells [6] and CD117 can be found in both tumors [7]. Beta-catenin expression, however has been shown to aide in the distinction since it is found in both the nucleus and cytoplasm of DT cells but only the cytoplasm of GIST cells.

Both medical and surgical therapies have been trialed for the treatment of DTs, though large randomized studies are not abundant due to the rarity and relative anatomic diversity of the tumors. What is clear is that despite the treatment modality, the recurrence rate for DTs is 30% to 40% [8]. Watchful waiting has been advocated for asymptomatic patients with interval cross-sectional imaging, and has been shown to be equivalent to medical therapy in progression-free survival [9]. For symptomatic patients, however, surgery is the mainstay of treatment. Though survival is not improved by surgical resection, and morbidities such as short gut syndrome can accompany resection of large or multiple tumors [3,10], a primary resection with negative margins is the most effective treatment [11]. Radiation and systemic drug therapy, such as tamoxifen, doxorubicin, and anti-inflammatory drugs, create the arsenal for medical therapy both in the neoadjuvant setting and cases of recurrence [1,12].

A high level of clinical suspicion is required in the diagnosis of desmoid tumors. Surgery has certainly been shown to be an important treatment modality for DTs regardless of the medical adjuncts that are used. Close follow up is also essential given the high recurrence rate despite effective treatment.

Conflicts of interest

None.

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Ethical approval

Exempt from IRB.

Consent

Written consent obtained from the patient.

Author contribution

Williams–writing the manuscript and literature review. Hightchew–pathological diagnosis. Siripurapu–primary surgeon.

Guarantor

Austin D. Williams.

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