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Multivisceral Resection for Massive Retroperitoneal Fibromatosis in an Adolescent: A Rare Surgical Challenge

Anwaar M. Bisri [1], Haneen M. Banjar [1], Zahrah Barnawi [1], Rawan Mohammed [2], Ehab M. Sadek [3]

1. General Surgery, King Abdullah Medical City, Makkah, SAU 2. General Surgery, King Abdulaziz Hospital, Makkah, SAU 3. Surgical Oncology, King Abdullah Medical City, Makkah, SAU

Corresponding author: Anwaar M. Bisri, bisrianwar@outlook.com

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Abstract

Desmoid tumors (DTs), also referred to as retroperitoneal fibromatosis, are fibroblastic neoplasms characterized by local aggressiveness. Although these tumors lack metastatic potential, their propensity for invasion and recurrence creates substantial clinical challenges. This case involves an 18-year-old female who presented with a one-year history of left-sided abdominal pain, progressive swelling, weight loss of 7 kg, and amenorrhea. CT imaging revealed a large epigastric mass measuring 18.4 × 19.1 × 11.9 cm, with a significant mass effect on surrounding structures. The patient underwent en bloc surgical resection, including left nephrectomy, distal pancreatectomy, splenectomy, and partial gastrectomy. Histopathological examination confirmed the diagnosis of fibromatosis, with tumor cells positive for vimentin and β-catenin. This case underscores the complexity of diagnosing and managing intra-abdominal DTs in young patients. Such tumors are rare and often present with nonspecific symptoms, complicating timely diagnosis. Surgical excision remains a primary therapeutic approach; however, cases involving multiple organ systems pose considerable challenges in surgical management. Importantly, this case highlights the necessity of including DTs in the differential diagnosis of abdominal masses in young adults, even in the absence of trauma or a family history of adenomatous polyposis. Intra-abdominal desmoids are well described, but giant retroperitoneal fibromatosis of this size (18.4 × 19.1 × 11.9 cm) in an adolescent female requiring complex multivisceral en bloc resection is exceedingly rare. This case illustrates that radical surgical clearance is feasible with acceptable morbidity and emphasizes the necessity of a multidisciplinary surgical approach to managing giant pediatric/adolescent retroperitoneal tumors.

Categories: General Surgery
Keywords: adenomatous polyposis, desmoid tumor, fibromatosis, histopathology, nephrectomy, pancreatic tumor

Introduction

Desmoid-type fibromatosis (DF) represents a rare subset of mesenchymal neoplasms characterized by benign yet locally aggressive fibrous tissue proliferation [1]. The term “desmoid” was initially introduced by Mueller in 1838 to describe the uniform appearance of the myofibroblastic cells with minimal atypia that constitute desmoid tumors (DTs). Clinically, DF is exceedingly rare, accounting for approximately 0.03% of all tumors and about 3% of soft tissue sarcomas, with an incidence of only five to six cases per million individuals annually [2-4]. DTs are well known for their slow growth and high recurrence rates, which can reach 87% [5]. Approximately 64% of DT cases occur in extra-abdominal locations, with 14% involving the upper extremities and 16% involving the lower extremities. Additionally, 16% arise in the abdominal wall, while 20% are located intra-abdominally [6]. A small proportion of cases are associated with mutations in the familial adenomatous polyposis (FAP) gene [7]. There was no family history of colorectal polyposis or clinical evidence of Gardner syndrome; however, formal APC germline genetic testing and screening colonoscopy were not performed. Therefore, FAP was clinically unsuspected rather than definitively excluded. DTs associated with FAP account for approximately 2-15% of all desmoid cases and are most commonly found in the mesentery of the small intestine [8]. Although DTs do not metastasize, their propensity for local recurrence is significant, with rates approaching 50% within five years [9].

Anatomically, these tumors commonly arise in the extremities and thoracic wall. In patients with FAP, intra-abdominal or abdominal wall involvement is also commonly observed [9,10]. DTs are significantly associated with estrogen levels, which may explain the increased incidence of both extra-abdominal and abdominal DTs in women during pregnancy and the postpartum period [11]. Mutations in the adenomatous polyposis coli and catenin beta-1 genes can account for up to 89% of DTs [12]. The biological behavior of DF is heterogeneous, ranging from indolent, slowly enlarging masses to rapidly progressive, aggressive lesions. The clinical course involves an initial phase of tumor growth, which may transition into a period of stabilization or even spontaneous regression [13]. Presenting symptoms are variable and often depend on tumor location but may include muscle pain, restricted range of motion, fatigue, nausea, malaise, and, in some cases, scarring [14].

For retroperitoneal DF (RPDF), medical therapy must be the first-line treatment, according to the guidelines

published by the European Desmoid Working Group [15]. Active surveillance and conservative or medical management are established first-line approaches for DF. Radical multivisceral surgery in our case was justified by the massive tumor dimensions, rapid progression, severe symptomatic mass effect, and immediate threat to adjacent vital visceral structures. Radiological examinations, such as ultrasound, CT, and MRI, play important roles in the diagnosis, management, and monitoring of fibromatosis [16]. Ultrasound is the first-line imaging tool, particularly for extremity or abdominal wall tumors, and can demonstrate distinctive "tail" and "staghorn" signs [10,17]. CT scans are the investigation of choice for diagnosing and monitoring intra-abdominal desmoids, whereas MRI is preferred for the follow-up of extra-abdominal and pelvic DTs [5]. Microscopically, DTs resemble scar tissue. Immunohistochemically, they are positive for β-catenin, vimentin, cyclooxygenase-2, c-KIT, androgen receptors, and beta-estrogen receptors [10,18].

Factors associated with an increased risk of DT recurrence following surgery include younger age (under 30 years), tumor location in the extremities, larger tumor size (greater than 5 cm), positive surgical margins, and a history of trauma at the tumor site. Tumors located in the extremities carry the highest recurrence risk, ranging from 30% to 90%. However, the addition of postoperative radiotherapy has been associated with lower recurrence rates, reported to range from 14% to 38% [6]. Evidence-based guidelines for DF are limited because of the rarity of the disease and the wide variation in its natural history. With local control rates as high as 80% at five years, many researchers have historically supported early surgery. Recent long-term studies have shown that many DFs remain stable or even regress during observation in 20-30% of cases [3,19,20]. Therefore, this report presents the clinical presentation, diagnostic assessment, surgical management, and outcomes of a rare and complex case of RPDF requiring multivisceral resection in an 18-year-old female, illustrating the complexities and challenges of managing such aggressive lesions in young adult patients.

Case Presentation

An 18-year-old woman was referred to our hospital from another city, with a notable clinical and surgical history. She reported experiencing abrupt, sporadic, stabbing pain localized to the left side of her abdomen for approximately one year. The pain was nonradiating and lacked clear precipitating or relieving factors. Associated symptoms included progressive swelling at the same site, as well as episodes of nausea and vomiting. The patient denied any changes in bowel movements or urinary symptoms. Secondary amenorrhea was clinically attributed to functional hypothalamic suppression secondary to severe weight loss (7 kg), systemic cachexia, and physical stress due to chronic illness. Preoperative hormonal assessment was not performed because the presentation was considered most consistent with functional disruption due to systemic catabolism and chronic disease burden.

On examination, the patient appeared alert, oriented, and cachectic. Her vital signs were as follows: blood pressure, 117/73 mmHg; pulse, 123 beats per minute; respiratory rate, 18 breaths per minute; temperature, 36°C; and oxygen saturation, 98% on room air. Respiratory examination revealed equal breath sounds bilaterally.

Gastrointestinal examination showed a distended abdomen with a large, firm, nontender mass occupying the epigastrium and left hypochondrium and measuring approximately 15 × 15 cm. The mass had limited mobility, with tenderness noted in the right lower quadrant. Percussion revealed dullness without a fluid thrill, and the pain score was 5 out of 10. Laboratory investigations are listed in Table *1*.

Laboratory parameter (unit)	Patient value	Reference range
Hemoglobin (g/dL)	9.4	13.5-17.5
White blood cell count (10^9/L)	5.1	3.9-11
Platelet count (10^9/L)	445	150-400
Albumin (g/dL)	3.29	3.5-5.2
Calcium (mg/dL)	8.36	8.4-10.2
Sodium (mmol/L)	143	136-145
Chloride (mmol/L)	108	98-106
Potassium (mmol/L)	4.2	3.5-5.1
Magnesium (mg/dL)	2.02	1.6-2.6
Alkaline phosphatase (U/L)	42	40-150
Aspartate aminotransferase (U/L)	19	8-48
Alanine aminotransferase (U/L)	11	12-78
Blood urea nitrogen (mg/dL)	8.75	8-24
Creatinine (mg/dL)	0.439	0.74-1.35

TABLE 1: Laboratory test results.

A CT scan of the abdomen and pelvis identified a massive, heterogeneously enhancing retroperitoneal soft tissue mass in the left hypochondrium, measuring up to 18.4 × 19.1 × 11.9 cm and causing significant mass effect, pancreatic displacement, and splenic compression, with mild-to-moderate free fluid (Figure *1*). Although preoperative imaging showed no gross vascular or direct organ infiltration, intraoperatively, the mass was found to be densely adherent to and encasing adjacent vital structures, making blunt dissection unsafe and necessitating en bloc multivisceral resection. The mild-to-moderate free pelvic fluid observed on the initial CT scan was interpreted as reactive or physiological fluid associated with mass effect and localized inflammation. Cytological sampling was not performed because malignant ascites was not suspected. Follow-up imaging confirmed complete resolution of the pelvic fluid postoperatively.

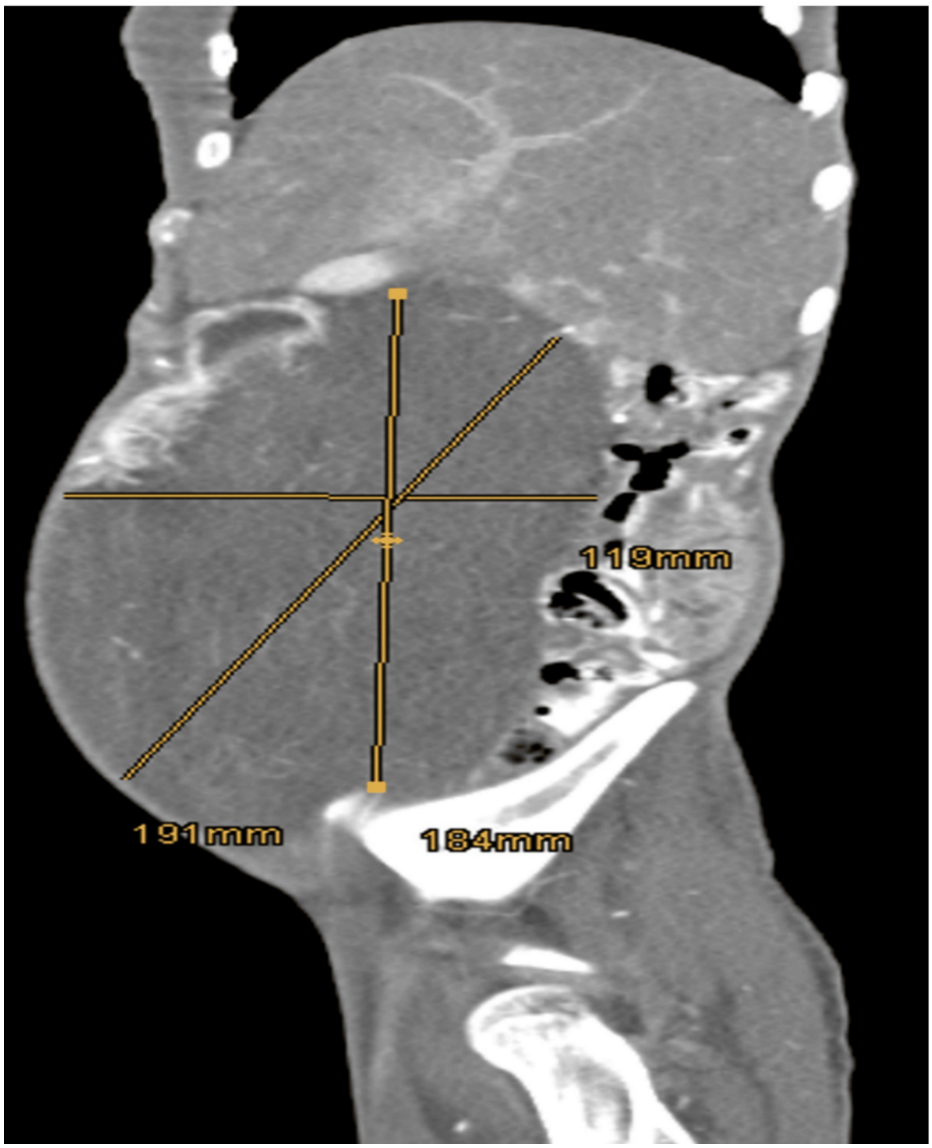


FIGURE 1: CT scan sagittal view of the tumor showing a large, fairly well-defined soft tissue mass in the epigastrium, measuring approximately 18.4 × 19.1 × 11.9 cm.

Histopathological examination of the core biopsy specimens showed collagenous proliferation of spindle cells without atypia, necrosis, or mitoses. A broad immunohistochemical panel was used to establish diagnostic confidence and systematically exclude possible histological mimics. Tumor cells were positive for β-catenin in a strong nuclear pattern and vimentin in a diffuse cytoplasmic pattern. Notably, neoplastic cells were negative for CD117 (c-KIT) and DOG1, excluding gastrointestinal stromal tumor (GIST). Furthermore, S100 negativity excluded a nerve sheath origin (e.g., schwannoma or neurofibroma), and negative immunostaining for desmin and smooth muscle actin (SMA) favored a non-smooth muscle neoplasm over leiomyoma or leiomyosarcoma. This immunophenotypic profile supported the diagnosis of DF. The status of the surgical margins was established separately by postoperative histopathological examination.

The patient underwent an exploratory laparotomy with en bloc resection of the retroperitoneal abdominal mass, measuring 18.4 × 19.1 × 11.9 cm and with an estimated specimen weight of approximately 6.5 kg. The procedure included left nephrectomy, distal pancreatectomy, splenectomy, partial gastrectomy, and placement of a feeding jejunostomy tube. Intraoperative findings included a large abdominal mass inseparable from the distal pancreas, left renal hilum, posterior wall of the stomach, and spleen (Figures *2*, *3*, Video *1*). The total operative time was eight hours, with an estimated intraoperative blood loss of approximately 700-800 mL.

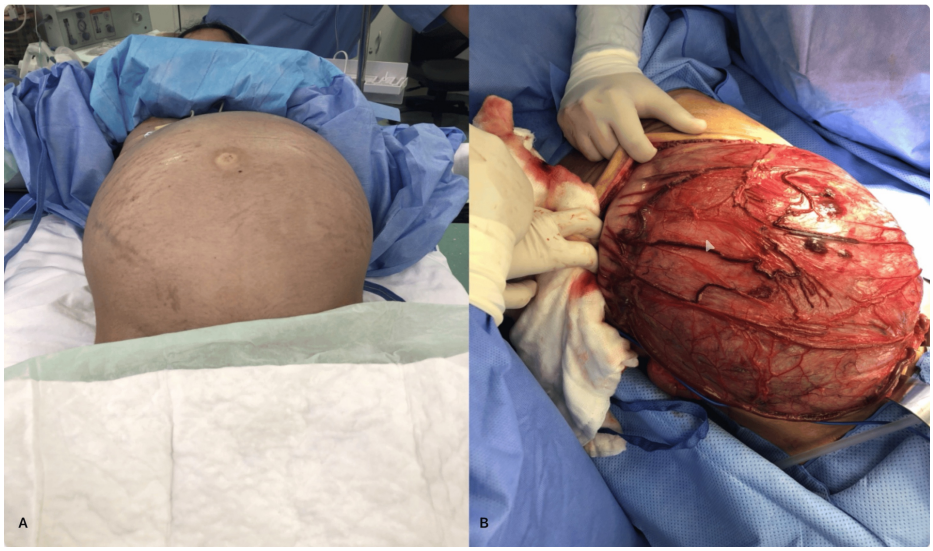


FIGURE 2: Preoperative clinical examination and initial surgical exposure.

(A) Preoperative clinical view displaying marked, asymmetrical abdominal distension. (B) Intraoperative exposure following midline laparotomy, demonstrating a massive, well-circumscribed retroperitoneal tumor protruding into the abdominal cavity.

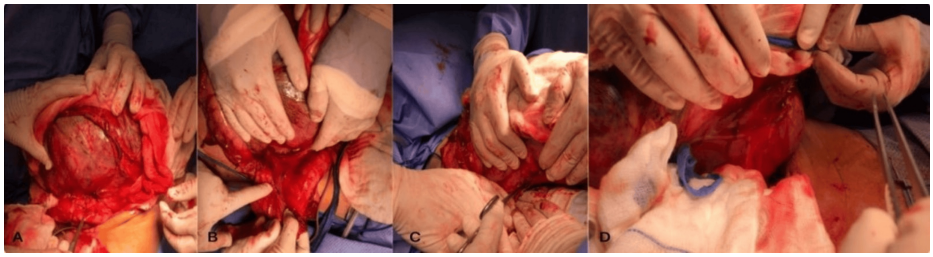


FIGURE 3: Intraoperative findings during radical en bloc resection.

(A) Surgical exposure of the retroperitoneal mass and transverse colon. (B) Dissection along the middle colic vessels. (C) Mobilization of the mass near the pancreatic body and major retroperitoneal vascular structures. (D) Dissection and control along the inferior mesenteric vessels.

VIDEO 1: Intraoperative footage demonstrating the excised retroperitoneal abdominal mass and the associated resections, including left nephrectomy, distal pancreatectomy, splenectomy, and partial gastrectomy.

View video here: https://vimeo.com/1225961549?share=copy&fl=sv&fe=ci

Following the procedure, the patient was transferred to the intensive care unit, where she was closely monitored and managed for four days before being transferred to the surgical ward in stable condition. R0 resection was confirmed postoperatively by histopathological examination, which verified that the surgical margins of the stomach, pancreas, and left ureter were completely free of tumor involvement. The spleen demonstrated focal infarction without direct tumor infiltration (Figure *4*). Mandatory post-splenectomy immunizations against encapsulated organisms, including *Streptococcus pneumoniae*, *Neisseria meningitidis*, and *Haemophilus influenzae* type b, were administered, along with counseling regarding long-term infection risks, in accordance with post-splenectomy prophylactic protocols.

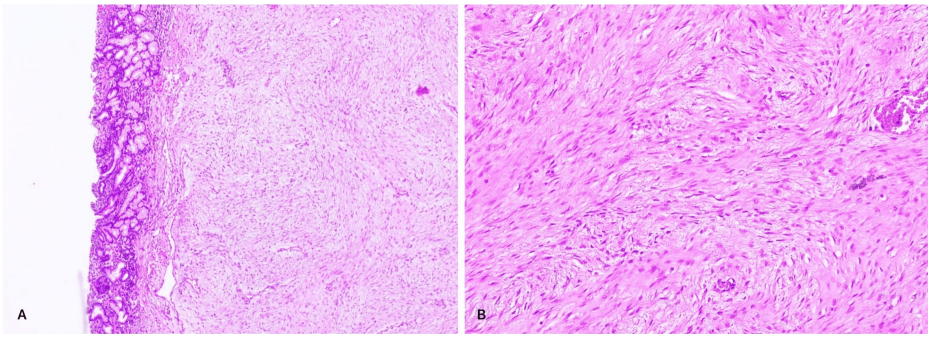


FIGURE 4: Postoperative histopathological findings of the resected mass.

(A) Low-power view demonstrating classical tissue architecture, tumor margins, and spindle cell proliferation (H&E, ×10 magnification). (B) High-power view displaying uniform, elongated spindle-shaped cells without significant nuclear atypia or atypical mitotic figures (H&E, ×40 magnification).

Despite the extensive multivisceral resection (partial gastrectomy, distal pancreatectomy, left nephrectomy, and splenectomy), the patient demonstrated excellent functional recovery without long-term metabolic complications, exocrine or endocrine pancreatic insufficiency, renal impairment, or severe nutritional deficits during follow-up.

Discussion

DF, also known as aggressive fibromatosis, is a rare mesenchymal neoplasm characterized by locally infiltrative growth and no metastatic potential [1]. Intra-abdominal DTs account for approximately 20% of all DTs and represent a major diagnostic challenge because of their nonspecific presentation [6]. These tumors are most common in women of reproductive age (20-40 years) and are often associated with high estrogen levels, trauma, or FAP, but they are rarely seen in adolescents without known risk factors [11].

In our case, an 18-year-old woman presented with a huge retroperitoneal mass, progressive weight loss, and amenorrhea, and an intra-abdominal malignancy was initially suspected. Up to 25% of DTs are associated with a history of trauma, and 68-86% of intra-abdominal DTs in patients with FAP are postsurgical [11]. However, our patient had no previous surgical history or clinical features suggestive of FAP. This emphasizes the need for a high index of suspicion for DF in young patients presenting with rapidly enlarging abdominal masses.

Aggressive fibromatosis of abdominal origin sometimes mimics GISTs, as shown in this case report and demonstrated by Abu-Jeyyab et al. in their study [16]. In their case, a middle-aged female presented with abdominal pain and a mass in the left hemiabdomen, which was confirmed by abdominal CT and ultrasound; however, there was no history of weight loss [16]. CT is a commonly used diagnostic modality for intra-abdominal DTs. This modality provides sufficient details of tumor localization and preoperative assessment to identify the relationship between the tumor and nearby structures and organs. CT imaging of abdominal fibromatosis typically shows uniform density [16]. However, necrosis and calcification may indicate soft tissue malignancy or DTs of the extremities.

In the present case, CT of the abdominal mass showed central necrosis, a rare finding that made the diagnosis more challenging. Imaging findings were inconclusive, leading to the decision to perform a core biopsy, which was required to confirm the diagnosis. Histopathological examination revealed collagenous spindle cell proliferation without atypia or necrosis. Immunohistochemistry was positive for vimentin and β-catenin (focal) but negative for CD117, CD34, S100, SMA, and desmin. These findings are consistent with the criteria described in the study by Ganeshan et al. [17].

The management of fibromatosis involves various approaches. Complete surgical resection with free margins has historically been considered important for managing aggressive fibromatosis and achieving favorable outcomes [17]. Penel et al. observed positive outcomes in patients with abdominal wall or intra-abdominal DTs who underwent complete (R0) surgical resection [2]. After a median follow-up of 25.7 months, median recurrence-free survival had not been reached. Although intra-abdominal DTs accounted for only 5.8% of the study population, these patients had fewer complications than those who underwent resection of extra-abdominal DTs [8]. Giant intra-abdominal DTs present major technical challenges for surgical intervention because of altered vascular anatomy and extensive local invasion into surrounding organs. In the present case, an exploratory laparotomy was performed, revealing a large mass involving the pancreas, kidney, stomach, and spleen. Complete macroscopic clearance required multivisceral resection (left nephrectomy, distal pancreatectomy, splenectomy, and partial gastrectomy) to achieve negative

surgical margins (R0), which are important because incomplete excision may contribute to local recurrence.

Similarly, Park et al. presented a case of a 23-year-old female with a 10-cm abdominal mass involving the distal pancreas and invading the posterior wall of the stomach and small intestine. Surgical resection with distal pancreatectomy, splenectomy, and partial gastrectomy was performed, with no recurrence during three years of follow-up [18]. The pancreatic region involved and the patient's age are similar to those in our case. Although the age of patients with DTs involving the pancreas varies, the most common site is the distal pancreas. Park et al. presented a table in their study showing that nine of 14 patients had DTs involving the pancreatic tail [18].

Sporadic intra-abdominal DF is rare, accounting for only 5% of cases. To date, fewer than 100 cases have been reported since the 1960s. Among these, tumors involving the pancreas are rare, with only 13 cases reported [18]. In the present case, the kidney was also involved and was removed during surgery along with the other affected organs.

The present case highlights the diagnostic and therapeutic challenges of intra-abdominal DTs, particularly in younger patients, in whom these tumors are uncommon. Diagnosis is often delayed because of the rarity of these tumors and their nonspecific presentation. DTs should be included in the differential diagnosis of abdominal masses, even in the absence of trauma or a history of FAP. Surgical resection remains an option for selected patients, but multiple organ involvement, as in our patient, complicates management because of the need to resect affected organs. Further research should focus on the genetic and molecular characteristics of these tumors involving multiple organs and assess their long-term treatment outcomes.

This case provides important clinical insights into the management of complex retroperitoneal masses in young patients. First, intra-abdominal fibromatosis should be considered in the differential diagnosis of adolescents presenting with rapidly growing abdominal masses, even in the absence of a history of FAP. Moreover, when there is extensive contiguous organ involvement, en bloc multivisceral resection may be indicated to achieve R0 resection and potentially reduce the risk of local recurrence. Finally, a definitive diagnosis requires detailed histopathological and immunohistochemical evaluation, particularly assessment of nuclear β-catenin expression and vimentin positivity.

Conclusions

This case report highlights the challenges of diagnosing retroperitoneal fibromatosis, particularly in young patients without risk factors such as trauma or FAP. The patient's nonspecific symptoms and weight loss highlight the difficulty of diagnosing this rare condition. Advanced imaging and histopathological examination were important in establishing the diagnosis. The successful resection highlights the importance of multidisciplinary management in providing optimal care. This case also emphasizes the need to include DTs in the differential diagnosis of abdominal masses. Further investigation of the genetic and molecular mechanisms underlying DTs and the evaluation of new therapies may help improve patient outcomes.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Anwaar M. Bisri, Rawan Mohammed, Zahrah Barnawi, Haneen M. Banjar

Drafting of the manuscript: Anwaar M. Bisri, Rawan Mohammed, Zahrah Barnawi, Haneen M. Banjar

Acquisition, analysis, or interpretation of data: Rawan Mohammed, Zahrah Barnawi, Ehab M. Sadek, Haneen M. Banjar

Critical review of the manuscript for important intellectual content: Ehab M. Sadek

Supervision: Ehab M. Sadek

Disclosures

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submitted work.

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