International Journal of Surgery Research
www.surgeryjournal.in
Online ISSN: 2664-6056, Print ISSN: 2664-6048
Received: 20-07-2022; Accepted: 08-08-2022; Published: 23-08-2022
Volume 4, Issue 2, 2022, Page No. 3-8

The hidden tumour: Delayed presentation of desmoid

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Abstract
Desmoid tumours are uncommon benign tumours that arise from the musculoaponeurotic layers. They're also known as aggressive fibromatosis, and they make up roughly 3% of all soft tissue tumours and 0.3% of all neoplasms. They have no metastatic potential, yet they have a strong proclivity to invade and recur locally. Patients between the ages of 30 and 40 are most typically diagnosed, with a female preponderance. 75-80% of cases are spontaneous, whereas the remainder are linked to familial adenomatous polyposis (FAP). Herewith, we have presented a case report of 37-year-old female patient presented to the surgical oncology outpatient department with a 15-month history of pain in the right side of her lower abdomen, followed by 8-month complaints of swelling in the same area. A lump was felt in the right paramedian region on clinical examination, which shrank in size when the patient raised his legs straight.

Keywords: hidden tumour, musculoaponeurotic layers, desmoid

Introduction

Desmoid tumors (DT) are aggressive fibromatosis that arise from muscle aponeurosis and the fascia that surround them. They are locally invasive and have a high tendency to reoccur, despite their benign nature. It is account for 3 percent of all soft tissue tumors and 0.03 percent of all cancers. This type of tumour is extremely rare, with only 3.7 percent of a million people diagnosed each year [1]. Based on location, it is categorized as extra-abdominal, accounting for 58 percent (posterior trunk, chest wall, lower extremities), abdominal wall 37 percent, or intra-abdominal 5 percent [2]. Females of reproductive age, pregnancy, trauma, previous surgeries, use of oral contraceptives pills (OCP), and hereditary (familial adenomatous polyposis and Gardner's syndrome) are all associated with a higher risk of DT. Although they have no metastatic potential, their local infiltrative nature can induce compression of neighbouring structures. Their anatomical location restrictions surgical access and can be fatal [1,2]. Its earliest symptoms are a bulging bulk, pain in the affected region, cramping and nausea and a loss of function.

Case presentation

A 37-year-old female patient presented to the surgical oncology OPD with symptoms of abdomen pain including the infra umbilical and right para median regions for the past 15 months. The pain was gradual in start, non-radiating, and nonspecific at first, then steadily grew in intensity. The pain worsens when bending forward to sit or stand up, and it was not associated to menstruation.

After 7 months of onset, she found a lump in the same area. She consulted a local physician, who advised an USG which reported suspected endometriosis and she was treated with hormonal supplements for two months. She began a native medication after two months of hormonal therapy had failed to improve her symptoms. Her symptoms continued to worsen, and the lump gradually increased in size. She describes a prolonged second stage of labour during her first pregnancy, which occurred nine years ago. As a result, she was advised to have lower segment caesarean section (LSCS) plus tubectomy for her second childbirth four years ago. She has no history of abdominal injuries, weight loss, or appetite loss. There is no history of skin coloration or oedema. There is no history of familial adenomatous polyposis (FAP) or colon cancer in the family. There are no other co-morbidities in this patient.

On physical examination, no oedema was visible. In the right paramedian region, a 3 x 2 cm lump was palpated 2-3 cm above the pubic symphysis. When straight leg lifting was performed, it had a hard consistency and shrank in size. USG scan revealed a well-defined 3 x 2.8 x 2.2 cm oval heterogeneously hyperechoic lesion

within the anterior abdominal wall's intramuscular region with no evidence of vascularity. The entire specimen was processed following a USG-guided biopsy.

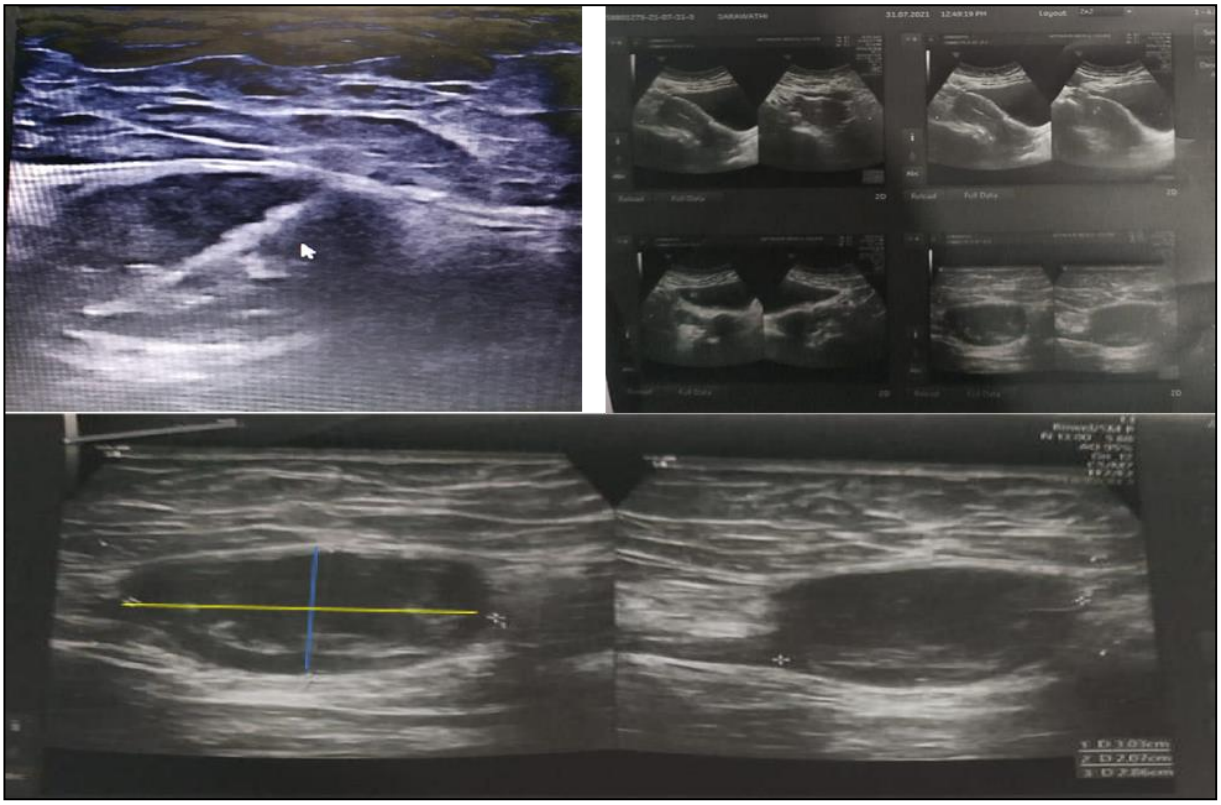

Fig 1: An USG guided biopsy was done. Arrow shows the biopsy needle. Shows an oval heterogenous hyperechoic lesion within the intramuscular region with no signs of vascularity and the tumour dimensions (3x2.8cms)

The section revealed spindle cells with pleomorphic nuclei and mildly eosinophilic cytoplasm under microscopy. Stroma revealed myxoid alterations with regions of bleeding. The histopathology result indicated a spindle cell tumour, most likely desmoid.

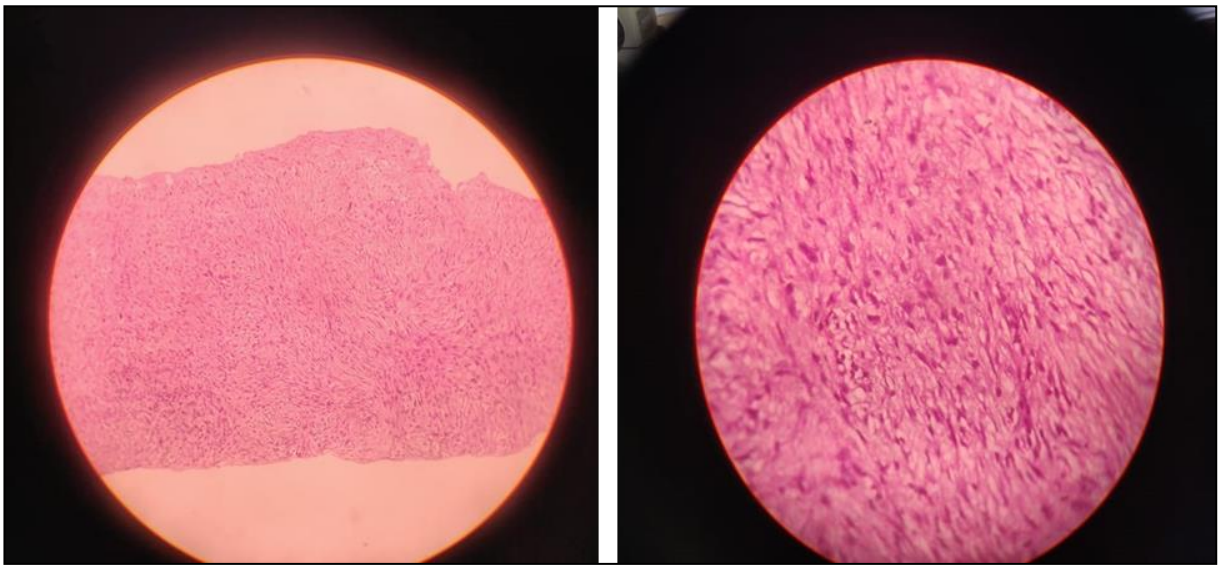

Fig 2: Low magnification 10x: Showed characteristics spindle shaped cells. High magnification 40x: Shows Pleomorphic nuclei with eosinophilic cytoplasm. Myxoid changes and haemorrhages were visualised in stroma

Within the right rectus abdominis, an MRI revealed a solitary well-defined T1 isointense and T2 hyperintense soft tissue lesion measuring 3 x 2.2 x 1.9 CMS. Intravenous contrast medium treatment resulted in uniform enhancement with no peritoneal/adjacent invasion. A benign mesenchymal tumour – desmoid / neurofibroma was found on the scan.

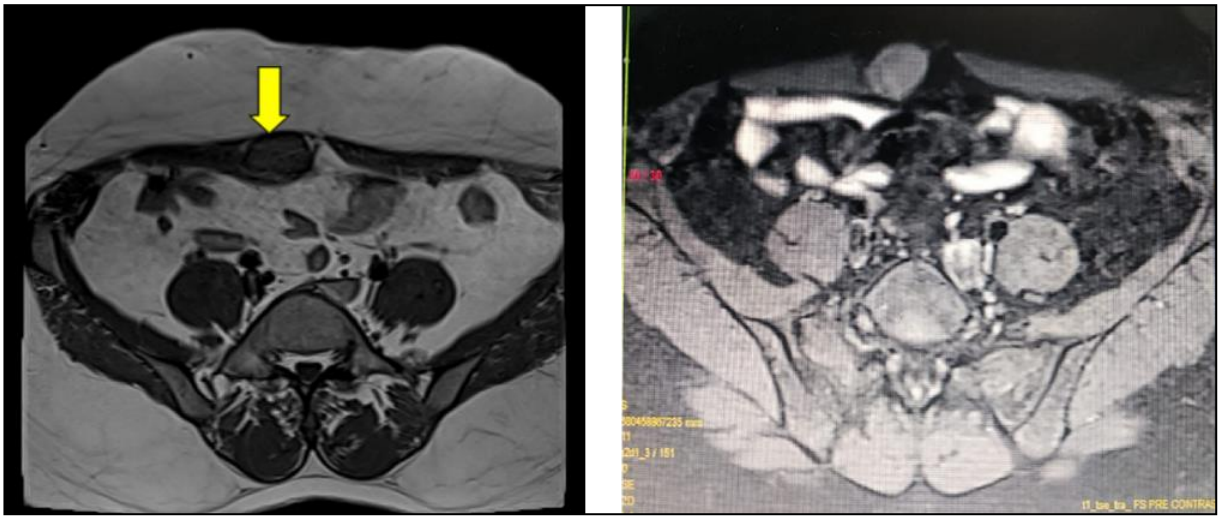

Fig 3: (a) MRI pelvis showing the tumour attached to rectus sheath (b) MRI with contrast showing well demarcated homogenous tumour

Based on these findings, patient was taken up for wide local excision of the tumour epidural anaesthesia. Wide local excision of the tumour was done with a margin of 2 cm of healthy tissue all around.

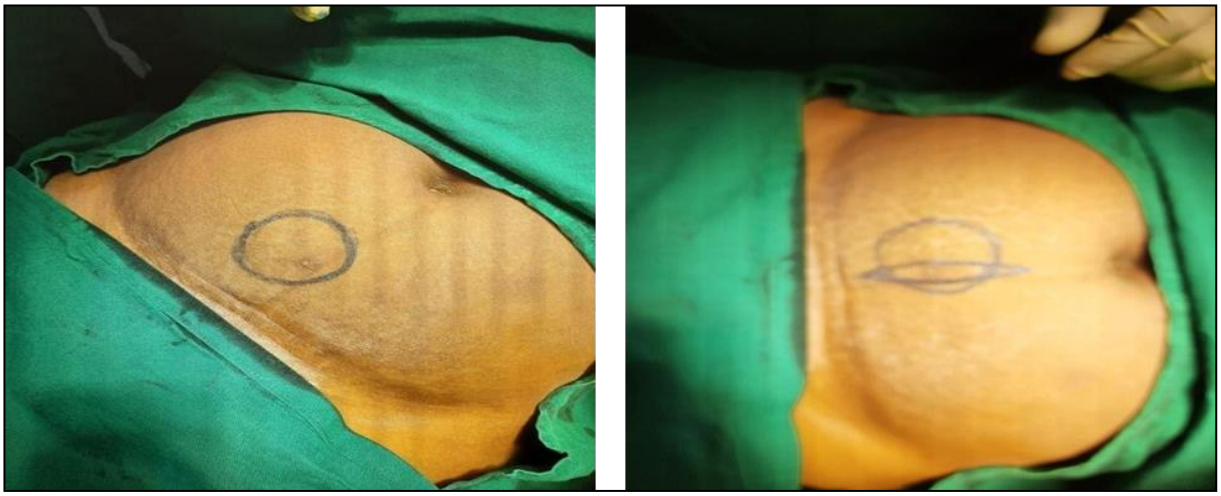

Fig 4: (a)Skin markings showing the location and extent of the swelling (b)Preoperative markings showing the skin incision to be taken surrounding the biopsy site.

Intra operatively

Tumour mass was excised in masse with skin [including the biopsy site], subcutaneous fat, rectus sheath, rectus muscle and peritoneum. The peritoneum was puckered. Anterior abdominal wall defect after resection was 7 x 6 CMS. Component separation was done. Lateral release of external oblique from the internal oblique was done up to the anterior axillary line. Rectus was approximated with loop PDS and reinforced with subcutaneous on-lay polypropylene mesh placement. Two drains were kept in the subcutaneous plane. Incision closed in layers. Patient was stable. Excised tissue was sent for histopathological examination.

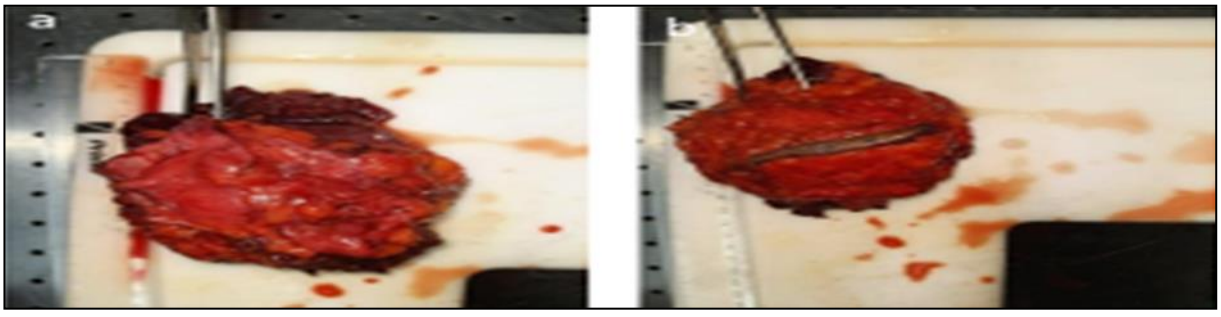

Fig 5: Shows the excised tumour with surrounding Adipose tissue and a part of excised rectus abdominisae muscle.

Histopathology

GROSS: specimen consisted of skin covered muscle tissue along with fatty tissue measuring 14 x 7 x 3 cm. External surface showed multiple grey white nodules with whorled appearance on cut surface. The tumour appeared to have raised from underlying skeletal muscle. Largest nodule measures 2.5x2x1.5cm.

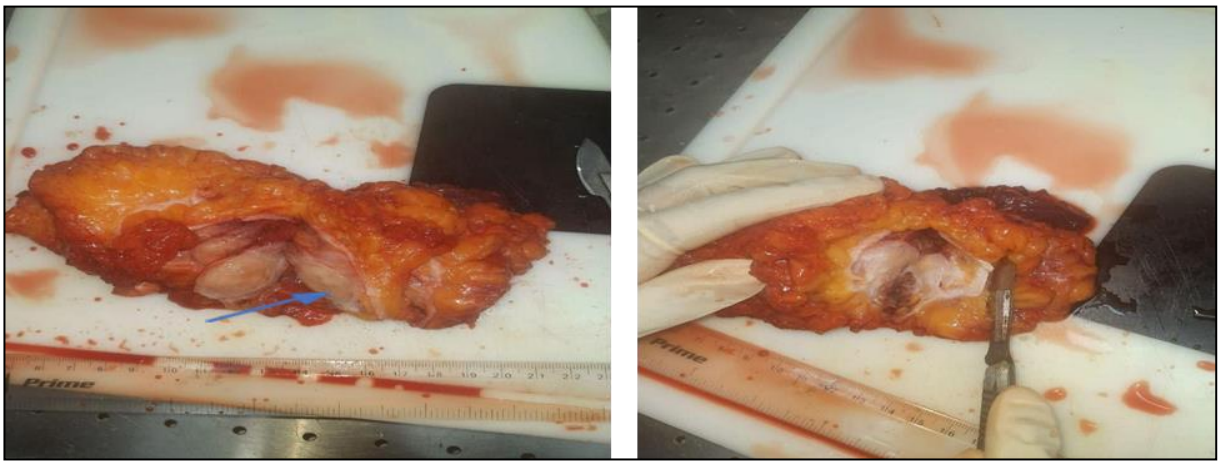

Fig 6: Gross specimen shows skin covered muscle tissue with multiple greyish white nodules

Microscopy

Sections studied showed long sweeping fascicles of elongated oval to spindle cells with pale nuclei and 1-3 nucleoli and moderate amount of eosinophilic cytoplasm set in collagenized stroma. Few congested and dilated blood vessels were seen. Occasional mitotic figures seen (<1/10HPF). The tumour is seen invading the adjacent skeletal muscle bundles and adipose tissue. All surgical margins are free of tumour. HPE report was consistent with desmoid tumour.

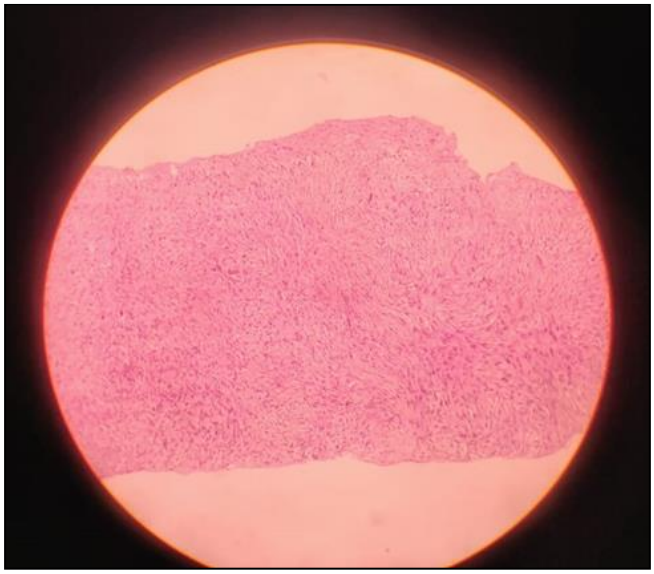

Fig 7: Shows oval-spindle cells with pale nuclei, moderate eosinophilic cytoplasm

Patient recovered well and was discharged on 4th post op day and is under follow up. Second opinion taken and was consistent with desmoid fibromatosis. Slide was sent for second opinion and findings correlated with primary pathology report. Patient is in regular follow up and has been advised colonoscopy to rule out FAP.

Discussion

Muller invented the term Desmoid, which comes from the Greek word "desmos," which meaning "tendon-like." Desmoid tumours are an uncommon type of deep fibromatosis that develops from mesenchymal stem cells with an infiltrating growth pattern. Spindle cells against a background of collagen stroma is the histological signature [3]. Myelofibroblastic cells are thought to be the source of desmoid tumours.

The majority of instances have been reported in people aged 15 to 60, with a peak incidence of 30 years. The ratio of females to males is 2:1. Oestrogen receptor positive was found in one-third of the cases, with equal sex distribution [5]. Because fibroblasts respond to oestrogen by proliferating, these tumours are typically encountered in young women during or after childbearing [6]. Hormonal and immunological variables may influence the course and severity of the condition during pregnancy [7]. An increase in volume is noticed in pre-

existing tumours on rare occasions during pregnancy. Desmoids are also infrequent after menopause [8]. Fibroblastic proliferation, which occurs after trauma and after surgery, can cause DTs if it is aggressive.

FAP-related or familial desmoids (10%) were bigger, numerous, and frequently intra-abdominal. They are linked to a mutation in the APC gene on Chromosome 5 in the germ line. The sporadic variety, on the other hand, is usually extra abdominal and more commonly encountered (90 percent) [9]. The related morbidity and mortality are due to their proclivity to invade quickly and return frequently [2].

The most common clinical manifestation is oedema, which can be accompanied by pain and weight loss. [10]

Fibrosarcoma, Liposarcoma, Rhabdomyosarcoma, Neurosarcoma, and Scar Endometriosis are the most prevalent differential diagnoses for Abdominal wall DTs [11]. Endometriosis, which is a hyper estrogenic state, is also a risk factor for DTs. The diagnosis of DTs is based on a combination of radiological and histological examinations. Initial examination with the USG is standard procedure.

On suspicion, a biopsy is performed. The desmoid tumours can be better characterised with CT and MRI scans.

There are no clear management guidelines in practise due to the low occurrence of DTs. It's best to use a multidisciplinary approach [12]. The mainstay of treatment is complete surgical excision with non-involved margins. In situations with unresectable tumours, a combination of radiation and hormone therapy has been used. The answer, however, is inconsistent [13].

DTs have a poor prognosis because to the danger of sequelae such bleeding, bowel perforation and obstruction, intestinal ischemia necrosis and gangrene, fistulisation, and urethral blockage with obstructive nephropathy [14].

The chance of recurrence is 25% in random tumours and 44% in familial tumours. The size, intra-abdominal placement, number, and young age of patients with desmoids are all characteristics that predict FAP. Despite the high probability of recurrence, the risk of death is minimal. Desmoids disease, on the other hand, is a leading cause of death among FAP patients [15].

Despite the existence of strongly suggestive risk indicators for desmoid tumour in our patient, there was a diagnostic delay. In addition, she began hormone therapy for suspected endometriosis, which exacerbated the tumour. As a result, the tumour had developed aggressively and invaded the peritoneum at the time of diagnosis, and if left untreated, it would have progressed. This emphasises the importance of considering DTs as a differential for abdominal swellings, despite their rarity and benign character.

The tumour was completely removed with tumour-free margins. With component separation and subcutaneous mesh rebuilding, the rectus sheath defect was closed and strengthened. Colonoscopy and regular follow-up have been recommended for the patient**.**

Conclusion

Desmoids tumours are exceedingly rare tumours, but their locally aggressive nature and proclivity for recurrence make it critical to consider them as a differential diagnosis in reproductive-age females with risk factors. The prognosis for these tumours is good if they are detected early and surgically resected completely. Long-term monitoring of these cases is critical for preventing complications and managing recurrence. Compliance with

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