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Case Report

Pathology Section

Retroperitoneal Desmoid Tumour Masquerading as Malignancy: A Case Report

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ABSTRACT

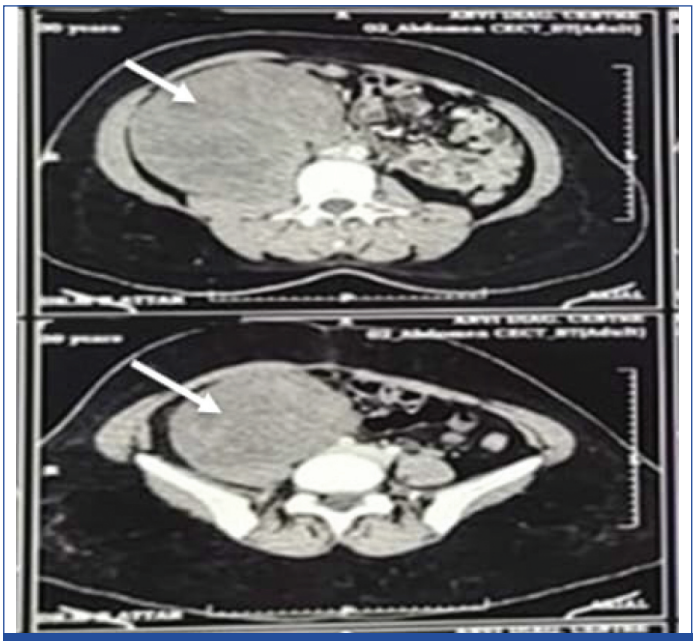
Desmoid tumour is a rare, benign soft tissue tumour. It is noted to be clinically aggressive but pathologically benign, showing a high local recurrence rate; however, it cannot metastasize. Although it is benign, it can impact functionality. The majority of these cases remain sporadic, and the aetiology is unknown. The retroperitoneum is a rare site, and there are few reports of desmoid-type fibromatosis occurring in this region. It is difficult to distinguish from other tumours and to identify the tumour's origin. Here, the authors present a case of a 36-year-old female who presented with abdominal pain and a mass in her abdomen. Imaging studies revealed a large retroperitoneal mass extending from the L5 vertebral body up to the inferior endplate of the L1 vertebral body. Exploratory laparotomy was performed, and the retroperitoneal mass was resected. Grossly, it was a capsulated dark brown to grey-coloured mass measuring 15×12×10 cm. The mass was sent for histopathology {Haematoxylin and Eosin (H&E)} study and reported as a retroperitoneal desmoid tumour, which was further confirmed on Immunohistochemistry (IHC) by showing immunopositivity for Desmin, Muscle-Specific Actin (MSA), and Beta-catenin. The patient is doing well with no further complaints to date. No disease relapse has occurred. Follow-up has been done every three months till the writing of the present report.

Keywords: Beta-catenin, Fibromatosis, Laparotomy, Sarcoma

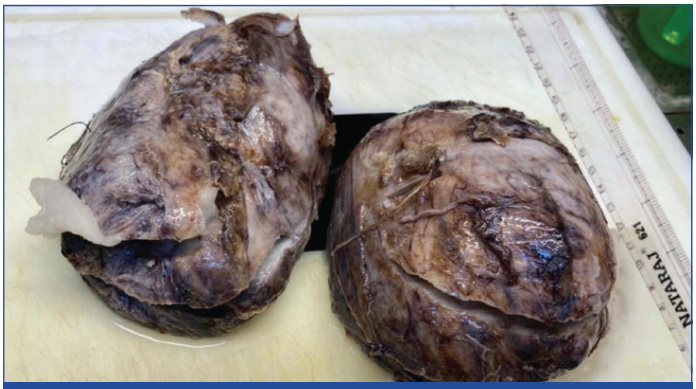
CASE REPORT

A 36-year-old female patient complained of a mass in her abdomen since two months, along with abdominal pain for the past one and a half months. The pain was insidious, dull, and aching, worsened by movement and palpation, but relieved after taking medication. The patient had undergone two Lower Segment Caesarean Sections (LSCS), one 19 years ago and the other 12 years ago. Her bladder and bowel habits, as well as her appetite, were normal. She did not have any other underlying diseases or conditions. During abdominal examination, a tender, firm mass measuring 10×10 cm was palpated in the right lumbar and iliac region. A Contrast-enhanced Computed Tomography (CECT) scan of the abdomen and pelvis revealed a large mass measuring 15×12.5×12 cm in the right retroperitoneal region, extending from the L5 vertebral body to the inferior endplate of the L1 vertebral body and abutting surrounding organs, suggesting a possibility of soft tissue sarcoma [Table/Fig-1]. There was no evidence of significant lymphadenopathy or metastatic deposits in the abdominal organs. The patient underwent exploratory laparotomy with excision of the retroperitoneal mass. The tumour was sent for histopathological study. Grossly, it was a well-circumscribed, encapsulated, globular solid mass measuring 15×12×10 cm [Table/Fig-2]. The external surface was dark brown to grey. At some places, the capsule was adherent to the tumour tissue. Cut section revealed a solid, pale white, trabeculated appearance [Table/Fig-3]. No areas of haemorrhage or necrosis were noted.

Histopathology revealed a well-encapsulated tumour tissue composed of elongated, slender, spindle-shaped cells arranged in interlacing bundles or fascicles. These cells were separated from each other by collagen and contained small, pale-staining nuclei with 1 to 3 tiny nucleoli and scant cytoplasm [Table/Fig-4]. Perivascular lymphocytic infiltrate was observed in some areas. Myxoid areas and congested blood vessels were present. A small focus of tumour tissue invasion into adjacent fibrofatty tissue was seen [Table/Fig-5]. There was no evidence of atypia, necrosis, or atypical mitoses. It was reported as a desmoid tumour, which was further confirmed by Immunohistochemistry (IHC) showing immunopositivity for Desmin



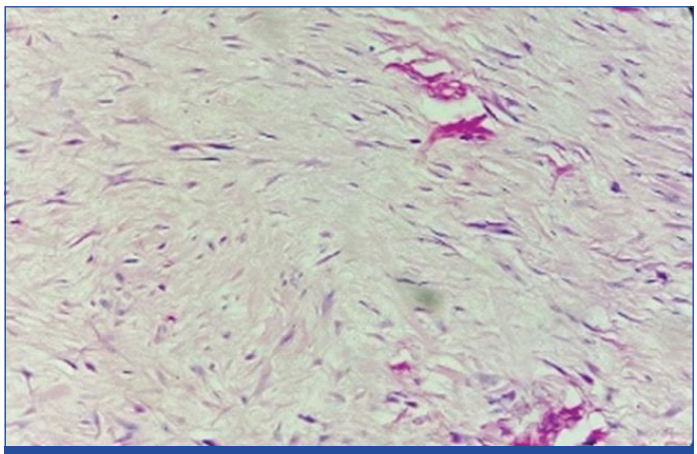
[Table/Fig-1]: Heterogenous soft-tissue density lesion seen in the abdominal cavity on right-side extending from L5 to L1 vertebral body.



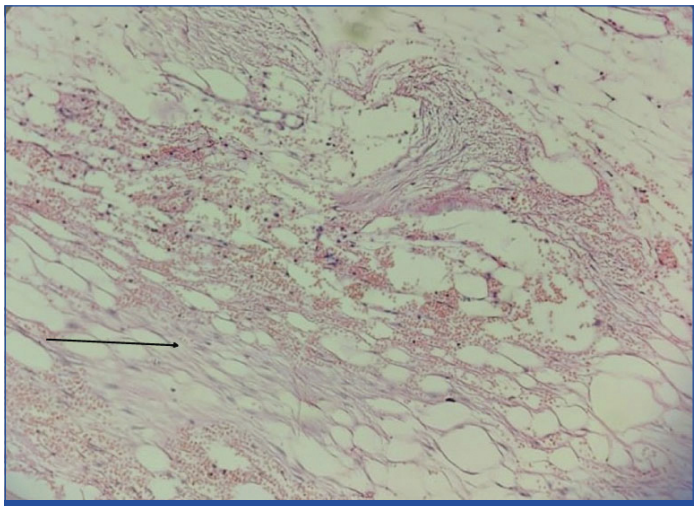
[Table/Fig-2]: External surface of tumour shows a well-encapsulated globular mass-dark brown to grey.

[Table/Fig-3]: Cut-section of tumour showing pale white, solid, trabeculated appearance.

[Table/Fig-6], MSA, and beta-catenin. It showed immunonegativity for DOG1 {Discovered on Gastrointestinal Stromal Tumour (GIST)1}, CD117, S100, Anaplastic Lymphoma Kinase (ALK), and CD34. Wide local excision of the mass was curative. The patient has not experienced any postsurgical symptoms and is recovering well.



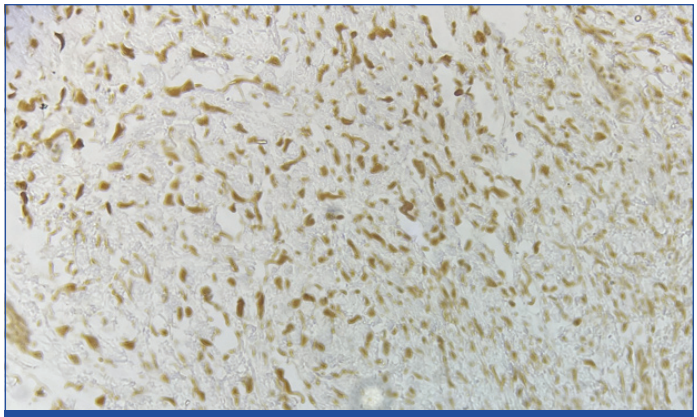
[Table/Fig-4]: Spindle shaped cells widely spaced and arranged in interlacing bundles (H&E, 400X).



[Table/Fig-5]: Tumour tissue invasion into adjacent fibrofatty tissue (H&E, 100X) (Black arrow).

DISCUSSION

Desmoid tumours account for 1.5 to 3% of all soft tissue masses and less than 1% of retroperitoneal tumours [1]. They are monoclonal, locally aggressive, fibroblastic proliferations that are often recurrent, non-metastasizing, and sometimes multifocal, presenting with variable symptoms [2]. These tumours primarily affect patients in their second and third decades of life and are more common in women [2,3]. They originate from the fascia and aponeurosis of the muscle, exhibit local infiltrating growth, and can invade adjacent structures, resulting in loss of function and causing visceral or



[Table/Fig-6]: Desmin showing diffuse and intense nuclear positivity (100x magnification, IHC marker).

neurological symptoms [4]. The majority of cases occur in extra-abdominal locations (approximately 60%), particularly in the anterior abdominal wall and shoulder girdle. Less common sites include the retroperitoneum and pelvic areas (less than 20%). Intra-abdominally, they usually arise from the mesentery, but they can also occur in other sites such as the extremities, rectus abdominis muscle, head, neck, and thorax [5].

The symptoms of desmoid tumours depend on the location of the tumour, its growth rate, and size [5]. When symptomatic, the main presentation is a painful mass with slow growth. Intra-abdominal cases are often asymptomatic but can also be associated with early satiety, nausea, bowel ischaemia, intestinal obstruction (in the case of large masses), ureteric obstruction, compression of vascular or neural structures, and bleeding or ulceration [5]. In the present case, the patient presented with an abdominal mass and abdominal pain. Similar symptoms were noted in the case of Park S; Njoku OC and Umezurike CC reported a painless abdominal mass [1,6]. Clinicians often face challenges in diagnosing these tumours correctly due to their uncommon sites, leading to misdiagnosis and confusion. A study conducted by Njoku OC and Umezurike CC reported a giant desmoid tumour mimicking a recurrent leiomyoma [6]. In that case, the patient developed an incisional hernia after 10 months, which was repaired unlike the present case where the patient is doing well and able to perform routine activities without any complications.

Many studies have reported desmoid tumours associated with Gardner's syndrome or Familial Adenomatous Polyposis (FAP). Desmoid tumours or colonic cancer are possible causes of death in FAP patients [1,4].

Desmoid tumour patients are often first assessed by ultrasound. On ultrasonography, desmoid tumours appear as a well-defined lesion with variable echogenicity [7]. However, the most common imaging modalities used are CT and Magnetic Resonance Imaging (MRI). On a CT scan, desmoid tumours may appear as a well-circumscribed solid mass with homogeneous or heterogeneous characteristics. They can be either hypointense, isointense, or hyperintense compared to the attenuation of muscles [8]. On a MRI, desmoid tumours show poor margination, low to isosignal intensity compared to muscle on T1-weighted images, and heterogeneity on T2-weighted images [8]. In present study, the mass exhibited moderate heterogenous postcontrast enhancement, whereas a study done by Imagami T et al., found mild enhancement, with weaker enhancement in the lower half of the mass [8].

Desmoid tumours often progress gradually with poorly-defined margins, an indistinct capsule, and a solitary, glistening white, round to oval mass with firm consistency [7]. On cut section, tan-white firm areas with ill-defined edges are noted. A few case reports have shown a whorled appearance on the cut section. Microscopically, the tumour consists of elongated spindle cells with small, vesicular, pale-staining nuclei, exhibiting one or two tiny nucleoli without hypochromasia. The cytoplasm is scant to moderate and eosinophilic. The background

shows a collagenous matrix. Desmoid tumours lack other malignant features [7]. The differential diagnosis of desmoid tumours on H&E sections includes pleomorphic sarcoma, fibrosarcoma, GIST, and leiomyoma. Therefore, pathologic confirmation is necessary before a definitive diagnosis can be made [1].

A fine-needle aspiration can be performed prior to surgical excision. Fine-needle aspiration smears of desmoid tumours usually demonstrate low cellularity. The tumour cells appear spindle-shaped or polygonal, exhibiting the characteristic features of fibroblastic/myofibroblastic cells with abundant basophilic cytoplasm [7]. The cytological differential diagnosis for bland spindle cells includes nodular fasciitis, scar tissue, myofibroma, superficial fibromatosis, collagenous fibroma, and fibromyxoid or spindle cell sarcoma [5].

Immunohistochemistry reveals non specific reactivity for MSA and smooth muscle actin. It is also positive for estrogen receptor beta, androgen receptor, and shows focal positivity for desmin. Additionally, more than 80% of cases exhibit beta-catenin positivity due to aberrant nuclear localisation. About 10% of these tumour cells are positive for c-KIT and cathepsin D. In the present case, desmin, MSA, and beta-catenin were positive [3]. There was a slight disagreement with the case of Ormonde M et al., where the tumour cells were positive for beta-catenin alone and negative for cytokeratin, S100, SMA, desmin, STAT6, CD34, and DOG1 [4].

The main mutation involved in sporadic desmoid tumours is in the beta-catenin-encoding gene CTNNB1 (in codons 41 or 45). As a result, nuclear beta-catenin accumulates, further activating the c-MYC or cyclin-D1 genes [2]. Mutations in the tumour suppressor gene APC, leading to Wnt signalling pathway activation, are noted in FAP-associated as well as sporadic desmoid tumours [9].

Desmoid tumour is regarded as a heterogeneous disease due to its biological behaviour. Desmoid tumours cause treatment-associated morbidity and mortality. The role of surgery is still under debate [2]. Surgical resection with a wide margin can be done for rapidly growing tumours, those suspicious of malignancy, cases with loss of organ function, or symptomatic cases. Even after complete resection, the recurrence rate is high (20-68%) [4]. Recent advances in local therapy include radiation therapy, cryoablation, and high-intensity focused ultrasound, as well as systemic therapy. Radiation therapy is effective and safe, and it is considered an alternative to surgery [5]. In cryoablation, a cryoprobe is used to continuously freeze and thaw the tumour, resulting in cell death [5]. High-intensity focused ultrasound involves the use of ultrasound energy concentrated inside the tumour results in the thermal coagulation of the targeted tissue [5]. Medical therapy includes antihormonal therapy, Non Steroidal Anti-inflammatory Drugs (NSAIDs), conventional chemotherapy, and tyrosine kinase inhibitors. Ongoing trials are exploring Wnt/beta-catenin inhibitors, gamma-secretase inhibitors, and immune checkpoint inhibitors [10].

CONCLUSION(S)

A differential diagnosis of a desmoid tumour has to be considered when evaluating a retroperitoneal mass. Since the clinical course of a desmoid tumour is uncertain, a multidisciplinary approach personalised to the individual patient is needed. It is difficult to distinguish it from other tumours and identify the tumour's origin. Therefore, a thorough clinical and histopathological work-up, along with a panel of IHC markers, is required.

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