Section: Pathology

Case Series

DESMOID FIBROMATOSIS: A CLINICOPATHOLOGICAL STUDY OF VARIABLE BIOLOGICAL BEHAVIOR

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ABSTRACT

Background: Desmoid fibromatosis is a rare fibroblastic/myofibroblastic soft-tissue neoplasm that is histologically benign but locally infiltrative, recurrent and biologically unpredictable. It may arise in the abdominal wall, intra-abdominal region, pelvis, trunk or extremities, and diagnosis requires clinicopathological correlation with immunohistochemistry, especially nuclear beta-catenin positivity. This case series highlights the variable biological behaviour of desmoid fibromatosis, ranging from indolent resectable lesions to aggressive unresectable pelvic disease. **Case Presentation:** Six cases of desmoid-type fibromatosis were evaluated. The patients included middle-aged males and females with abdominal wall or pelvic masses, some associated with previous surgery, caesarean section, hysterectomy, colorectal carcinoma treatment or family history of colorectal carcinoma. Presentations varied from incidentally detected abdominal wall swelling to abdominal pain, urinary obstruction, hematuria, hydronephrosis, bowel adherence and locally invasive pelvic disease. Radiology showed well-defined or infiltrative soft-tissue masses involving rectus muscle, bladder, bowel, ureter, prostate, mesorectal fascia or pelvic structures. Histology revealed spindle-cell proliferation in collagenous stroma with minimal atypia and absence of necrosis. Immunohistochemistry confirmed desmoid fibromatosis by strong nuclear beta-catenin positivity, with exclusion of important spindle-cell differentials. Management included surgical excision with clear margins, chemotherapy, diversion colostomy, follow-up, planned targeted therapy and referral for advanced pelvic surgery. **Conclusion:** Desmoid fibromatosis shows marked clinicopathological variability despite its non-metastatic nature. Accurate diagnosis depends on histopathology and beta-catenin immunostaining, while management must be individualized according to site, symptoms, resectability, organ involvement and biological aggressiveness.

INTRODUCTION

Desmoid fibromatosis, also known as desmoid-type fibromatosis or aggressive fibromatosis, is a rare fibroblastic or myofibroblastic soft-tissue neoplasm arising from deep connective tissues, fascia, aponeuroses and musculoaponeurotic structures.[1] It is classically regarded as histologically benign because it does not metastasize, yet its locally infiltrative growth pattern, tendency for recurrence and unpredictable clinical course make it biologically distinct from benign fibrous proliferations. These tumours may occur in the abdominal wall, intra-abdominal region, trunk, extremities, head and neck, and they may present as a painless mass or with site-specific symptoms due to compression, pain, deformity, restricted movement or involvement of adjacent organs.[2]

The clinical importance of desmoid fibromatosis lies in its variable biological behaviour. Some lesions remain indolent or stable for long periods, whereas others enlarge progressively, infiltrate surrounding structures and produce significant morbidity despite the absence of distant spread. This heterogeneity creates difficulty in predicting outcome at diagnosis.[3] The disease is seen more often in young and middle-aged adults and shows female predominance in many series, although it can occur at any age and at almost any deep soft-tissue site. It may be sporadic or associated with familial adenomatous polyposis and Gardner syndrome. Molecularly, most sporadic cases are linked to dysregulation of the Wnt/β-catenin pathway,

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commonly through CTNNB1 mutation, while syndromic cases are related to APC mutation. This molecular background supports the clonal neoplastic nature of the lesion and explains the diagnostic role of β-catenin immunohistochemistry.[2]

Pathologically, desmoid fibromatosis is characterized by long sweeping fascicles of uniform spindle cells arranged in a collagenous or variably myxoid stroma. The tumour often shows infiltrative margins with entrapment of skeletal muscle, adipose tissue or adjacent soft tissue.[4] Cytological atypia is usually minimal, mitotic activity is low and necrosis is typically absent. However, its morphology may overlap with scar, nodular fasciitis, low-grade fibromyxoid lesions, gastrointestinal stromal tumour and low-grade sarcoma, particularly in small biopsy specimens. Therefore, clinicopathological correlation is essential, including tumour site, radiological appearance, growth pattern, histomorphology and immunohistochemical findings. Immunohistochemistry is particularly useful when morphology is equivocal, with nuclear β-catenin positivity supporting the diagnosis along with variable smooth muscle actin expression and usual absence of markers that suggest neural, vascular, epithelial or gastrointestinal stromal differentiation.[5]

Management of desmoid fibromatosis has changed considerably in recent years. Earlier, wide surgical excision was commonly considered the main treatment, but high recurrence rates, functional morbidity and recognition of spontaneous stabilization or regression have shifted the approach toward individualized, risk-adapted care.[6] Active surveillance is now accepted as an initial strategy for many asymptomatic or non-progressive tumours, while surgery, radiotherapy, systemic therapy or targeted therapy may be selected for progressive, symptomatic or anatomically threatening disease. Thus, pathological diagnosis alone is not sufficient; assessment of biological behaviour and clinical context is equally important, because the same histological diagnosis may correspond to different clinical outcomes.[7]

A clinicopathological study of desmoid fibromatosis is therefore valuable because it allows systematic evaluation of age and sex distribution, anatomical location, clinical presentation, tumour size, microscopic patterns, immunohistochemical profile, treatment modality and follow-up outcome. Such analysis helps identify the spectrum of behaviour ranging from indolent lesions to aggressive locally recurrent tumours. It also assists in distinguishing desmoid fibromatosis from histological mimics and supports appropriate multidisciplinary decision-making. Since the tumour is uncommon and biologically unpredictable, documentation of clinicopathological features in a defined patient population can contribute to better understanding of its natural history, improve diagnostic accuracy and guide patient-specific management.

CASE DESCRIPTION

Case 1: A 44-year-old male, previously operated for colorectal carcinoma with clear resection margins followed by chemotherapy and radiotherapy, presented after 8 years with intermittent urinary obstruction, later progressing to hematuria, urinary retention and obstructive symptoms for 3 months. USG and CT showed a pelvic/rectal mass suspicious for recurrence, infiltrating the urinary bladder, bilateral VUJ and prostate. PET-CT revealed an FDG-avid soft tissue lesion arising from the postoperative bed, extending along the left lateral pelvic wall and rectovesical pouch, measuring 10 × 7 × 8 cm with SUVmax 5.7, involving the left ureter, left seminal vesicle, left lobe of prostate, mesorectal fascia and presacral region. CT-guided biopsy showed fibrocollagenous tissue with proliferating stellate spindle cells, minimal pleomorphism, 2 to 3 mitoses/10 HPF, stromal hyalinization and no necrosis. IHC showed strong nuclear beta-catenin positivity with patchy mild SMA, SAT-B2 and desmin expression, while CK, S-100, ALK-1, CD34, STAT6 and CD117 were negative. Ki-67 was 8 to 10%, confirming desmoid-type fibromatosis/aggressive fibromatosis. As the tumour was unresectable, liposomal doxorubicin was given but was not tolerated; the patient developed three vesicorectal fistulae requiring diversion colostomy. Later, methotrexate, vinblastine and tamoxifen combination therapy was given, but it caused severe bone marrow suppression. At present, the patient is off medication, clinically stable with intermittent urinary obstruction, and sorafenib is being planned, indicating aggressive biological behaviour.

Case 2: A 34-year-old male was incidentally detected to have a rapidly growing left abdominal wall mass on ultrasound, measuring 4 × 3 cm and appearing well defined. No significant previous history was mentioned in the file. The lesion was surgically resected with clear margins. Histopathology showed spindle cells with abundant collagen, and IHC demonstrated strong diffuse beta-catenin positivity, weak patchy CD34 positivity, and negativity for STAT6, ALK-1 and CD117, confirming desmoid-type fibromatosis. The case showed benign biological behaviour, and no further complication or recurrence detail was mentioned.

Case 3: A 32-year-old female with a history of three previous caesarean sections, the last performed 6 years earlier, presented with lower abdominal pain for 8 months, not associated with the menstrual cycle, along with intermittent urinary obstruction. USG showed a well-defined mass measuring 10.5 × 9.5 × 9 cm. MRI revealed a lobulated mass compressing the urinary bladder and involving the right rectus muscle. CT-guided biopsy was performed, and IHC confirmed the diagnosis of desmoid tumour. The patient is currently receiving chemotherapy and is doing well, with reduction in obstructive urinary symptoms.

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Case 4: A 34-year-old male, with a history of descending colon polyp resection 3 years earlier and a family history of colorectal carcinoma in his father, presented with a rapidly growing left abdominal wall mass arising from the postoperative site. USG showed an ill-defined mass measuring 6 × 5 × 3.5 cm. CT-guided biopsy diagnosed a spindle cell lesion, and IHC confirmed desmoid-type fibromatosis. The patient underwent surgery with partial colon resection and anastomosis. The resected sample was sent for next-generation sequencing to evaluate possible germline mutations. Postoperative outcome was not mentioned in the file.

Case 5: A 37-year-old multiparous female with a history of three vaginal deliveries presented with an umbilical mass. USG showed a well-defined suprapubic mass measuring 9.5 × 8.5 × 7 cm involving the right rectus muscle. Although the lesion was well defined, it was adherent to the ileum, so surgical removal was performed along with the adherent intestinal segment, achieving clear resection margins. Histopathology suggested a spindle cell neoplasm, with desmoid tumour as the first differential diagnosis, along with inflammatory myofibroblastic tumour and solitary fibrous tumour as differentials. IHC showed strong beta-catenin positivity, confirming desmoid-type fibromatosis. The patient is stable and remains on follow-up.

Case 6: A 40-year-old female, previously treated with total abdominal hysterectomy with bilateral salpingo-oophorectomy 3 years earlier for abnormal uterine bleeding with fibroid, presented with intermittent urinary obstruction and abdominal pain. USG showed a large 13 × 12 × 10 cm mass located between the urinary bladder and colon, adherent to both. MRI showed that the mass was adherent to and infiltrating the bowel, with partial compression of the bowel, rectum and colon, and extension to the right ureter causing mild hydronephrosis. Initially, the patient was kept on follow-up for a few months, but symptoms became aggressive with rising creatinine, hematuria and recurrent urinary tract infection. Due to progressive local invasion and urinary complications, total pelvic exenteration was suggested. The patient then went to a higher centre in Mumbai for further management, and final treatment outcome was not available.

Histopathology Images of the Cases

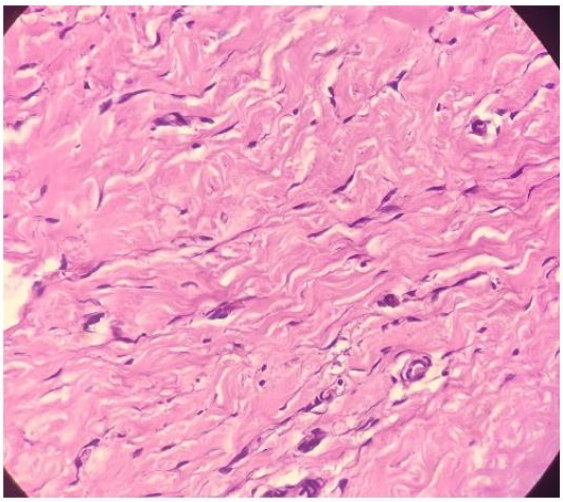

Figure 1: Spindle cells in densely collagenized fascicles

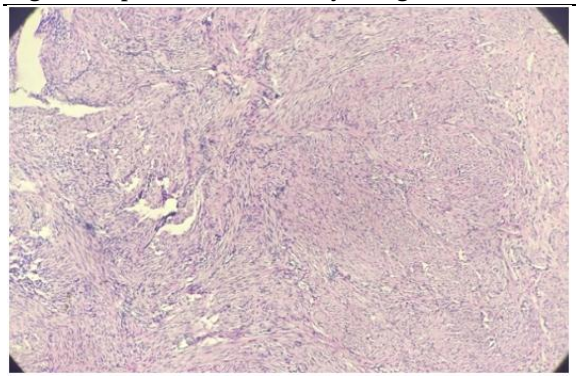

Figure 2: Spindle cell fascicles in collagenous stroma

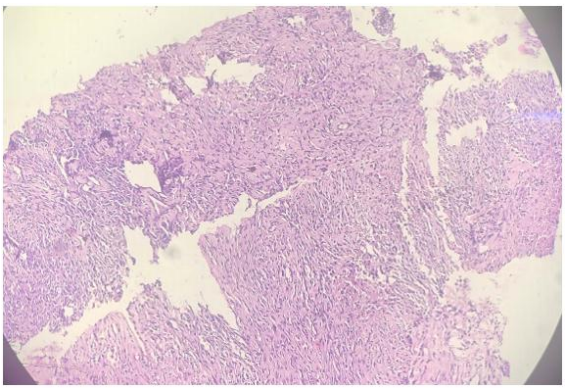

Figure 3: Low-power H&E showing desmoid-type fibromatosis

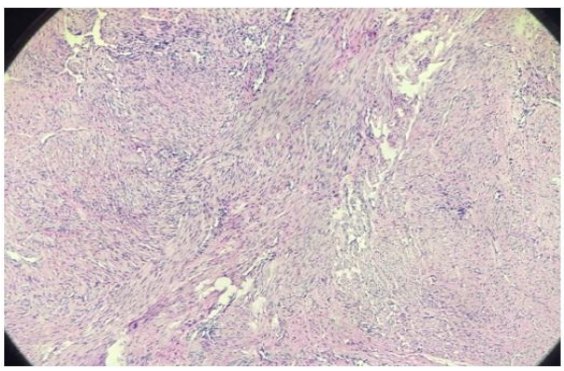

Figure 4: Bland spindle cells in densely collagenized stroma

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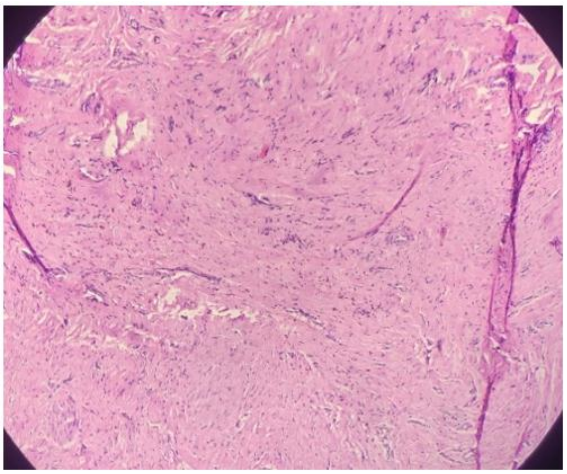

Figure 5: Bland spindle cell neoplasm on low-power H&E

DISCUSSION

The present cases highlight the wide clinicopathological spectrum of desmoid-type fibromatosis, ranging from a small, incidentally detected and completely resectable abdominal wall lesion to deeply infiltrative pelvic disease producing urinary obstruction, hematuria, ureteric involvement and treatment-limiting morbidity. This heterogeneity closely corresponds with the Desmoid Tumor Working Group consensus by Alman, Kasper, Gronchi, Gounder and colleagues (2020), which described desmoid tumour as a rare, locally aggressive fibroblastic proliferation with an unpredictable clinical course, where management has shifted from routine upfront surgery to individualized conservative or active treatment strategies.[7] In our series, Case 1 and Case 6 represent the aggressive end of this spectrum. Case 1 developed a large postoperative pelvic mass after previous colorectal carcinoma treatment, radiologically mimicking recurrence, with infiltration of bladder, ureter, prostate, seminal vesicle and presacral region; histology and strong nuclear beta-catenin confirmed desmoid-type fibromatosis. Case 6 similarly showed a large pelvic mass after hysterectomy, adherent to bladder and colon, compressing bowel and ureter with hydronephrosis, rising creatinine, hematuria and recurrent UTI, for which pelvic exenteration was advised. These cases support the concept that desmoid tumours are non-metastatic but can cause major local organ morbidity due to infiltrative behaviour. The diagnostic approach in our cases was strongly supported by histology and immunohistochemistry. An et al. (2021), in a clinicopathological study of desmoid-type fibromatosis, confirmed the diagnostic value of beta-catenin immunostaining and CTNNB1 mutation analysis, reporting nuclear beta-catenin localization in 56 of 70 cases and CTNNB1 mutations in 43 cases.[8] This is directly comparable with our cases, where beta-catenin positivity confirmed desmoid fibromatosis in the abdominal wall and pelvic lesions, while exclusion of CK, S-100, ALK-1, CD34, STAT6 and CD117 helped rule out important spindle-cell differentials. Case 3, a female with three previous caesarean sections and rectus muscle involvement, and Case 5, a multiparous female with rectus muscle involvement, support the recognized association of abdominal wall desmoid tumours with pregnancy-related and surgical trauma. Drabbe et al. (2023) reported that pregnancy-associated desmoid fibromatosis often occurs in the abdominal wall and may be related to physiological trauma during pregnancy and caesarean-section-related surgical trauma.[9] The more favourable behaviour in Case 2 and Case 5, where complete surgical excision with clear margins was achieved, is consistent with Nishida et al. (2021), who studied 34 abdominal wall desmoid cases and found that active surveillance was successful in 15 cases, while less-invasive surgery showed low recurrence, with only one recurrence among 15 surgically treated patients at a mean follow-up of 45 months.[10] In contrast, Case 1 required systemic therapy due to unresectability, but liposomal doxorubicin was poorly tolerated and methotrexate-vinblastine with tamoxifen caused severe marrow suppression. The planned use of sorafenib is evidence-based, as Gounder et al. (2018), in a randomized trial of advanced or refractory desmoid tumours, reported significantly better 2-year progression-free survival with sorafenib compared with placebo, 81% versus 36%, with objective response rates of 33% versus 20%.[11] Case 4 also emphasizes the need for genetic evaluation, because prior colonic polyp resection and family history of colorectal carcinoma raise concern for an APC/FAP-related background; therefore, sending tissue for NGS was appropriate. Overall, these cases demonstrate that desmoid fibromatosis should not be judged only by histological blandness, as its clinical behaviour depends on site, size, organ involvement, resectability, surgical history, genetic background and response to systemic therapy.

CONCLUSION

Desmoid fibromatosis demonstrates a wide clinicopathological spectrum despite its non-metastatic nature. The present cases show that tumour behaviour varies from small, resectable abdominal wall lesions with favourable outcome to large pelvic masses causing urinary obstruction, bowel involvement, fistula formation and major treatment difficulty. Histopathology with nuclear beta-catenin positivity remains central for diagnosis and for excluding spindle-cell mimics. Prior surgery, pregnancy-related trauma, intra-abdominal location and possible genetic background may influence presentation and progression. Therefore, desmoid fibromatosis requires individualized

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multidisciplinary management, balancing observation, surgery, systemic therapy and follow-up according to site, symptoms, resectability and biological aggressiveness.

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