DOI: 10.7860/JCDR/2023/64000.18257

Case Series

Pathology Section

# Dermatofibrosarcoma Protuberans at Unusual Sites: A Case Series

SUBRAT BURMA[1], GAYATRI RATH[2]



## ABSTRACT

Dermatofibrosarcoma Protuberans (DFSP) is a rare, locally aggressive malignant subcutaneous soft tissue tumour that often goes undiagnosed due to its indolent nature and non protuberant growth. Recurrences after surgical excision are a common issue. While the trunk and extremities are commonly affected sites, rarer locations such as the genitalia and breast have been reported. The rarity of the lesion, its occasional unusual presentation sites, the indolent yet locally aggressive nature, and the diagnostic and therapeutic challenges are the distinctive features of this tumour. This study presents three unusual sites of DFSP: scrotum, vulva, and pubic region, one of which is an uncommon variant known as pigmented DFSP (Bednar tumour).

**Keywords:** Bednar tumour, Cutaneous sarcoma, Immunohistochemical staining

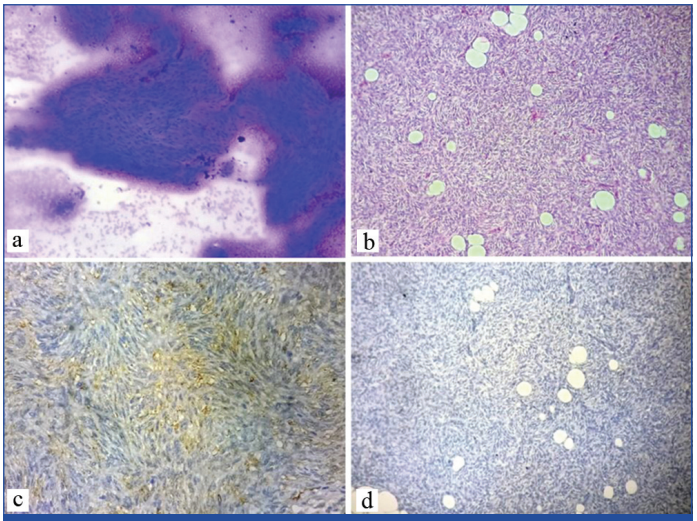
## INTRODUCTION

DFSP is a rare, locally aggressive superficial mesenchymal neoplasm of fibroblastic origin. Due to the peculiar indolent nature of the tumour, patients often present at an advanced stage. The late presentation, low but existing risk of distant spread, and high rate of local recurrences even after wide local surgery pose therapeutic challenges for treating physicians. The trunk (42-72%), followed by the proximal extremities (16-30%), head and neck (13%), and genitalia (1%), are common sites of affection [1]. About 90% of DFSPs are low-grade sarcomas (malignant potential very low, <5%), while 10% are mixed with a high-grade sarcomatous component [2]. Metastasis to regional lymph nodes is rare, and distant spread is even rarer. The lungs are the most common site of distant spread, with other sites including the brain, bone, and other soft tissues [3]. The rarity of the lesion, its uncommon presentation sites, the indolent yet locally aggressive nature, and the diagnostic and therapeutic challenges have prompted the undertaking of this study. Here, three cases of DFSP are reported at three different and very uncommon sites like scrotum, vulva, and pubic region.

## CASE SERIES

### Case 1

A 38-year-old male presented to the surgical Outpatient Department (OPD) with a slowly enlarging mass in the scrotum over the past five years. He had no other associated co-morbidities. On local examination, the swelling measured approximately 10×8×5 cm, was soft to firm, and non tender. Fine Needle Aspiration Cytology (FNAC) was performed on the mass, which showed interlacing fascicles of benign spindle cells with a mild degree of nuclear pleomorphism [Table/Fig-1a]. This was reported as a benign spindle cell tumour with a differential diagnosis of benign fibroblastic spindle cell tumour, and biopsy was recommended. Routine haematological investigations were normal, except for mild anaemia Haemoglobin (Hb) 10.2 gm/dL). The mass was resected and sent for histopathological study. Gross examination revealed a partially skin-covered mass measuring about 8x6x4 cm. On the cut section, a greyish-white solid tumour was observed in the dermis. Microsections showed a well-circumscribed proliferation of spindle-shaped cells arranged in a distinct storiform pattern with a grenz zone between the epidermis and tumour mass [Table/Fig-1b]. Individual tumour cells exhibited mild nuclear pleomorphism with a low mitotic rate (2-3 per 10 high-power field (hpf)). A provisional diagnosis of DFSP with a differential diagnosis of cutaneous leiomyosarcoma was made. Immunohistochemical (IHC) study was conducted to reach a definitive diagnosis. The tumour cells showed strong CD34 positivity and negative Desmin immunostaining [Table/Fig-1c,d]. Therefore, the final diagnosis of DFSP was established. The patient was advised to report at four-month intervals for a period of one year after surgery. No recurrences were observed, and neoadjuvant or radiotherapy was not administered.

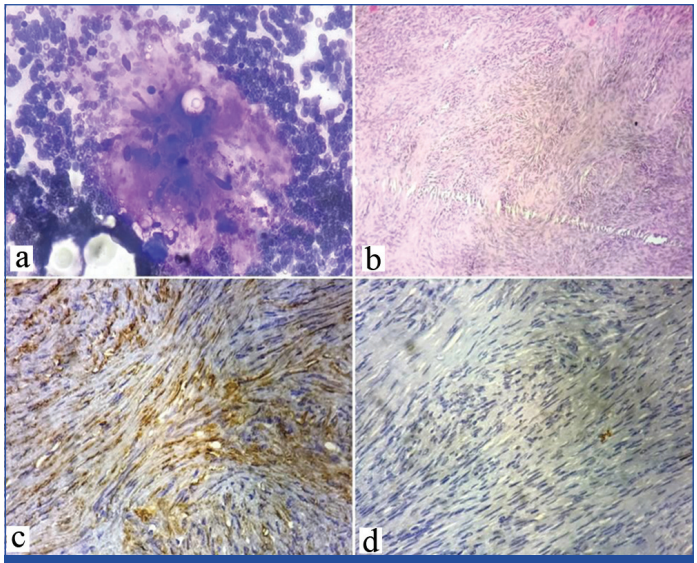



**[Table/Fig-1]:** a) Cytosmear shows interlacing fascicles of benign spindle shaped cells with mild nuclear pleomorphism (Diff Quik, 10X). b) Histopathological sections showing spindle shaped cells arranged in storiform pattern with entrapped adipocytes (H&E, 10X); c) CD34 positive; d) Desmin negative.

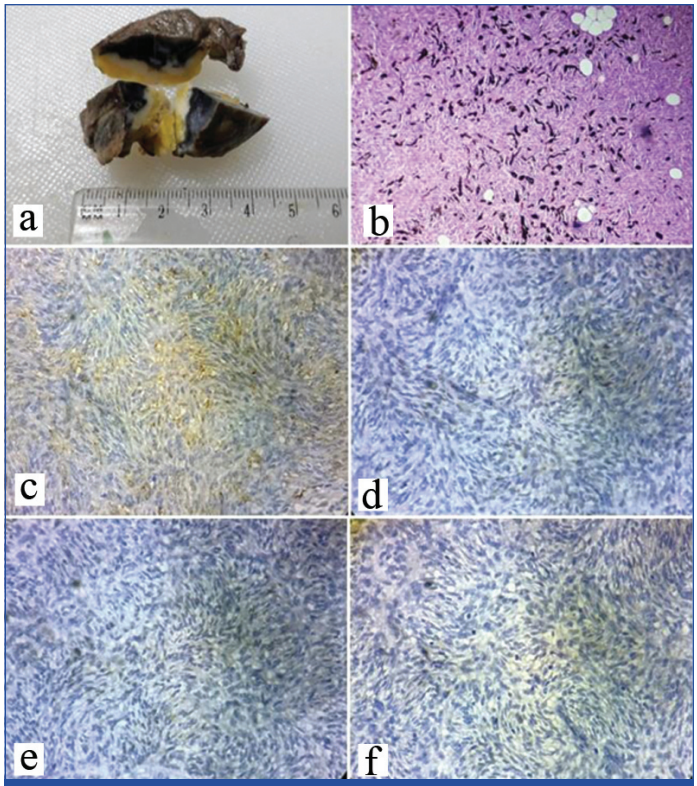
### Case 2

A 40-year-old female presented with a slowly growing mass in the vulva for a duration of four years. On local examination, the mass measured approximately 6×4×3 cm. It was firm, tender, and had mild surface ulceration. FNAC was performed, and the report indicated a benign spindle cell tumour [Table/Fig-2a]. The mass was then resected and sent for histopathological study. Grossly, a partially skin-covered mass measuring about 5×4×2 cm was received. Microsections revealed a lobulated mass with well-defined borders that extended to the deep resected margin. The tumour cells were arranged in short interlacing fascicles and occasionally showed a vague storiform pattern [Table/Fig-2b]. Some areas exhibited acellular regions resembling Verocay bodies seen in Schwannoma. Tumour cell infiltration was observed at the resected margins. The mitotic rate was 4-5 per 10 hpf. IHC analysis showed that the

tumour cells were positive for CD34 [Table/Fig-2c] and negative for S100 [Table/Fig-2d]. Therefore, the final diagnosis was DFSP with involvement of the deep resected margin. The patient was advised to follow-up every two months for one year postsurgery. However, during subsequent follow-ups, the patient did not exhibit any signs of recurrence. As a result, no further medical therapy or surgical intervention was recommended.




**[Table/Fig-2]:** a) Cytosmear shows spindle shaped cells present over eosinophilic matrix substance in discohesive manner (Diff Quik, 40X); b) Histopathological sections showing spindle shaped cells arranged in short interlacing fascicles with hypocellular areas in between (H&E, 10X); c) CD34 positive; d) S100 negative.




**[Table/Fig-3]:** a) Gross picture shows irregular firm mass with brownish black discoloration of superficial part of the tumour; b) Micro section showing spindle shaped cells arranged in short interlacing fascicles with brownish black pigment deposition within the tumour cells (H&E, 10X); c) CD34 positive; d) S100 negative. e) Desmin negative; f) HMB45 negative.

### Case 3

A 45-year-old female presented at the surgery OPD with a slowly growing mass in the pubic region persisting for six years. On local examination, the mass measured approximately 6×5×3 cm. It appeared blackish in colour, firm, and non tender. Routine haematological investigations showed results within normal limits. FNAC was performed, and a diagnosis of malignant spindle cell tumour with a differential diagnosis of desmoplastic malignant melanoma was provided. The mass was subsequently resected and sent for histopathological study. Grossly, the mass measured about 5×4×3 cm and exhibited brownish-black pigmentation [Table/Fig-3a]. Microscopic examination revealed spindle-shaped cells arranged in short interlacing fascicles and a storiform pattern, with the presence of brownish-black pigments [Table/Fig-3b]. The final diagnosis was pigmented variant of DFSP (Bednar tumour), with a differential diagnosis of desmoplastic malignant melanoma. On IHC study, the tumour cells were positive for CD34 but negative for S100, Desmin, and HMB45 immunostaining [Table/Fig-3c-f], respectively. The patient was advised to follow-up every two months for a period of one year. However, since the patient did not attend the follow-up appointments and was from a remote location, further monitoring was not possible. The demographic, clinical, FNAC diagnosis and follow-up details of all three cases are given in [Table/Fig-4].

## DISCUSSION

The DFSP is a rare, locally aggressive superficial mesenchymal neoplasm of fibroblastic origin, accounting for <0.1% of all malignancies and <1% of all soft tissue sarcomas [4]. The estimated occurrence of DFSP in the United States (US) ranges from 0.8-4.5 cases per million persons per year [5], and in India, it is 0.8-4.1 cases per million people annually. DFSP commonly affects individuals between 25-45 years of age, with a slight female preponderance. In 90% of DFSP cases, a chromosomal translocation t(17;22)(q22;q13) involving chromosome 17 and chromosome 22 is found. This translocation activates the platelet-derived growth factor beta polypeptide fusion gene, leading to overproduction of platelet-derived growth factor, cell proliferation, and tumour formation [6].

Clinical presentation of DFSP can vary from a flat plaque-like lesion to a nodular subcutaneous mass. In this study, all three cases presented as nodular masses. The first case was an uncomplicated case of DFSP presenting at a rarer site like scrotum. Paravathaneni M et al., reported a case of DFSP scrotum in a 24-year-old male patient, which was also an uncomplicated case of DFSP [7]. The second case presented in a relatively advanced stage, as there was tumour cell infiltration in deep resected margin. Goyal LD et al., reported a case of recurrent DFSP vulva in a 35-year old female patient [8]. Microscopic examination of DFSP typically reveals uniform spindle cells arranged in fascicles and a storiform pattern. Variants such as those with giant cells, pigmentation (Bednar tumour), myxoid differentiation, pseudo cystic change, atrophic, features, and sarcomatous transformation can also be encountered [9]. Tumour cells typically exhibit strong and diffuse CD34 immunoreactivity [10]. Sardesai VR et al., reported a case of

| Case No. | Age (years) | Gender | Site | Size | Clinical diagnosis | FNAC diagnosis | IHC markers | HP diagnosis | Follow-up |
|---|---|---|---|---|---|---|---|---|---|
| 1 | 38 | Male | Scrotum | 10×8×5 cm | Scrotal mass (Leiomyoma) | Benign spindle cell tumour (Fibroblastic) | CD34 (+) Desmin (-) | DFSP | No recurrence till last visit |
| 2 | 40 | Female | Vulva | 6×4×3 cm | Vulval angiofibroma | Benign spindle cell tumour | CD34 (+) S100 (-) | DFSP | No recurrence till last visit |
| 3 | 45 | Female | Pubic area | 5×4×3 cm | Melanocytic nevus/ Melanoma | Malignant spindle cell tumour/ Desmoplastic melanoma | CD34 (+) S100 (-) Desmin (-) HMB45 (-) | Pigmented DFSP (Bednar tumour) | No follow-up visit |

**[Table/Fig-4]:** Summarises the clinical, FNAC, histopathological diagnosis and IHC studies, along with follow-up details of the three cases reported here.

recurrent DFSP in a rare site like pubic region in a 40-year old male patient [11]. The third case included in this study also presented at pubic region, but was a variant (pigmented) of DFSP that is Bednar tumour. Bednar tumour may undergo malignant transformation to fibrosarcoma. Hence, these cases should be followed-up regularly.

The standard treatment is surgery with wide local excision. Mohs' micrographic surgery is done for small and cosmetically sensitive regions, which has a high cure rate with lowered incidence of recurrence [12]. Patients unsuitable for surgery are managed with radiation and targeted therapies [13]. According to Chen YT et al., adjuvant radiotherapy may be considered for all patients after surgical excision irrespective of the surgical margin [14]. Recurrence rate in those treated with wide local excision and those treated with Mohs Micrographic Surgery are 7.3% and 1%, respectively [15]. Recurrences are treated with re-resection, or are managed with radiotherapy and/or Imatinib therapy. Postsurgical periodic surveillance should be done to monitor recurrence and metastasis.

## CONCLUSION(S)

The general prognosis for DFSP is excellent; however, timely diagnosis and early treatment are of paramount importance. Additionally, even seemingly benign lesions should not be ignored and should be promptly brought to the attention of a surgeon, particularly if there are any changes in dimensions, colour, or other characteristics. Early treatment will undoubtedly help prevent complications and improve patient survival.

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**PARTICULARS OF CONTRIBUTORS:**
1. Assistant Professor, Department of Pathology, SLN Medical College, Koraput, Odisha, India.
2. Associate Professor, Department of Pathology, FM Medical College, Balasore, Odisha, India.

**NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:**
Dr. Gayatri Rath,
N4/288, IRC Village, Nayapali, Bhubaneswar-751015, Odisha, India.
E-mail: drgrath1968@gmail.com



**PLAGIARISM CHECKING METHODS:** [Jain H et al.]
- Plagiarism X-checker: Mar 13, 2023
- Manual Googling: May 09, 2023
- iThenticate Software: Jun 12, 2023 (8%)

**ETYMOLOGY:** Author Origin

**EMENDATIONS:** 6

**AUTHOR DECLARATION:**
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Mar 10, 2023**
Date of Peer Review: **Apr 17, 2023**
Date of Acceptance: **Jun 15, 2023**
Date of Publishing: **Aug 01, 2023**