International Surgery Journal
Aggarwal M et al. Int Surg J. 2021 Oct;8(10):3171-3175
http://www.ijsurgery.com
pISSN 2349-3305 | eISSN 2349-2902

DOI: https://dx.doi.org/10.18203/2349-2902.isj20214018

Case Report

Rare giant dermatofibrosarcoma protuberans with uncommon fibrosarcomatous transformation: case report and review of literature

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Received: 04 August 2021
Accepted: 04 September 2021

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ABSTRACT

Dermatofibrosarcoma protuberans (DFSP) is a rare soft tissue sarcoma with aggressive local behavior but with a low metastatic potential. Although slow growing and indolent, they rarely reach huge sizes. Very uncommonly, these locally invasive tumors undergo fibrosarcomatous transformation with a more aggressive clinical course, with higher rate of recurrence risk and distant metastases. A 32-years-old lady, presented with a gradually progressive lump in the upper central back for the past 6 years, with rapid progression in size during the last 6 months. On examination, she had a single lump of size 18×18 cm in the midline of the upper back, with prominent veins over its surface. Magnetic resonance imaging (MRI) showed no connection with the spinal canal and appeared flush with the paraspinal muscles. Core needle biopsy showed DFSP. The patient underwent a wide local excision with split skin grafting. The histopathology now showed a fibrosarcomatous transformation of DFSP. The patient again underwent a wide re-excision with a 3 cm margin. Histology reported no evidence of tumor cells in the specimen. The patient's postoperative period was uneventful and she was referred for adjuvant radiotherapy. DFSP is a rare, slow-growing malignant fibroblastic mesenchymal skin tumor with low metastatic potential. However, in any patient with long standing DFSP with a recent increase in size, this fibrosarcomatous transformation must be kept in mind as it represents an uncommon form of DFSP that tends to follow a more aggressive clinical course, with higher rate of recurrence risk and distant metastases.

Keywords: Dermatofibrosarcoma protuberans, Wide local excision, Recurrence, Adjuvant radiotherapy, Imatinib

INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a rare soft tissue sarcoma with aggressive local behavior but with a low metastatic potential, with an annual incidence of approximately 4 per million. DFSP constitutes approximately 1% of all sarcomas and <1% of all malignancies.[1]

Although slow growing and indolent, they rarely reach huge sizes. Very uncommonly, these locally invasive tumors undergo fibrosarcomatous transformation with a more aggressive clinical course, with higher rate of recurrence risk and distant metastases.[2]

We here present a report of one such patient who had a slow growing lump over her back for many years which reached a giant size, and then started rapidly increasing in size.

CASE REPORT

A 32-years-old lady, presented to our surgery outpatient with complaints of a gradually progressive lump in the upper central back for the past 6 years, with rapid progression in size during the last 6 months, along with pain over the lump for the past 4 months (Figure 1 and 2). On examination, she had a single lump of size 18×18 cm in the midline of the upper back, with prominent veins over

its surface, with a firm consistency and having a bosselated surface, with no regional cervical or axillary lymphadenopathy. Core needle biopsy showed DFSP with mild focal CD 34 positivity, and smooth muscle antibodies (SMA) negative. Magnetic resonance imaging (MRI) of the cervical and dorsal spine revealed a large 13.3×13.6×9.3 cm mass in cervicodorsal region, hypointense, with few incomplete septations compressing the paraspinal muscles on sagittal T1W image. On T2W coronal image the mass was hyperintense with multiple tiny flow voids suggestive of vessels. On T2W sagittal image the mass was slightly hyperintense. On axial T1W image the mass showed no connection with the spinal canal and appeared flush with the paraspinal muscles (Figure 3-6).

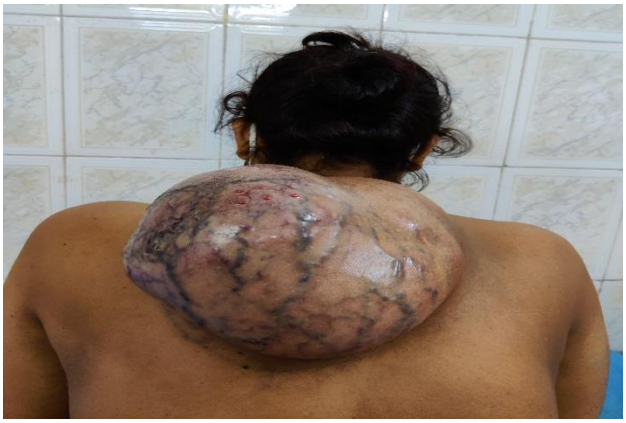

Figure 1: Clinical image depicting the large mass located in the upper central back.

Figure 2: Clinical image (side view) of the same mass.

With the diagnosis of giant DFSP, the patient underwent a wide local excision with split skin grafting. Histological examination revealed a relatively circumscribed tumor, with storiform to whorled pattern of tumour cells. Focal areas showed high cellularity with tumor cells arranged in long and short intersecting fascicles giving a herring-bone appearance. Tumor cells had elongated spindled nuclei, coarse granular chromatin, inconspicious nucleoli and bipolar eosinophilic cytoplasm. Brisk mitosis was seen >20/10 hpf. Tumor was abutting underlying skeletal muscle, however not infiltrating it, and numerous tongues of tumor reaching upto reticular dermis and destroying the adnexal structures was noted. The lateral and superior soft tissue margin was involved by the tumor; all skin margins were free of tumor. On immunohistochemistry, tumor cells showed focal, patchy positivity for SMA, whereas they were negative for CD 34, EMA, cytokeratin, desmin, myogen, CD99, Bcl-2 and S-100 immunostains (Figure 7).

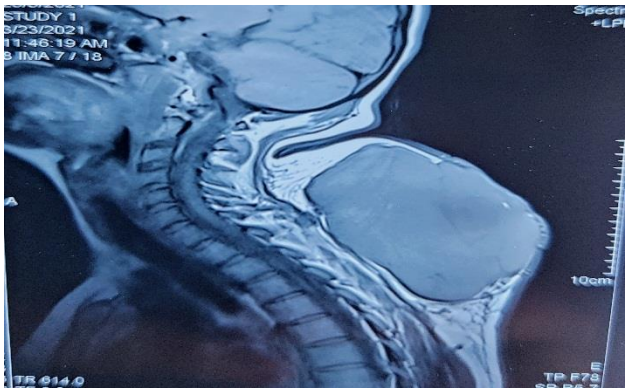

Figure 3: Sagittal T1W image on MRI demonstrating large 13.3×13.6×9.3 cm mass in the cervicodorsal region, hypointense, with a few incomplete septations compressing the paraspinal muscles.

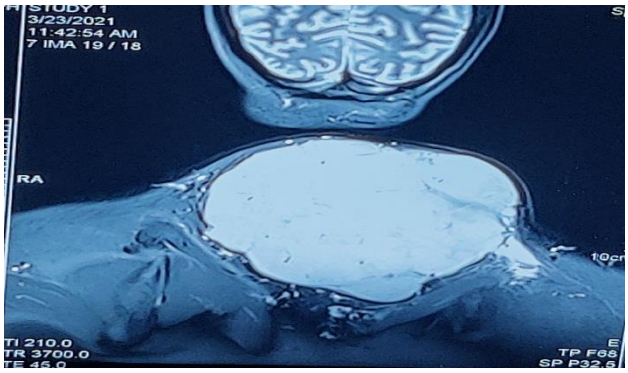

Figure 4: T2W coronal image on MRI depicting the mass was hyperintense with multiple tiny flow voids suggestive of vessels.

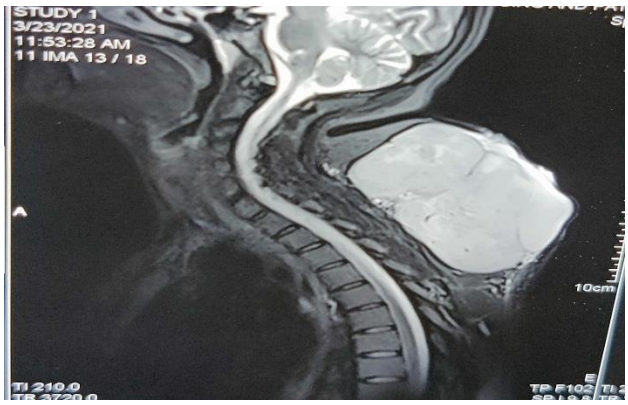

Figure 5: T2W sagittal image on MRI showing the mass was slightly hyperintense.

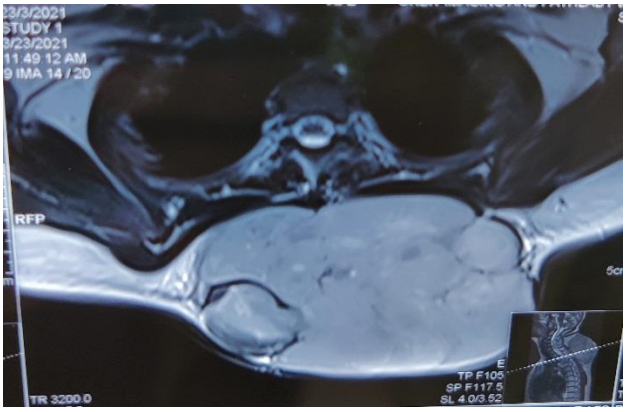

Figure 6: On axial T1W image on MRI the mass showed no connection with the spinal canal and appears flush with the paraspinal muscles.

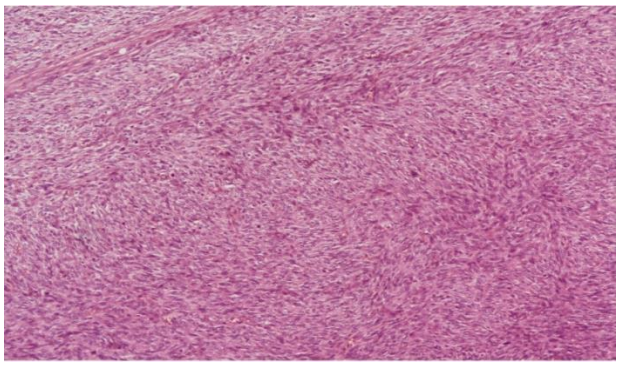

Figure 7: HPE image showing herring bone pattern, classical of fibrosarcomatous transformation of a DFSP.

The diagnosis was made of a fibrosarcomatous transformation of DFSP. The patient again underwent a wide re-excision with a 3 cm margin. Histology reported no evidence of tumor cells in the specimen. The patient's postoperative period was uneventful and she was referred for adjuvant radiotherapy.

Patient is doing well on follow-up.

DISCUSSION

DFSP is a rare, slow-growing malignant fibroblastic mesenchymal skin tumor which constitutes less than 0.1% of all malignant neoplasms and 1% of all soft tissue sarcomas.[3] DFSP most commonly affects patients in their mid- to late 30s; however, the disease can occur at any age. The most common location of DFSP is the trunk (42-72%) followed by proximal extremities (20-30%), and head and neck (10-16%).[4] DFSP sites include surgical scars, old burns, trauma, radiation dermatitis, vaccination sites, central venous line puncture sites and even insect bites.[4,5] Blacks have slightly higher incidence than whites. Both men and women are equally affected.[6] DFSP classically presents as a solitary, frequently asymptomatic, plaque with violaceous to blue hue. The tumor exhibits slow growth. The Bednar tumor is a rare pigmented variant of DFSP.[7]

Approximately 90% of DFSP are low grade, rest are intermediate or high grade because of presence of fibrosarcomatous transformation.[8] More than 90% of DFSP feature a translocation between chromosomes 17 and 22, resulting in the fusion between the collagen type Iα1 gene (COL1A1) and the platelet derived growth factor (PDGF) β-chain gene (PDGFB). Thus, the growth of DFSP is a result of the deregulation of PDGF β-chain expression and activation of PDGF receptor (PDGFR) protein tyrosine kinase.[9,10]

Lymph node metastases occur in approximately 1% of cases, and distant metastases, principally to lung, occur in approximately 4% of DFSP cases. DFSP-associated mortality is low. A fibrosarcomatous variant, DFSP-FS, represents an uncommon form of DFSP that tends to follow a more aggressive clinical course, with higher rate of recurrence risk and distant metastases.[2]

MRI studies are not specific since they may not always distinguish DFSPs from other soft tissue sarcomas. Therefore, histological examination is the only definitive diagnostic method. Histologically DFSP is characterized by diffuse infiltration of the dermis and subcutis, usually sparing the epidermis and skin appendages, and is composed of monomorphous, dense spindle cells arranged in a storiform pattern and embedded in a sparse to moderately dense fibrous stroma.[11] Irregular projections (tentacle-like) of the tumor are common and may account for the high incidence of local recurrence after excision. Atypia is minimal, and mitoses are rare.[12] Immunohistochemically, tumor cells stain for vimentin, CD34, apolipoprotein D, nestin, and may be for EMA. Desmin, S100 protein, FXIIIa, stromelysin III, HMGA1&2, tenascin, D2-40, CD163, and keratins are negative. In myoid nodules, tumor cells stain for SMA. Fibrosarcomatous DFSP may show loss of CD34 positivity and increased expression of TP53.[12-14] In our case, CD 34 was negative and p53 patchy weak to moderate nuclear positive.

Treatment options for DFSP include wide local excision (WLE) and Mohs micrographic surgery (MMS). WLE with a minimal margin of at least 3 cm of surrounding skin, including the underlying fascia, without elective lymph node dissection is advocated.[15] The likelihood of local recurrence is directly proportional to the adequacy of surgical margins. Reconstructive surgery may be required to restore tissue defects after excision using a local skin flap, skin graft or myocutaneous flap.[16] For well-defined tumors located on trunk or extremities, WLE is likely to achieve tumor clearance with satisfactory cosmetic and functional result. However, extirpation of tumor by MMS, using frozen sections with or without confirmation by examination of paraffin-embedded sections, may be beneficial in sites where maximum conservation of normal tissue is required. The MMS technique consists of

successive horizontal sectioning (5-7 µm) during resection and immediate frozen microscopic examination until a tumor-free margin is succeeded.[4] There are reports of local cure rates of 93-100%.[17,18]

Alternative treatment options for DFSP include RT and chemotherapy. RT was used selectively in a number of cases if surgical resection was not possible or would result in major cosmetic or functional deficit, with good local response.[19,20] Radiotherapy should be considered in cases of positive or inadequate margins, in cases of recurrence, or cases of unacceptable functional or cosmetic results after wide excision, in combination with surgery.[21,22] A PDGF receptor inhibitor, imatinib, has been used with clinical success in advanced disease.[23,24] Imatinib inhibits the tyrosine kinase of PDGF and seems effective in treating DFSP in patients with t (17;22) translocation.[25] Patients with DFSP should be followed closely for evidence of local or regional recurrence or metastatic disease. DFSP has a tendency to recur locally. The average time for recurrence is within the first 3 years. Although metastases are rare, multiple local recurrences appear to predispose to distant metastases.[26]

CONCLUSION

DFSP is a rare, slow-growing malignant fibroblastic mesenchymal skin tumor which constitutes less than 0.1% of all malignant neoplasms and 1% of all soft tissue sarcomas. Our patient had a slow growing swelling over her central back for past 6 years, but with a more rapid increase for the last 6 months due to an uncommon fibrosarcomatous transformation. This fibrosarcomatous variant must be kept in mind in a patient with DFSP as it represents an uncommon form of DFSP that tends to follow a more aggressive clinical course, with higher rate of recurrence risk and distant metastases.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Aggarwal M, Manchikanti D, Misra S, Thomas S, Arsia A, Pusuluri R, et al. Rare giant dermatofibrosarcoma protuberans with uncommon fibrosarcomatous transformation: case report and review of literature. Int Surg J 2021;8:3171-5.