Case Report

Low-grade dermatofibrosarcoma protuberance - A rare case report

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Abstract

Dermatofibrosarcoma protuberans (DFSP) is a slow-growing, soft-tissue tumour of early or mid-adult life, affecting both the genders equally. The most common sites are soft tissue of the trunk (50 to 60%), followed by proximal extremities (20 to 30%) and the head and neck region (10 to 15%). Its metastatic potential is low though the local recurrence rate is high. Here, we report a case of a female patient with a large soft tissue growth located at the right cheek, chin and neck region. Local excision was done under the impression of a benign tumour such as lipoma or sebaceous cyst. Histological evaluation showed bland spindle cells arranged in a storiform pattern questioning the provisional diagnosis of the lesion. Further evaluation with the immunohistochemistry (IHC) panel confirmed the diagnosis of DFSP. Since it is a rare tumour of the head and neck region with non-alarming initial presentation and the potential for erroneous diagnosis as another lesion, we present a case of DFSP.

Keywords: Cartwheel, CD34, dermatofibrosarcoma protuberans, DFSP, storiform

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INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a soft tissue lesion originating from the dermal layer of the skin,[1] comprising approximately 1.8% of all soft tissue sarcomas and 0.1% of all cancers.[2] They are low-grade tumours with low metastatic potential. In about 5% to 20% of cases, DFSP have high-grade fibrosarcomatous (FS) components and this transformation is responsible for their high incidence of local relapse and distant metastases.[3] It was first identified by Taylor, RW in 1890.[4] Then described in 1924 by Darier J,[4] and Ferrand M, as a progressive recurrent dermatofibroma, and was later given the term DFSP by Hoffmann in 1925.[5]

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It has an annual incidence of 0.8-4.5 per million persons per year[6] and primarily affects adults of different ages (peak age 20 to 50 years)[7] with an almost equal incidence in both sexes,[3] and the presentation during childhood is rare with only 6% of tumours found in patients less than 16 years of age.[8] The most common location of DFSP is on the trunk, mainly the chest and shoulder.[9]

This mesenchymal tumour is thought to be derived from dermal stem cells or undifferentiated mesenchymal cells with fibroblastic, muscular and neurologic features.[6] Histopathologically, DFSP is identified by a pattern of monomorphous proliferation of

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Figure 1: Hyperpigmented diffuse growth seen on the right side of the cheek, chin and neck region

Figure 2: Hyperpigmented diffuse growth seen on the right side of the cheek, chin and neck region

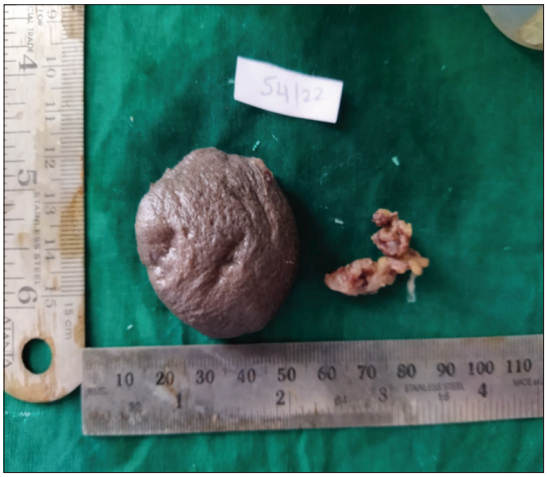

Figure 3: Grossing of the specimen

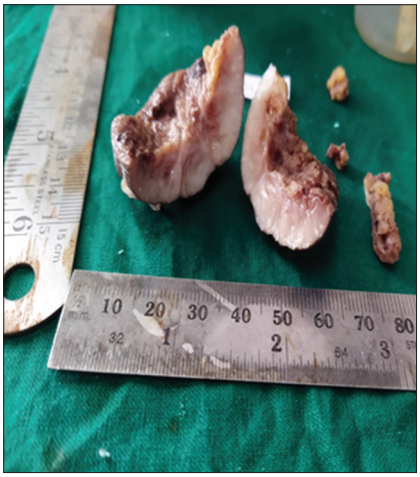

Figure 4: Cut section of the grossed specimen

cytologically bland spindle cells with a visible storiform or whorled (rushmat-like) architecture.[10-12]

Other characteristic features are low mitotic activity and deep, honeycomb infiltration into subcutaneous adipose tissue.[13] The immunohistochemical markers used for DFSP are CD34 and vimentin.[14]

The treatment of choice for DFSP is Mohs micrographic surgery or, if unavailable, conventional surgery with wide margins greater than 3 cm is advised. The prognosis for metastatic cases is very poor with a survival rate of less than 2 years following detection of metastatic disease.[2] The tumour has a local invasion, with a high recurrence rate of about 10%-60%.[15]

Case history

An 18-year-old female patient visited our hospital due to multiple areas of hyperpigmented growth on the right side of the chin, cheek and neck region which was present since 12 years. Initially, the lesion was flat, which gradually attained the present size.

On extra oral examination, hyperpigmented diffuse growth was seen on the right side of the cheek, chin and neck region measuring about 6 cm mediolaterally and 15 cm anteroposteriorly. On palpation, the lesion was soft and painless. Clinically, the diagnosis was made as neurofibroma or giant melanocytic nevus. Later, the patient was referred to the oral and [Figure 1] maxillofacial surgery department, [Figure 2] for an excisional biopsy.

Macroscopically, the received biopsy specimen was brownish, soft to firm in consistency with an irregular surface texture measuring about 3.7 × 3.5 × 2 cm. The cut section appears whitish cream in colour. The specimen was fixed in 10% buffer formalin, processed and sections of 4-micron thickness were made [Figures 3 and 4].

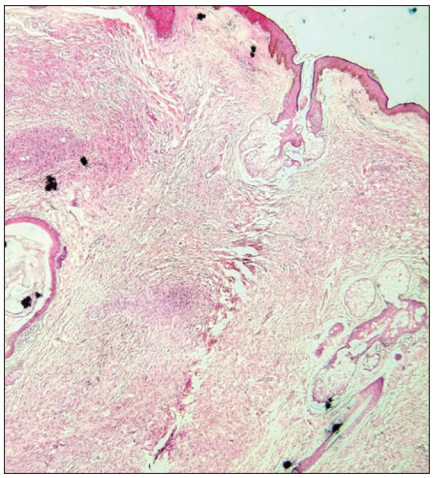

Figure 5: H&E Stain (4×) - Densely packed, monomorphous spindle cells. Poorly circumscribed with diffuse infiltration

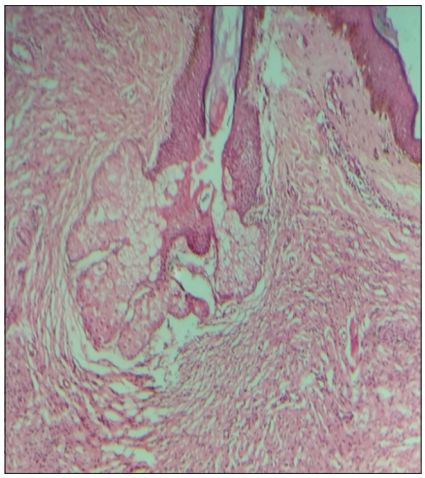

Figure 6: H&E Stain (10×) - Tumour cells extending to the surface, sparing adnexal structures

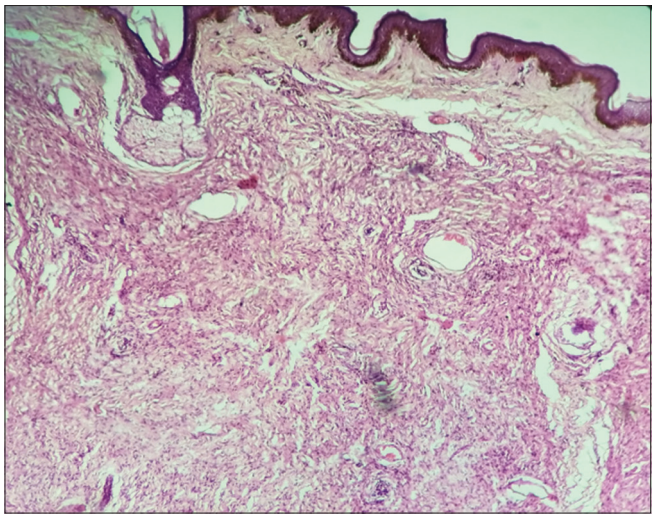

Figure 7: H&E Stain (10×) - Storiform pattern of spindle cell arrangement, with infiltrative growth pattern

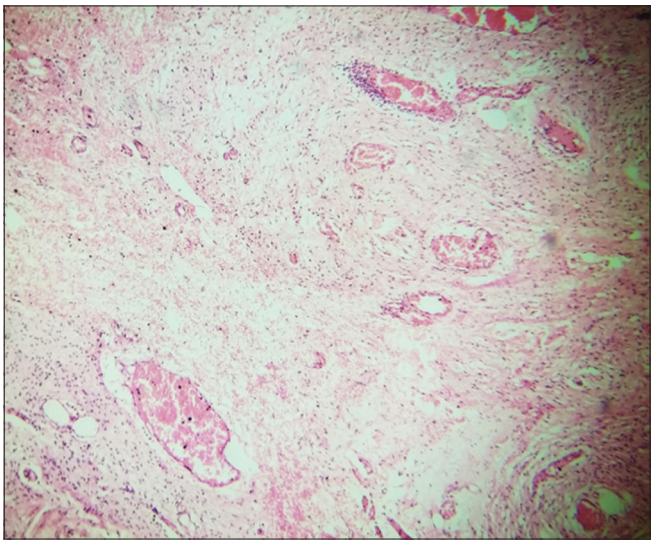

Figure 8: H&E Stain (10×) - Vascularity was predominant within the lesion, especially at the periphery of the lesion

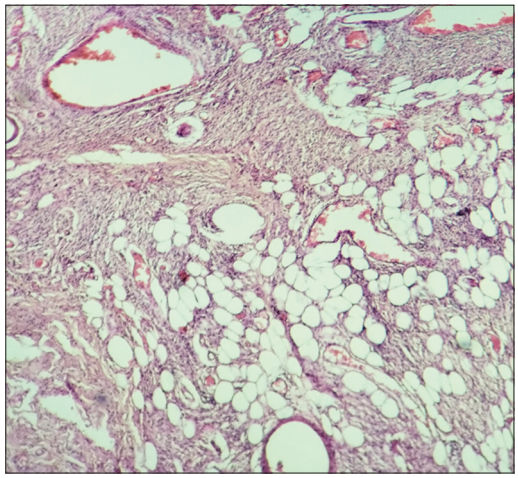

Figure 9: H&E Stain (10×) - Densely packed spindle cells infiltrating subcutis and extending around fat cells in a honeycomb fashion

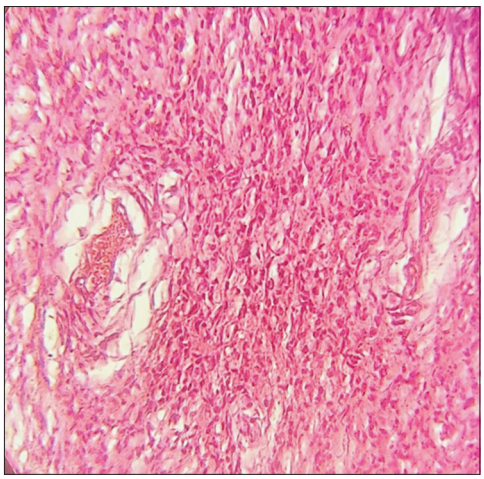

Figure 10: H&E Stain (40×) - Proliferating spindle cells

Microscopically, the section showed densely packed, monomorphous spindle cells arranged in a storiform [Figure 5] fashion which were poorly circumscribed with diffuse infiltration. [Figure 10] Tumour cells were extending

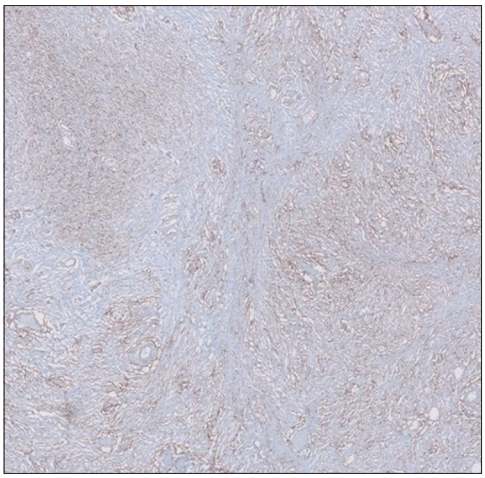

Figure 11: 4×, CD34(+)

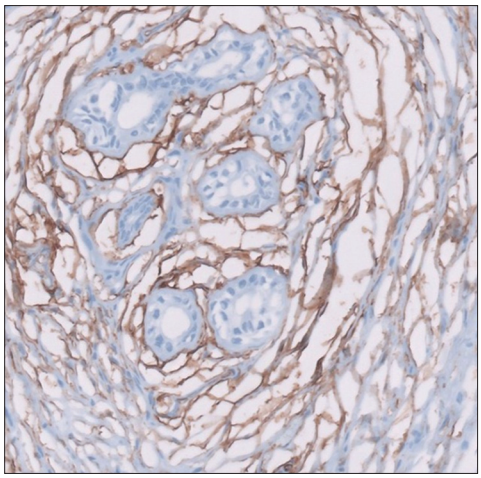

Figure 12: 40×, CD34(+)

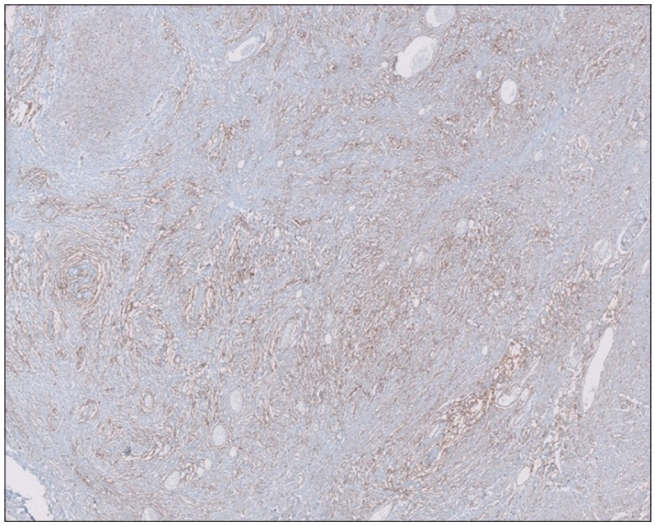

Figure 13: 4×, CD34(+)

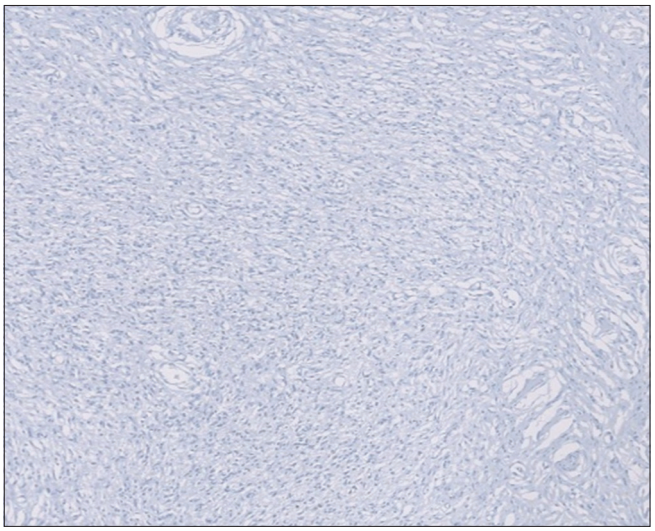

Figure 14: 4x,S-100(-)

to the surface, [Figure 7] sparing adnexal structures. Vascularity was predominant within the lesion, especially at the periphery of the lesion. [Figure 6], [Figure 8] Densely packed spindle cells were seen infiltrating subcutis and extending around fat cells in a honeycomb pattern. [Figure 9] Immunohistochemistry (IHC) panel of CD34 and S-100 was done. The tumour cells showed positivity for CD34 and were S-100 negative. Based on the histological and immunohistochemical findings, the diagnosis of DFSP was made [Figures 11-14].

DISCUSSION

DFSP is a rare mesenchymal soft tissue sarcoma with low to intermediate malignancy.[7] Studies have implicated that in DFSP, there is chromosomal translocation, resulting in the fusion protein COL1A1-PDGFB, which promotes tumour growth through the overproduction of platelet-derived growth factor (PDGF).[6]

Table 1: Differential diagnosis of dermatofibrosarcoma protuberance (DFSP) and dermatofibroma[4]

DFSP	Dermatofibroma
Grossly nodular configuration	Slightly elevated nodule with discoloured surface
Infiltration of subcutaneous tissue	No infiltration of subcutaneous tissue
Recurrence rate >5%	Recurrence rate <5%
Dendritic cells containing melanin	Spindle cells with xanthomatous and hemosiderin-laden histiocytes and inflammatory cells

It most commonly affects patients between the age group of 20 and 50 years. Congenital DFSP is a recognized entity but is extremely rare.[1] Fifty per cent of all reported cases are located on the trunk; mainly the chest and shoulder region. Other unusual locations of DFSP are the limbs (30-40%) and head and neck (10-15%).[4] The pigmented variant of DFSP (Bednar tumour) also occurs predominantly in black patients.[6]

Table 2: Differential diagnosis of dermatofibrosarcoma protuberans[16]

Tumour	Clinical feature	Histology	Immunostain
Dermatofibroma	Elevated, pedunculated or dome-shaped. More frequent in extremities, young (20-49 years) and females predominant	More pleomorphic with both small spindle-shaped fibroblastic cells and larger histiocytes admixed with chronic inflammatory cells in the dermis. Hyperkeratosis, acanthosis and pigmentation in epidermis.	XIIIa(+) CD34(-)
Schwannoma	Round, ovoid, well-circumscribed, solid mass, most common on the limbs, between 20 and 50 years	Well circumscribed with fibrous capsule, biphasic growth patterns with Antoni A (highly ordered wavy hyperchromatic spindle cells arranged in palisades) and with Antoni B (myxoid hypocellular components)	S100(+)
Cutaneous neurofibroma	Skin coloured, painless, slowly growing, solitary, soft, rubbery nodule. Frequently occurred in younger patients (20 to 40 years)	Mixed multiple cell types including Schwann cells, perineurial-like cells, fibroblastic cells, entrapped axons interspersed with shredded carrot collagen, mast cells and lymphocytes.	S-100(+) Sox10(+) CD34(+) Collagen IV(+) αSMA(-) XIIIa(-)
Intradermal spindle cell lipoma	Slowly growing, skin coloured, raised, polypoid lesion with well-defined margins.	Bland spindle cells admixed with more or less or not matured lipocytes associated with delicate ropey/ refractile collagen bundles	CD34(+) Rb(-) S-100(-) αSMA(-)

Murphy *et al*. and Moureau-Zabottoa *et al*. studied the clinical characteristics at early stage and further classified three different forms of non-protruding DFSP: (i) Morphea-like, characterized by the formation of a white or brown indurated plaque with the appearance of a scar, morphea, morpheaform basal cell carcinoma or dermatofibroma plaque; (ii) Atrophoderma-like, characterized by a soft depressed white or brown plaque that appears similar to atrophoderma or anetoderma; and (iii) Angioma-like, the least common form, made up of indurated or soft, red or violaceous plaques that have a clinical appearance similar to vascular malformations or such as morphea-like plaques, and congenital cases, such as atrophoderma-like, are more commonly seen when the lesions are located on the trunk. The initial size of the lesion at the early stage is 2-5 cm. Later, the lesion may reach even 25 cm in diameter or larger.[4]

Staging system of DFSP[16]

Stage I - Non-protuberant lesions including atrophic or sclerotic plaque, macula or small nodules.

Stage II - Protuberant primary tumour.

Stage IIA - Superficial tumour: without invasion of the underlying fascia.

Stage IIB - Deep tumour: either superficial to the fascia with infiltrating the fascia or occurred beneath the superficial fascia.

Stage III - Lymph node metastasis.

Stage IV - Distant metastasis to other organs.

Clinical differential diagnosis includes dermatofibroma, epidermal inclusion cyst, keloid and hypertrophic scar, melanoma, lymphoma, fibrosarcoma, etc. [Tables 1 and 2][4]

Grossly, DFSP is commonly a white to yellow coloured soft tissue mass which is poorly circumscribed, without a smooth outer surface. The cut surface is white to yellow, poorly encapsulated, solid and has a fish flesh-like texture. Haemorrhagic and/or cystic changes can be observed in larger tumours (>5 cm).[16]

Diagnosis may be suspected based on the tumour's clinical appearance, while physical examination may assess the extension of the tumour. Lymphatic or haematogenous dissemination is rare. Magnetic resonance imaging (MRI) is very useful for the estimation of tumour invasion, mainly in cases of large tumours or large recurrent lesions.[4]

In non-protuberant stage I with dermal plaque or subcutaneous thickening, the presence of elongated spindle cells is loosely scattered in the upper dermis without involving the grenz zone.[16]

In stage II and later with protuberant lesions, DFSP is typically featured with uniformly monomorphous spindle cells, with little atypia and mitotic activity, arranged in a storiform or whorl pattern, in the subcutaneous and dermal layers. The cellular nuclei are elongated with mild hyperchromasia, small to inconspicuous nucleoli and low to moderate quantities of cytoplasm.[16]

The neoplastic cells often infiltrate into subcutaneous adipose tissue in a honeycomb pattern which is also seen in our case. This poses a challenge to determine the true extent of the tumour tissue. All margins need to be carefully grossed and examined for residual tumour cells.[16]

DFSPs have multiple histological variants including myxoid, pigmented, giant cell, giant cell fibroblastoma, granular cell, sclerotic and fibrosarcomatous components;

these variants reflect the morphologic heterogeneity which is associated with the spindle cell differentiation during tumour development. They do not bear significant clinical manifestations and outcomes, except for the FS variant with an increased risk of local recurrence and metastatic potential.[16]

Immunohistochemically, spindle cells typically show strong and diffuse cytoplasmic expression of CD34, but negative expression for other immunohistochemical stains, such as alpha-smooth muscle actin, factor XIIIa, S-100 and melan-A. Additionally, immunostaining with CD34 as a marker helps identify tumour cells at the surgical margins, particularly when treating recurrent DFSP in which tumour cell fascicles are often interspersed with the scar tissue. CD34 expression is not unique to DFSP.[16]

Surgical excision is the standard treatment of DFSP including stages I and II, even III and IV whenever feasible.[16]

The general prognosis for DFSP is excellent. The overall rate of distant metastasis is only 5% and regional metastasis is 1%.[1]

CONCLUSION

DFSP is a cutaneous sarcoma, presented here for its rarity. In our case, the tumour was confined to the skin and subcutaneous tissue of the head and neck region. One of the most challenging areas is the head and neck, with an increased rate of local failure, due to critical structures and aesthetic difficulties in reconstruction. It is a clinically challenging tumour because it is highly invasive and aggressive locally with a high potential for local recurrence if not treated properly. Diagnostic delay or even misdiagnosis is not uncommon due to its indolent nature. Long-term follow-up is strongly recommended to rule out recurrences and metastasis. Hence, a detailed evaluation of the clinical presentation and morphologic features along with IHC are needed to make an accurate diagnosis.

DFSP of the cheek, though uncommon, does occur. Correct diagnosis through routine histopathology is difficult, and therefore IHC is required for proper diagnosis and appropriate management. Awareness of this entity can facilitate prompt diagnosis and proper management of the disease.

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Conflicts of interest

There are no conflicts of interest.

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