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Case Report

Clinical insights into dermatofibrosarcoma protuberans

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ABSTRACT

Dermatofibrosarcoma protuberans (DFSP) is a rare, locally aggressive cutaneous soft tissue sarcoma, it is the second most common skin sarcoma after Kaposi's sarcoma. The cause of DFSP remains unknown. The case of a 54-year-old female patient with a diagnosis of Dermatofibrosarcoma protuberans is presented, displaying a typical clinical presentation. It is characterized by an initial lesion in the form of a reddish spot on the anterior region of the chest, which showed slow growth until the development of a multinodular and irregular lesion with multiple recurrences. The lesion is resected, confined to the superficial layers of the skin, with 3 cm margins, confirming the histopathological diagnosis of dermatofibrosarcoma protuberans with clear margins. DFSP is an uncommon cutaneous sarcoma that is typically low- to intermediate-grade, and while it has a limited likelihood of metastasis, it exhibits a notable tendency for local recurrence. The risk of recurrence is closely linked to the extent of surgical resection.

Keywords: Dermatofibrosarcoma protuberans, Cutaneous soft tissue sarcoma, Mohs micrographic surgery

INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a relatively uncommon yet locally aggressive cutaneous soft tissue sarcoma. It poses diagnostic challenges due to its varied clinical presentation and histological characteristics. This article provides an in-depth overview of DFSP, its etiology, clinical features, diagnosis, and treatment options. By shedding light on this rare malignancy, healthcare professionals can enhance their understanding and clinical management of DFSP.

CASE REPORT

A 54-year-old female patient sought medical evaluation in 2007 after noticing a lesion in the infraclavicular region. Initially, this lesion appeared as a red spot and received topical treatment with antifungals and corticosteroids, but showed no improvement. The lesion continued to grow slowly for 15 months, changing in appearance to become bulky and multilobulated, measuring approximately 2 cm.

She was referred to the surgical oncology service due to the observed characteristics, leading to the decision to perform an excisional biopsy with wide margins on suspicion of dermatofibrosarcoma. The histopathological study confirmed clear margins and corroborated the diagnosis.

She experienced recurrences in 2012, 2016, and 2020, receiving surgical treatment on each occasion, supplemented with imatinib. Due to these multiple recurrences, it was determined that extension studies were necessary. The tomography ruled out locoregional invasion or metastasis. The most recent recurrence occurred in 2023, presenting a lesion measuring 2.5 cm in the anterior region of the chest, over a scar from a previous surgery. Resection with 3 cm margins was performed. The lesion was located in the skin and subcutaneous cellular

tissue without extension to the muscle plane, confirming the diagnosis of dermatofibrosarcoma protuberans with clear margins through histopathology.

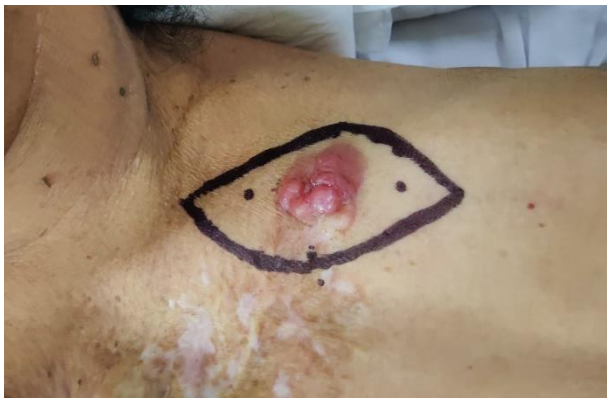

Figure 1: Multilobulated nodular lesion of approximately 02 cm. Compatible with dermatofibrosarcoma.

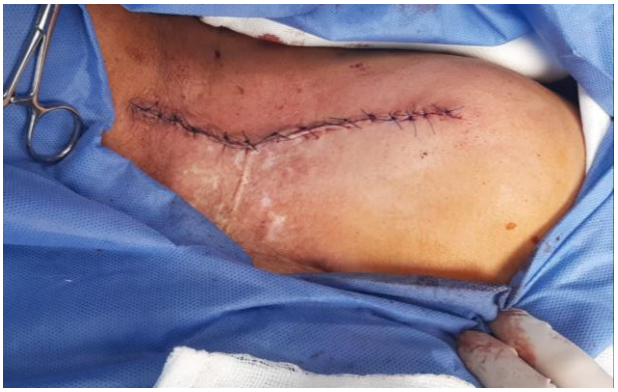

Figure 2: Resection of multilobulated lesion in the anterior region of the thorax. Scars from previous resections are observed.

DISCUSSION

DFSP is a rare, locally aggressive cutaneous soft tissue sarcoma. It has an incidence rate ranging from 0.8 to 4.5 cases per million people per year. DFSP accounts for approximately 1% to 6% of all soft tissue sarcomas and 18% of all cutaneous soft tissue sarcomas.[1] While it is a rare tumor (less than 1 per 100,000 people per year), it is the second most common skin sarcoma after Kaposi's sarcoma.[4] There is some disagreement in studies regarding whether it slightly affects females or males more.[1]

The cause of DFSP remains unknown, although potential risk factors include skin lesions in the affected area, such as existing scars or tattoos.[1] Most DFSP cases occur sporadically, but a subset with unique characteristics, such as multicentricity and early onset, is observed in children with adenosine deaminase-deficient severe combined immunodeficiency (ADA-SCID), a condition characterized by immunodeficiency and faulty DNA repair mechanisms.[4]

The cellular origin of DFSP is not yet determined, but possible candidates include histiocytes, fibroblasts, and dendritic cells. It is currently categorized among fibroblastic-myofibroblastic soft tissue tumors.[4] The prevailing belief is that the mesenchymal tumor originates from a dermal stem cell or an undifferentiated mesenchymal cell with fibroblastic, muscular, and neurological traits.[1]

Cytogenetic and molecular studies have shown that over 90% of DFSP cases are characterized by either supernumerary ring chromosomes derived from chromosomes 17 and 22 or a chromosomal translocation t(17;22)(q22;q13). This results in the fusion of the COL1A1 gene on chromosome 17q22 and the PDGFB gene on chromosome 22q13.[2]

Most DFSP cases occur sporadically. In its very early clinical stage, DFSP presents as a non-protuberant (flat) lesion, often pink or violet in color, which gradually evolves into the typical protuberant form.[4] Dermatofibrosarcoma protuberans usually appears as an asymptomatic, skin-colored to red-brown indurated plaque that eventually develops multiple raised violaceous to red-brown nodules. These growths are slow and can reach several centimeters in diameter.[1] Particularly in its early stages, DFSP can resemble keloids or dermatofibromas, leading to frequent misdiagnosis. As they grow larger, some can ulcerate and become painful. Most DFSPs occur on the trunk (50%), followed by the extremities (35%), and then the head and neck (15%). The shoulder and pelvic regions are particularly characteristic areas for dermatofibrosarcoma protuberans. Normally, DFSP extends into subcutaneous fat but rarely affects the fascia, muscle, or bone unless it has been long standing or recurrent.[1]

Distant metastasis is infrequent, occurring in 1% to 4% of cases and usually only after multiple local recurrences. The lung is the most common site of metastasis through hematogenous spread. Regional lymph nodes are seldom involved. The fibrosarcomatous variant of dermatofibrosarcoma protuberans has a higher risk of local recurrence (14% to 52%) and distant metastases (8% to 29%).[1]

DFSP should be considered in patients with a history of a firm, slow-growing cutaneous nodule. Dermoscopy findings can aid in suspecting DFSP, although a core needle or incisional biopsy is necessary for a definitive diagnosis.[5] The dermoscopic pattern of DFSP often consists of a pink background, depigmented areas without structure, light brown areas without structure, and mostly linear and arboriform vessels in fully developed plaques. Bright white veins and a fine pigment network may also be present, though these findings are not exclusive to DFSP and may sometimes resemble morpheaform basal cell carcinoma.[1,3]

Classic DFSP is a low-grade malignancy that typically infiltrates the dermis and subcutis extensively. Tumor cells grow along the fibrous septa of the subcutaneous tissue and intercalate with fat lobules, creating a honeycomb-like (or storiform) pattern. DFSP is composed of cytologically uniform spindled cells with minimal cytological atypia and typically low mitotic activity. Rarely, DFSP infiltrates deep soft tissues.[4] In cases of recurrent fibrosarcomatous DFSP, a CT scan of the lungs is recommended because the lungs are the most common site of metastasis.[3]

The primary treatment for dermatofibrosarcoma protuberans is surgical excision, ideally using Mohs micrographic surgery (MMS), a surgical technique that ensures complete histopathological control of margins during surgery. MMS is favored over wide local excision due to DFSP's tendency to have unpredictable subclinical extension. In specific situations or when MMS is unavailable, a wide local excision with 2 to 4 cm margins can be performed. However, local recurrence is relatively common with wide local excision, even with clear surgical margins.[1,4]

Considering that the COL1A1-PDGFB fusion leads to increased PDGFB signaling through an autocrine activation loop, targeted therapy has demonstrated significant antitumor activity. Notably, the tyrosine kinase inhibitor imatinib4 is currently FDA-approved for unresectable recurrent and metastatic DFSP. Imatinib has also been proposed for neoadjuvant treatment of large primary lesions, given its 50% overall tumor response rate, which supports less invasive surgery.[4] In cases of metastatic DFSP, systemic treatment with tyrosine kinase inhibitors, such as imatinib, may be necessary. Imatinib can also serve as a second-line option for unresectable lesions.[3]

The overall prognosis for dermatofibrosarcoma protuberans is generally favorable, with a 10-year survival rate of 99.1%. Since metastasis is rare, local recurrence poses a more common challenge. Risk factors for local recurrence include age over 50, high mitotic index, increased cellularity, Black race, male gender, and location on the head, neck, or limbs.[1,3] Given the elevated risk of local recurrence, it is recommended to conduct follow-up examinations every 6 months for at least 5 years.[3]

CONCLUSION

Dermatofibrosarcoma protuberans, although rare, presents unique clinical and histopathological features that demand careful consideration. Accurate diagnosis and prompt treatment are pivotal in achieving successful outcomes for patients with DFSP. The utilization of Mohs micrographic surgery and targeted therapies like imatinib has significantly improved the management of this condition. As our understanding of DFSP continues to evolve, ongoing research and clinical vigilance are crucial in further enhancing the prognosis and quality of life for those affected by this unusual soft tissue sarcoma.

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REFERENCES

1. Brooks J, Ramsey ML. Dermatofibrosarcoma Protuberans. In: StatPearls. Treasure Island (FL): StatPearls Publishing. 2023.
2. Hao X, Billings SD, Wu F, Stultz TW, Procop GW, Mirkin G, et al. Dermatofibrosarcoma protuberans: Update on the diagnosis and treatment. J Clin Med. 2020;9(6):1752.
3. Apalla Z, Liopyris K, Kyrmanidou E, Fotiadou C, Sgouros D, Patsatsi A, et al. Clinical and dermoscopic characteristics of cutaneous sarcomas: A literature review. Diagnostics (Basel). 2023;13(10):1822.
4. Mocellin S. Soft tissue tumors: A practical and comprehensive guide to sarcomas and benign neoplasms. Cham: Springer International Publishing. 2021.
5. Bernard J, Poulalhon N, Argenziano G, Debarbieux S, Dalle S, Thomas L. Dermoscopy of dermatofibrosarcoma protuberans: a study of 15 cases. Br J Dermatol. 2013;169(1):85-90.

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