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Case Report

Dermatofibrosarcoma Protuberans in Juvenile: A Rare Case Of Soft Tissue Sarcoma Of The Skin

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ABSTRACT

Introduction and importance: *Dermatofibrosarcoma protuberans (DFSP) is an uncommon soft tissue sarcoma that primarily affects middle-aged adults but can also occur in children. Despite its low metastatic potential, DFSP displays aggressive local behavior and a high local recurrence rate. Diagnosis in children can be delayed due to confusion with other skin conditions. This case report aims to discuss the case management of DFSP in juvenile female.* ***Presentation of case:*** *A 13-year-old female patient presented with a progressively enlarging lump on the left eyelid, initially resembling small brown spots that coalesced over 7 months. Intermittent bleeding and pain were reported. Physical examination revealed a dark brown mass in the left frontotemporal region, with solid, non-fragile characteristics. Clinical diagnosis suggested a suspicious malignant skin tumor, confirmed as basal cell carcinoma.*
Treatment involved wide excision and flap reconstruction. Postoperative care included infusion, antibiotic injection, and pain management. The patient's prognosis was favorable, though functional impairment was possible. Follow-up revealed histopathological findings of pigmented DFSP. ***Discussion:*** *DFSP is a rare but significant malignancy that poses diagnostic challenges due to its varied clinical presentations and*

histopathological features. While primarily affecting young to middle-aged individuals, it can also manifest in pediatric populations. Diagnosis typically involves histopathological assessment, confirming characteristic spindle cell proliferation with CD34 expression. Surgical excision remains the cornerstone of treatment, complemented by adjuvant therapies like imatinib mesylate or radiation in select cases. Regular surveillance, comprising frequent evaluations in the initial post-excision period followed by yearly monitoring, is imperative for promptly detecting any recurrence or metastasis. ***Conclusions:*** *The structured approach in DFSP ensures optimal patient management and outcomes. Surgical management with Mohs micrographic surgery or Wide local excision is the gold standard in treating DFSP.*

1. INTRODUCTION

Soft tissue sarcomas are an uncommon group of tumors, comprising less than 1% of all malignant tumors. Dermatofibrosarcoma protuberans (DFSP) is a rare soft tissue sarcoma of the skin. DFSP is a low- to intermediate-grade and slow-growing malignant sarcoma primarily affecting middle-aged adults. Originally termed keloid sarcoma, it was later renamed DFSP in 1925 by Hoffman. DFSP originates in the dermis but can infiltrate underlying tissues. Despite its low metastatic potential, DFSP has a high rate of local recurrence.[1]

DFSP, though the most common cutaneous sarcoma, accounts for less than 0.1% of all malignant neoplasms and 1% of soft tissue sarcomas. Its estimated annual incidence in the United States ranges from 4.2 to 4.5 cases per million individuals. Despite its locally aggressive nature, recurrence rates range from 10% to 60%, attributed to its irregular morphology with frequent finger-like extensions. While recurrence is common, regional (1% of cases) or distant metastasis (4%-5% of cases) is rare. DFSP predominantly affects the trunk (42%-72% of cases), followed by the extremities (20%-30% of cases), and the head and neck (10%-16% of cases). These lesions often occur at sites of previous trauma, including burn scars, surgical scars, vaccination sites, and radiation dermatitis.[2]

DFSP in the pediatric population has an aggressive local behavior. Diagnosis is often delayed due to confusion with other skin conditions, particularly in the early stages. DFSP typically presents as a slowly enlarging, rubbery lump, evolving in size, color, and consistency over time. In rare instances, DFSP may manifest as either a depressed area of skin or an ulcerated exophytic lesion, further complicating the diagnostic process.[3]

Histopathologically, DFSP is characterized by uniform spindle cell fascicles arranged in a storiform pattern and exhibits strong CD34 immunoreactivity. However, its histological features overlap with other benign and malignant lesions, necessitating careful differentiation. Ultrastructurally, DFSP displays spindle or stellate cells with ramified processes resembling dermal dendrocytes. Cytogenetically, most DFSP cases

feature a characteristic t(17;22)(q22;q13) translocation, resulting in COL1A1-PDGFB fusion transcripts.[4]

Early suspicion and referral to specialized units proficient in oncologic resection and reconstruction are crucial in cases of DFSP. DFSP originates in the dermis and grows with finger-like projections, necessitating meticulous excision techniques to ensure complete removal, especially in aesthetically or functionally sensitive areas.[5] This case report aims to discuss the case management of DFSP in a 13-year-old female.

2. CASE PRESENTATION

A 13-year-old female patient was presented to the Surgical Polyclinic with a chief complaint of a progressively enlarging lump on the left eyelid. Initially, there were small brown spots in the size of green beans, which gradually enlarged and protruded to the size of a marble over the course of 7 months. The patient reported intermittent bleeding from the lump accompanied by pain. No visual impairment was noted, with regular eye-opening and closing movements and an intact blink reflex. The patient denied cough, dyspnea, or epigastric pain. There was no significant past medical history; family history was negative for similar lesions.

The patient was in good general condition with compos mentis during the physical examination. Vital signs were within normal limits, and clinical examination of the head revealed non-anemic conjunctiva and non-icteric sclera. Examination of the thorax, abdomen, and extremities revealed no abnormalities. Local examination of the left frontotemporal region revealed a dark brown mass with raised edges, lacking ulceration. Palpation revealed a solid, non-fragile mass with indistinct borders, an uneven surface, and fixed dimensions measuring 3x2x1 cm. The lesion did not demonstrate a propensity for bleeding. Illustrative representations of the lesion's position and the initial clinical condition were provided in Figure 1.

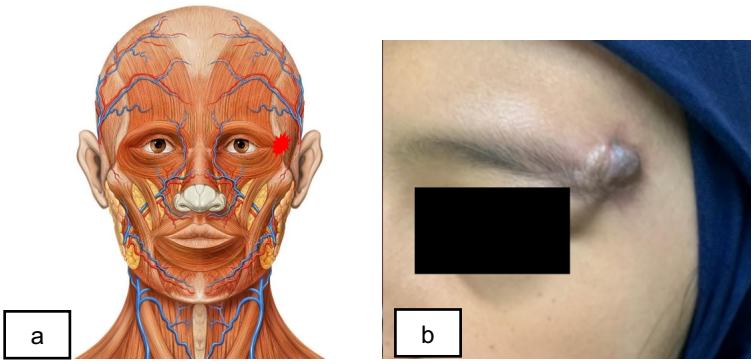

Fig. 1. (A) The position of the lesion on the frontotemporal sinistra region (B) Clinical examination of DFSP

Clinical diagnosis concluded the presence of a suspicious malignant skin tumor in the left frontotemporal region, which had infiltrated the orbicularis oculi muscle, and infiltration into the orbital bone was not yet determined. Histopathological examination revealed suspicious basal cell carcinoma in the left frontotemporal region. The patient was advised to undergo a skull X-ray and biopsy—the treatment plan involved wide excision and flap reconstruction. Postoperative instructions included lactated Ringer's infusion at 20 drops per minute, ceftriaxone injection 2x1g, and administration of

ketorolac 3x30mg. The prognosis was generally favorable, with a possibility of functional impairment.

On postoperative day 1, the patient regained consciousness and was in good general condition. Vital signs were within normal limits. Physical examination of the thorax, abdomen, and extremities revealed no abnormalities. Local examination of the postoperative area showed edema (+), no bleeding, and no pus (Figure 2). The patient was discharged with a prescription for cefadroxil 2x500mg and ibuprofen 3x400mg, and a follow-up appointment was scheduled for 1 week.

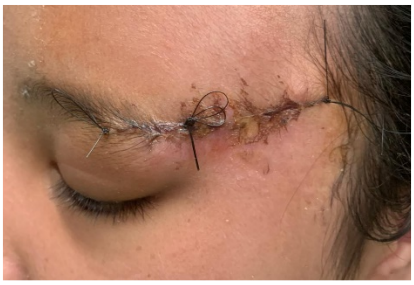

Fig 2. Post-operative (H+1) examination

On postoperative day 6, the patient presented with reduced swelling and dry wounds that felt painful and itchy. Physical examination revealed no abnormalities. Local status in the left frontotemporal region showed tenderness to palpation (+), no bleeding, and no pus. Histopathological examination results concluded pigmented DFSP in the left frontotemporal region, and the surgical wound margins did not contain malignant tumor cells. Histological features of the lesion are presented in Figure 3. No infiltration was found in the orbital bone. The patient was prescribed cefadroxil 2x500mg and ibuprofen 3x400mg, with instructions for a follow-up appointment in 1 week for suture removal.

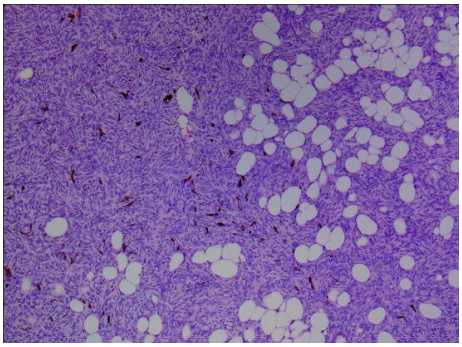

Fig 3. Histological features reveal a tumor mass composed of hyperplastic spindle cells arranged in groups exhibiting a storiform and fascicular pattern. The nuclei of the cells display polymorphism, hyperchromatism, regular nuclear membranes, and mitotic activity. The stroma exhibits hyaline degeneration with vascular dilation.

3. DISCUSSION

In this article, we reported a case of DFSP in a 13-year-old female. DFSP is a slow-growing, malignant skin tumor originating in the dermis and extending into subcutaneous tissues, fascia, bone, and muscle. DFSP can affect individuals of all ages but is most frequently diagnosed between the second and fifth decades of life, with a peak incidence around 40-50 years old. While earlier reports suggested a potential male predominance, recent studies indicate an equal distribution between genders. While

DFSP is rare in both adults and children, it stands as the most common dermal sarcoma. In pediatric cases, DFSP can manifest in the neonatal period or at any later stage, with an estimated incidence of approximately one per million in individuals under 20 years old.[1,6,7]

The diagnosis of DFSP is frequently delayed, often by months to years. The tumor initially presents as asymptomatic, erythematous to violaceous patches, progressing slowly to plaque or papule and then to nodular or tumoral phases. In its early stages, DFSP must be differentiated from hypertrophic scars, keloids, epidermal cysts, lipomas, dermatofibromas, and nodular fasciitis. DFSPs can exhibit radial growth as the disease progresses and become atrophic, sclerotic, ulcerative, and protuberant, eventually invading subcutaneous tissues, fascia, muscles, and bone. Differential diagnosis should include other soft tissue sarcomas, Kaposi sarcoma, pyogenic granuloma, and amelanotic melanoma at later stages.[5]

Clinical suspicion of DFSP is confirmed through histopathologic assessment, as with most solid tumors. Histopathologically, DFSP typically exhibits a storiform or fascicular proliferation of bland spindled cells extending from the dermis into the subcutis. DFSP commonly shows positivity for CD34 and negativity for factor XIIIa. Distinguishing DFSP from dermatofibroma requires immunohistochemical assessment for cathepsin K, apolipoprotein D, nestin, factor XIIIa, and CD34. Routine diagnostic imaging is typically not performed initially unless metastatic disease is suspected.[4]

DFSP tumors typically exhibit free cells between the lesion and the overlying epidermis, with low-to-moderate mitotic activity. Immunohistochemical staining reveals positive expression for CD34 and negative expression for factor XIIIa, aiding in the differential diagnosis of DFSP.[8,9] However, immunohistochemical staining may not always be sensitive, with estimated sensitivity ranging from 84% to 100%. Various histopathologic variants of DFSP have been documented, including granular cell, mixed, Bednar, fibrosarcomatoid, atrophic, myxoid, and sclerosing variants. Approximately 10%-15% of DFSP tumors undergo fibrosarcomatous transformation, often associated with a more aggressive clinical course.[1]

The initial management of DFSP is surgical, requiring careful planning based on tumor size, location, risk for metastasis, and cosmesis considerations. Efforts should be made toward achieving complete resection (R0) during the primary surgery to minimize the risk of recurrence. Histopathologic assessment of all surgical margins is essential, and reconstruction without confirmation of negative margins is discouraged to avoid tumor seeding. If positive margins are identified, re-resection is recommended to achieve clear margins. Mohs micrographic surgery (MMS) is an alternative technique offering high local control rates, mainly when meticulous pathologic margin evaluation is ensured.[2]

Conventional chemotherapy is rarely effective in DFSP treatment; however, imatinib mesylate, a tyrosine kinase inhibitor, has shown clinical activity against DFSP tumors containing t(17;22)(q22;q13) translocation, which results in overexpression of PDGF-B receptor[10,11]. Neoadjuvant imatinib mesylate therapy may be considered for locally advanced or recurrent DFSP cases to reduce tumor burden before surgery[11,12]. Adjuvant radiation therapy is commonly recommended, especially for cases with positive margins or where wide radical excision alone may result in significant cosmetic or functional deficits. Adjuvant irradiation can reduce the risk of recurrence. At the same

time, definitive radiation therapy may improve cosmesis but carries a risk of late toxicities related to functionality, mainly if major joints are included in the radiation field. Frequent follow-up after radiotherapy is crucial, as a subset of DFSP tumors may transform into more aggressive phenotypes. The radiotherapeutic dose varies based on the treatment setting, tumor characteristics, and risk factors for recurrence.[2,4]

Regular surveillance, initially every six months for the first five years post-excision, followed by yearly evaluations, is essential for monitoring DFSP patients, aiming to detect any recurrence or metastasis promptly. This structured monitoring schedule allows for early intervention and appropriate management, ensuring optimal patient outcomes by promptly addressing any signs of disease progression.[13]

4. CONCLUSION

DFSP is a low to intermediate-grade malignancy commonly found in young to middle-aged individuals. It exhibits histological features of bland spindle cells in a storiform pattern. Wide local excision is the typical approach for DFSPs on the trunk and extremities, while MMS may be suitable for smaller lesions in sensitive areas. Adjuvant therapies like targeted therapy and radiation are options for patients ineligible for surgical excision. Regular surveillance, initially every six months for five years post-excision, followed by yearly evaluations, is essential for early detection of recurrence or metastasis.

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Conflict of Interest Statement:

The author declares that the case report was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.