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DERMATOFIBROSARCOMA PROTRUBERANS-A RARE ENTITY AND INFREQUENT DIAGNOSIS

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ABSTRACT

Dermatofibrosarcoma protuberans (DFSP) is a rare slow developing sarcoma of soft tissue which has significant local recurrence and infrequent distant metastasis. It makes up 0.1% of all malignancies and 1% of all sarcomas. The vast majority of around 90% of DFSP are low grade sarcomas, whereas the remainder are classified as intermediate or high grade because of the presence of high grade fibrosarcomatous component. Although the disease can affect people of any age, DFSP most frequently affects patients in their mid- to late-30s. Blacks have a 1.5 to 2 fold higher incidence than whites. Both men and women are equally affected. The literature search revealed that both non-congenital mutation as well as trauma serve a role in the development of this dermal neoplasm.

INTRODUCTION:

Darier and Fernandes first described dermatofibrosarcoma protuberans (DFSP) in 1924, but Hoffman first defined the term "DFSP" in 1925. This soft tissue sarcoma has a high local recurrence rate and a low rate of distant metastasis. It extends into the fascia planes and through the dermis and subcutaneous tissue. The distinctive feature for high local recurrence is related to the existence of tentacle like extensions from the main tumor mass termed pseudopods. Therefore, wide local excision with sufficient margins is the preferred course of action. Recurrence is a characteristic of DFSP when excision is insufficient. DFSP most typically found in young people on the trunk and limbs. Children with severe combined immunodeficiency (ADA-SCID) that lacks adenosine deaminase typically experience it sporadically.

The trunk is where DFSP is most frequently found, followed by the proximal extremities, head, and neck. The list of DFSP sites also includes bug bites, old burns, trauma, radiation dermatitis, vaccination sites, and puncture sites for central venous lines. Although the head and neck region is a potential site, it rarely happens there.

Case Report:

A 34 years old male came to surgical outpatient department with a large protuberant mass in the lower back on the right side for about two years. The patient reported the mass is initially small and it gradually increases our the course of time and attained the present size. There was no rapid increase in size and no ulceration and not associated with any pain or bleeding into the mass and not associated with any trauma. The patient denied any recent weight loss, fever ,night sweats or chills.

On physical examination ,there was a firm mass which is painless and multinodular and shows induration to the underlying subcutaneous tissue with no signs of localised heat or redness. There was no palpable inguinal lymph nodes. There was neither personal nor familial history of malignancy.

A soft tissue high frequency ultrasound was performed which shows poorly defined, heterogenous cutaneous tumor, measuring 4x4 cm .

Magnetic resonance imaging (MRI) demonstrated an 4.4x3.7x3.0 cm heterogenous tumor with peripheral enhancement, extending into the subcutaneous layers but without infiltration of the adjacent muscular or bony structures.

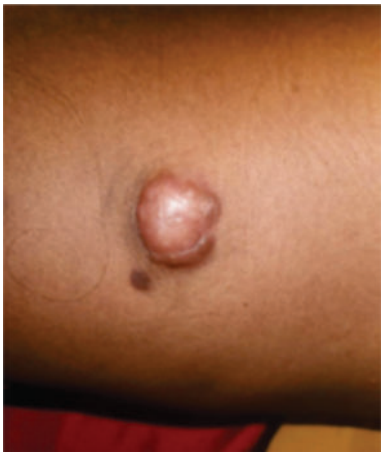

Image showing the mass in the lower back with surrounding skin appears normal and no ulceration and bleeding.

With the heterogeneous nature of the tumor, a soft tissue adnexal tumor was suspected and patient underwent wide local excision in the operation theatre under tumescence and specimen sent for histopathological examination. A well circumscribed tumor with hematoxylin and eosin stained sections showed a cellular spindle cell neoplasm with vague cellular borders and relatively uniform elongated nuclei in storiform pattern.

On immuno histochemical stains, the spindle cells shows diffuse positivity for vimentin and CD34 antigen. There was no positivity for smooth muscle actin ,Desmond, s100protein, CD68, Cd57, CD 117, and keratin 8\18. Based on the histological and Immunohistochemicsl findings,the diagnosis of DFSP was made.

Owing to the adequacy of wide margin in three dimensions there was no additional margin excision needed. Post operative course was uneventful and he was discharged on the second post operative day after being referred for adjuvant radiotherapy if required. Twenty months after surgery , no local recurrence is evident.

Figure 2: showing post operative excision of tumor with adequate margin on all dimesions

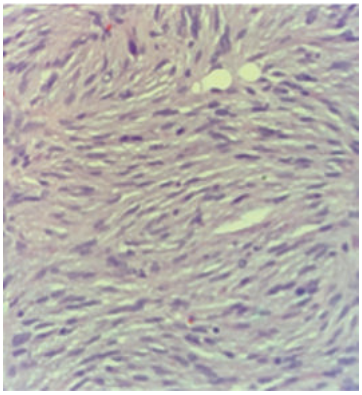

Figure3: shows spindle shaped cells in storiform pattern and with adipose subcutaneous tissue

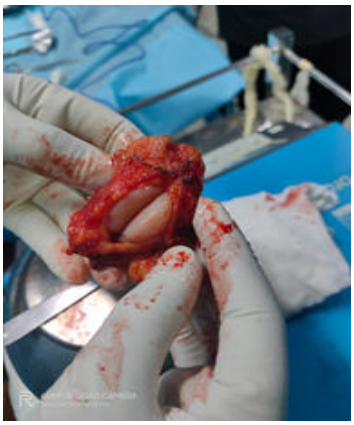

Figure 4; shows cut section of tumor shows homogenous soft tissue nature with in the lesion.

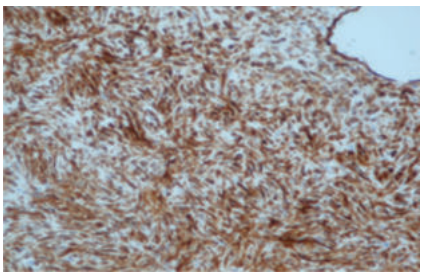

Figure 5: immunohistochemistry of the tumor cells showing diffuse positivity for CD34 antigen.

DISCUSSION:

Less than 0.1% of all malignant neoplasms are malignant fibroblastic mesenchymal skin tumours, such as DFSP, are slow-growing. Although the pathophysiology of DFSP is not well known, it may be influenced by a variety of factors, including an abnormal tumour suppressor gene (TSG) or a history of localised trauma or scarring. It has nothing to do with prior UV exposure. The translocation between chromosomes 17 and 22 that causes the fusion of the collage type I alpha 1 (COL1A1) and the platelet derived growth factor (PDGF) beta chain gene (PDGFB) is present in more than 90% of DFSP cases. Thus the expansion of DFSP is a result to the dysregulation of PDGF B-CHAIN expression and activation of the PDGF receptor (PDGFR) protein tyrosine kinase.

Clinical and pathological features:

DFSP classically presents as a solitary, frequently asymptomatic,multilobulated plaque with a pink and violacious hue. Lesions may be subtle and are occasionally mistaken for scars , with a deep induration palpable on exam. Clinically, it may be difficult to differentiate from a dermatofibrosarcoma or a keloid. The tumor exhibits slow but continual growth, and lesions may be present for years prior to definitive diagnosis. Most commonly affected sides are trunk and less frequently the extremities ,head and neck but it may occur anywhere. BEDNAR tumor is a rare pigmented variant of DFSP.

Histologically, DFSP arises in the dermis and is composed of monomorphous, dense spindle cells arranged In a storiform pattern and embedded in a sparse to moderately dense fibrous stroma. Deep invasion into the subcutis is common and may account for the high incidence of local recurrence after excision.

Immunohistochemistry shows strong positive for CD34 and vimentin and negative for S100,desman and SMA. Fibrosarcomatous variant of DFSO shows reduced expression of CD34, but nesting levels remains unchanged and staining for nestin can be used in conjunction with CD34 for diagnosis.

Staging:

DFSP is prone to local recurrence but distance metastasis is rare, as is mortality. There is no universally accepted staging system for this rare tumor. AJCC 8th edition groups DFSP with other soft tissue sarcoma. Nearly localised DFSP as stage 1 disease with low grade histologic features. The fibrosarcomatous variant of DFSP is an uncommon but well recognised variant of DFSP that tends to follow a more aggressive clinical course, and these lesions with higher grade histological features are classified as stageII or III, depending on size of the primary tumor. Rare cases of regional or distant metastasis are classified as stage IV.

The lack of epidermal hyperplasia, relative cellular homogeneity, a lesser amount of collagenous matrix and diffuse subcutaneous infiltration distinguish DFSP from benign and cellular fibrous histiocytoma (dermato fibrosarcoma).

Treatment of choice is wide local excision with negative margins of 3+5cm from the tumor edge including the skin, the subcutaneous tissue, and the underlying fascia. In cases with possible bone involvement, the periosteum or even a portion of the bone may also need to be excised to achieve negative resection margins. In series where resection margins of five cms were used , recurrence rates were less than 5%. Reconstructive surgery may be required to restore tissue defects after excision using a local skin flap, skin graft or myocutaneous flap. In our case, a local skin flap reconstruction was chosen.

An alternative to wide local excision is MOHs micrographic surgery which is considered by may as the treatment of choice for DFSP. The technique consists of successive horizontal sectioning (5-7um) during resection and immediate frozen microscopic examination until a tumor free

margin is succeeded, There are reports of local cure rates if 93-100%.

Regarding adjuvant treatment, imatinib mesylate, a tyrosine kinase inhibitor, is used in the treatment of un resectable, recurrent and/or metastatic disease. Imatinib inhibits the tyrosine kinase of PDGF and seems effective in treating DFSP in patients with t (17; 22) translocation. 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. Due to the large size of the original tumor we referred our patient for adjuvant radiotherapy, to avoid potential relapse. Post-operative radiotherapy is reported to have a cure rate of 85 %. Combination of conservative excision and adjuvant radiotherapy has demonstrated a reduced local recurrence rate of 5 %.

The recurrence rate is high. Most local recurrence appears in first three postoperative years, with 50% presenting within the first year of surgery. However, recurrence after five years are also reported. Thus, it is important to follow up these patients for long term.

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