Dermatofibrosarcoma protuberans on the leg: pearls in diagnosis and surgical management

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Introduction: Dermatofibrosarcoma protuberans (DFSP) is a rare, slow-growing soft tissue tumor accounting for less than 0.1% of all malignant neoplasms and approximately 1% of all soft tissue sarcomas[1].

Case Summary: We report a case of a 27-year-old female who presented with a 1-year history of a gradually enlarging firm, erythematous to violaceous nodule on the right anterolateral leg. Shave excision biopsy was done, and revealed spindle-shaped fibroblasts arranged in a storiform pattern around an indistinct vasculature. Histopathologic diagnosis revealed dermatofibrosarcoma protuberans. There was prominent, diffuse, positivity for CD34 by immunohistochemistry. The cytogenetic analysis revealed a t(17;22) translocation, confirming our working impression.The patient underwent wide excision with frozen section. All surgical margins of resection were negative for tumor. The patient was referred to plastic surgery for post-excision coverage defect. Split-thickness skin graft was applied over the defect. Patient tolerated the procedure well, and has not experienced recurrence of the tumor.

Conclusion: A protuberant, well-circumscribed tumor that seems benign may incidentally be a sarcoma. Skin biopsy, immunohistochemistry and cytogenetic studies using Fluorescence In-Situ Hybridization (FISH) are the most essential laboratory investigations to validate a diagnosis of DFSP. Proper surgical excision with adequate margins will prevent recurrence of the tumor.

Keywords: dermatofibrosarcoma, tumor, malignancy.

INTRODUCTION

Dermatofibrosarcomma protuberans (DFSP) is a rare, slow-growing soft tissue tumor accounting for less than 0.1% of all malignant neoplasms and approximately 1% of all soft tissue sarcomas. [1]

It is a nodular skin tumor that has a slow growth with a long pre-clinical duration that may likely begin in childhood and manifest clinically during young adulthood.[2] The lesion has a "protuberant" appearance in its fully developed form. The risk of local recurrence correlates well with the extent and adequacy of wide excision. [3]

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CASE REPORT

A 27-year-old female presented at our clinic with a tumor on the lateral aspect of the right leg. Past medical history and family history were unremarkable. The mass was first noticed as a small pink, painless, non-pruritic papule 1 year prior to consult, and slowly enlarged to its present size of 3.0 x 2.0 x 2.0 cm. No lymph node involvement was noted at the time of physical examination.

Shave excision biopsy was done which revealed spindle-shaped fibroblasts arranged in a distinct storiform pattern in the dermis around inconspicuous blood vessels. Minimal nuclear pleomorphism and low mitotic activity were noted. Cytogenetic analysis of the paraffin-embedded specimen revealed a t(17;22) translocation. The translocation involves the COL1A1 gene on chromosome 17 and the platelet derived growth factor B gene on chromosome 22, which validated the diagnosis of dermatofibrosarcoma protuberance.

The patient underwent surgical excision with frozen section. The tumor was excised to include a 2.2-cm margin of grossly normal tissue, and was sent to pathology for frozen section. All surgical margins of resection were negative for tumor. Wide excision of the mass including

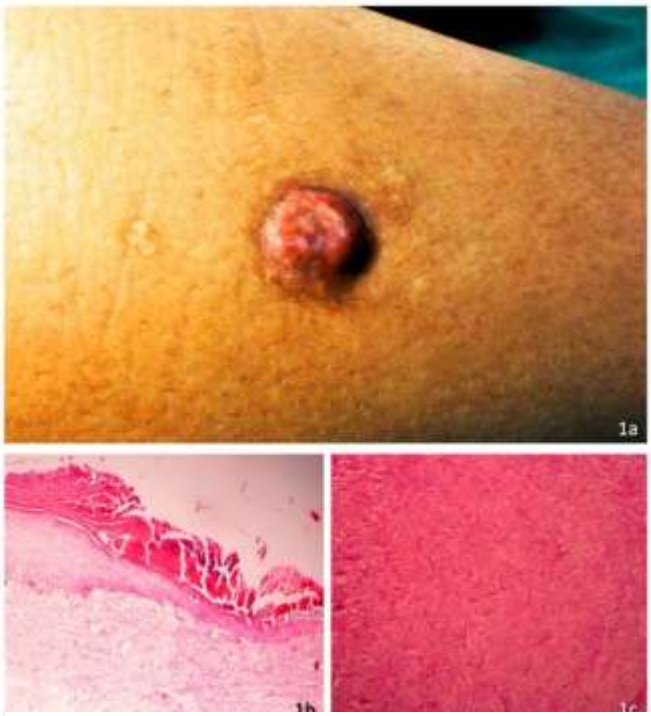

Figure 1a. A solitary erythematous to violaceous nodule measuring 3.0 x 2.1x 2.0 cm on the right anterolateral leg.
Figure 1b. Histopathology revealed proliferation of spindle-shaped cells infiltrating the dermis (H and E, x100)
Figure 1c. Higher magnification shows fibroblasts arranged in a monotonous storiform pattern around inconspicuous blood vessels(H and E, x400)

subcutaneous fat and fascia was done.The patient was also referred to plastic surgery for split-thickness skin graft on the post-excision coverage defect.

DISCUSSION

Microscopically, DFSP diffusely infiltrates the dermis and subcutaneous fat. In the deeper regions, the tumor extends along connective tissue septa and between adnexae. It may come in contact with the lobules of subcutaneous fat creating a lace-like or "honeycomb" look.[4]

The main part of the tumor has slender or spindly fibroblasts arranged in a distinct storiform pattern around a discrete vasculature. There is usually little nuclear pleomorphism with low mitotic activity. DFSP can be distinguished among other fibrohistiocytic tumors due to its significant uniformity and distinct storiform pattern.[5]

DFSP commonly stains positive for CD34+ marker in 20-50% of cases. Cytogenetic studies using Fluorescence In-Situ Hybridization (FISH) is more specific in the sense that it shows a t(17;22)(q22; q13) reciprocal translocation in more than 90% of cases leading to formation of a ring chromosome.[6]

The tumor can locally recur if not adequately excised with proper margins. The risk of local recurrence correlates well with the extent of wide excision.[7] Parker, et al. showed that a 2.5 cm surgical margin through the deep fascia cleared all tumors. DFSP tumors that measured less than 2 cm were completely cleared with a 1.5 cm surgical margin. None of the patients who were excised with a 2.5 cm margin through the deep fascia had a tumor recurrence. [8]Regional and distant metastasis is rare, and usually occurs after multiple recurrences. In those reported to experience metastases, three-fourths had hematogenous spread to the lungs, and one-fourth had lymphatic spread to the regional lymph nodes. Metastasis to the brain, bones and heart have also been documented.[2] Due to the low incidence of regional lymph node metastasis, routine node dissection is not warranted. Radiotherapy is recommended for large, unresectable tumors where repeated surgery may cause mutilation or functional impairment.[9,10]

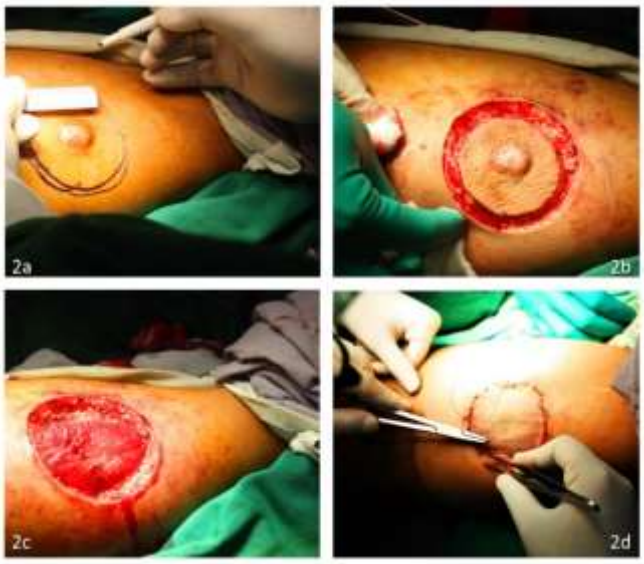

Figure 2: (a & b) Surgical excision of the tumor with a 2.2 cm margin of grossly normal tissue; (c) skin and subcutaneous fat were all removed together with the tumor; (d) Split-thickness skin graft done by plastic surgeon on the post-excision coverage defect

CONCLUSION

A protuberant, well-circumscribed tumor that seems benign may incidentally be a sarcoma. Therefore, it is imperative to do an excisional biopsy with 2.5 cm margins through deep fascia (as recommended by Parker, et al.) to rule out dermatofibrosarcoma protuberans, and to prevent recurrence. A skin biopsy, immunohistochemistry and cytogenetic studies using Fluorescence In-Situ Hybridization

(FISH) validate the diagnosis of DFSP. Metastasis is rare, and usually occurs only after multiple recurrences.

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