Deep dermatofibrosarcoma protuberans: a pitfall in the ultrasonographic diagnosis of lipoma-like subcutaneous lesions

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ABSTRACT

Dermatofibrosarcoma protuberans (DFSP) is an indolent sarcoma known for its propensity for local invasive growth and recurrence. It typically presents as a protuberant tumor mass. Rare nonprotuberant presentations have recently been described; these invariably present as pigmented or depressed plaques. Lesions arising in the subcutaneous compartment and without cutaneous manifestations have rarely been reported or emphasized in the literature. Here we report a case of deep DFSP that lacked discernible epidermal or dermal changes, was mistaken for a lipoma, and localized entirely within the subcutaneous compartment. Ultrasonography may not be useful in differentiating DFSP from benign tumors. In addition, a review of the English language literature revealed that these deep-seated tumors might be more common than originally believed. For this purpose, the current case is presented to raise awareness for DFSP, which can be present entirely in the subcutis without cutaneous manifestations and thus easily overlooked.

KEYWORDS
Cutaneous manifestations
Deep variant
Dermatofibrosarcoma protuberans
Subcutaneous
Ultrasonography

Introduction

Dermatofibrosarcoma protuberans (DFSP) is a common cutaneous sarcoma representing approximately 6% of all malignant tumors with soft tissue involvement.1 Clinically, it is associated with unique cutaneous findings compatible with a tumor of dermal origin, typically presenting as a protruding “protuberant” mass or less commonly as a depressed and/or pigmented plaque.2 While it rarely metastasizes, this indolent tumor has great propensity for deep local invasion. Thus, wide surgical excision or Mohs micrographic surgery with or without radiotherapy are generally considered to be the best treatments. Here we describe a case of DFSP lacking the cutaneous findings it typically associates with, and is located entirely in the subcutaneous compartment. A review of the literature reveals few cases of deep DFSP presenting without cutaneous features.3-5

Case report

A 34-year-old man with a history of a left upper chest wall tumor excised two years ago presented at our clinic with a right upper back mass of two years’ duration. Compression symptoms during sleep were noted in the previous few weeks. No pain, localized heat or redness was otherwise reported. There was neither personal nor familial history of cancer.
Deep dermatofibrosarcoma protuberans revealed a large subcutaneous tumor (Figure 2A) composed of spindle cells forming storiform growth patterns with scattered mitotic figures and entrapped fat cells forming a honeycomb (Figure 2B).

Immunohistochemical study of the sample showed the tumor cells were positive for CD34, and negative for factor XIIIa (Figures 3A and 3B, respectively); a diagnosis of deep DFSP was made. Magnetic resonance imaging (MRI) revealed a large subcutaneous tumor (Figure 2A) composed of spindle cells forming storiform growth patterns with scattered mitotic figures and entrapped fat cells forming a honeycomb (Figure 2B).

Figure 1  (A) A movable (non-fixed) and slightly indurated mass without any epidermal change on the posterior aspect of the right shoulder. (B) Soft tissue ultrasonography revealed a poorly defined heterogeneous subcutaneous tumor measuring 2.17 x 0.74 cm, without posterior enhancement.

Figure 2  (A) The tumor is grossly located in the subcutaneous compartment. (B) Histopathological examination revealed a large subcutaneous tumor composed of spindle cells forming storiform growth patterns with scattered mitotic figures and entrapped fat cells forming a honeycomb (H&E, 200×).

Figure 3  (A) The tumor cells were positive for CD34 (H&E, 400×). (B) The tumor cells were negative for factor XIIIa (H&E, 400×).
revealed post-operatively-enhanced tissue abutting the superficial fascia of supraspinatus tendon but without deep invasion. Chest film, abdominal ultrasonography, computed tomography scan and bone scan failed to reveal distant metastasis. Definitive treatment via wide excision with 2.5 cm lateral margins and a 4.5 cm deep margin combined with subsequent rotational fasciocutaneous flap reconstruction was performed. Subsequent adjuvant radiotherapy was also ongoing.

Discussion

DFSP is believed to be a cutaneous sarcoma of the dermis with potential for deep invasion of subcutaneous structures but rarely metastasizing. It is typically located over the trunk and proximal extremities but may occur on any part of the body. Incidence rate has been estimated to be between 0.8 to 5 cases per million persons per year. This represented approximately 6.2% of the 39,179 malignant soft tissue tumors diagnosed during a 10-year retrospective analysis.

Traditionally, DFSP is considered a cutaneous tumor fixed to the dermis and thus able to move freely over deeper-lying tissue. However, recent reports have shown primary DFSP presenting in sites without dermal involvement. DFSP confined to the subcutaneous layer have also been recognized as “deep DFSP” or “subcutaneous DFSP”. These cases challenges the conventional belief of DFSP as a cutaneous tumor with primary growth in dermis. Further research into the true histogenetic etiology of DFSP is needed to include cell types of the hypodermis and vascular compartments, instead of or in addition to those lineages typically seen in the dermal compartment.

Regardless of tumor location, previous reports of DFSP almost invariably presented with skin surface findings, either due to dermal involvement or secondary epidermal changes. Two primary groups have been elucidated, including “classical” protruding DFSP with elevation or protrusion, and “non-protruders” DFSP with atrophy or depression. A case of pedunculated DFSP clinically believed to be neurofibroma or fibroepithelioma has also been reported. Previously, Ramakrishnan et al reported a lesion without any evidence of skin involvement; they noted a DFSP enveloped in adipose tissue without any dermal infiltration in histopathological analyses of both the core biopsy sample and the excised tumor. This presentation was similar to our case: both DFSP presented entirely without epidermal changes, elevation-depression, erythema, or pigmented alterations on the skin surface and both were confined within the subcutaneous compartment.

Epidermal involvement may be deficient due to the deep location of the tumors, which were 0.4 cm and 1 cm in case of the current and the Ramakrishnan et al study, respectively, since the level of epidermal hyperplasia has been noted to be inversely proportionate to the tumor depth in dermatofibromas and DFSP. Specifically, the critical distance necessary for DFSP-induced epidermal hyperplasia was 0.19±0.16 mm as measured from stromal proliferation to the tip of the dermal papilla. Observation of epidermal hyperplasia was thus proposed to be attributable to cytokine diffusion to the epidermis. Therefore, purely subcutaneously located DFSP presenting without any skin surface changes should not be regarded as coincidental, but should instead be expected to have little cutaneous effect. This lack of epidermal or even dermal changes is particularly alarming, since DFSP are typically diagnosed after biopsy of a clinically suspect lesion.

The true epidemiology of these deep-seated lesions is not known, since they may be present, as in our case, entirely devoid of skin surface changes for many years. This is clinically alarming, since deep lesions would lack the typical clinical presentations of DFSP or other malignant tumors that would otherwise warrant further biopsy. In those unable or not compliant to biopsy, MRI may be an early viable alternative. In a series of 10 DFSP patients who underwent MRI, the results were found to be consistent with other soft tissue sarcomas. Notably, 3 of these 10 cases had tumors primarily located in the subcutis; one of these three was purely subcutaneous after a histopathologic review. Unfortunately, clinical presentation was lacking due to the retrospective nature of the study. Although limited in case number, this study nevertheless raises the possibility that DFSP has high propensity (30%) to be located deep in the dermis.

In 2005, Martin et al reviewed questionnaires from 143 cases of DFSP. Sixty-two were initially nonprotuberant (43%). Of these, 18 (29%) were “morphea-like”, 12 (19%) were “atrophoderma-like” and 26 (42%) were “angioma-like”. The pathological, immunohistochemical and cytogenetic findings of “non-protubers” DFSP and “classical” protuberant DFSP are otherwise identical and believed to be at different stages of the same disease spectrum. Progression from nonprotuberant to protuberant DFSP took an average of 7.6±9.3 years (mean±SD). With its proclivity for deep invasion, subsequent upward growth of subcutaneous DFSP toward the dermis is unlikely to occur to any great extent. The true prevalence of DFSP may therefore be underestimated and lacking a “protubers” morphology. Thus, the current methodology of classification via clinical presentations should be reconsidered.

In clinical settings, ultrasonography is frequently used in early assessment of tumor characteristics. Sonography is an excellent imaging modality to determine the cystic or solid nature of a mass and its anatomic relation to adjoining structures. Masses can be characterized in terms of size, number and vascularity in addition to the presence of calcification or ossification. Because of their heterogeneous but mostly reproducible echogenicity and relatively typical sonographic appearance, subcutaneous lesions of various
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origins, especially fluid cysts, can be easily distinguished from each other. Although lipomas have been classically described as homogeneous and hyperechoic, there are frequent exceptions. As a consequence, the diagnosis of benign lipomatous tumors can often be problematic, since echogenicity corresponds to that of the surrounding fatty tissue and is in case of fibrolipoma, hyperechoic compared to the adjacent tissue or in case of an angiolipoma, hypoechoic. Diagnosis of a lipoma on sonography should be made with caution, because there can be a myriad of malignant or benign fat-containing lesions, including low-grade liposarcoma and angiolipoma which can mimic lipomas. Similarly, ultrasonographic features of DFSP have varied as well. Studies have shown echogenicity to be primarily hyperechoic or mixed hyperechoic, while margins are typically well-defined or irregular with pseudopodia-like protrusions; vascularity, a feature favoring malignancy, has also been varied. Similar to DFSP, lipoma has been shown to vary in echogenicity, margin delineation and tumor location in several publications.

DFSP can be mainly or completely located in the subcutaneous compartment, and thus may be clinically indistinguishable from lipomas or other subcutaneous tumors. Without clinical indicators as to the aggressive nature of these non-cutaneous, asymptomatic, slow-growing and deep-seated DFSP, a dichotomous distinction between these and other more benign lesions may not be readily evident. It is possible that these clinically unidentifiable DFSP are underdiagnosed. Enlargement of any mass regardless of lesion duration should prompt immediate suspicion of malignancy; therefore, aggressive cutaneous biopsy of all progressive lesions is recommended for early diagnosis. Of the available imaging modalities, MRI may aid the diagnostic process, since features of DFSP consistent with soft tissue sarcoma are often seen. Ultrasonography may be insufficient for differentiating between deep DFSP and otherwise benign tumors such as lipoma, but more cases are needed to clarify its role.

Acknowledgment

We would like to thank Dr Meng-Yun Hsieh for her editorial assistance.

References