A pearly nodule on an indurated plaque

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Abstract

We present an 81-year-old man who presented for evaluation of an indurated plaque with an exophytic, pearly, skin-red-brown colored nodule with central ulceration on his chest that evolved over the course of several months and was initially suspected to be basal cell carcinoma. Biopsy demonstrated histological features of dermal spindle cell proliferation in a storiform fashion with CD34 positivity confirming a diagnosis of dermatofibrosarcoma protuberans (DFSP). Dermatofibrosarcoma protuberans are rare, slowly progressive soft tissue sarcomas. The rate of DFSP is greatest among African Americans (8.3/1,000,000), occurring nearly twice as frequently when compared to Caucasians. Aside from race/ethnicity, age, and skin trauma, no specific risk factors are associated with DFSP. Complete excision is curative. Given its pearly skin colored appearance, papular/nodular/atrophic morphology variants, and tendency to form indurated plaques, DFSP may be mistaken for nodular and morpheaform basal cell carcinoma subtypes, as well as a variety of other conditions. This case highlights the importance of maintaining DFSP on the differential diagnosis of slowly progressive nodules and indurated plaques, especially in African Americans.

Keywords: African-American, basal cell carcinoma, dermatofibrosarcoma protuberans, pearly nodule

Introduction

Dermatofibrosarcoma protuberans (DFSP) are rare, slowly progressive tumors characterized by their locally microscopic invasive nature, accelerated nodule stage growth, and high recurrence rate. First described in the early 1900s, its name derives from the protruding nodules that develop within a firm red-violaceous scar-like plaque. It accounts for 1% of all soft tissue sarcomas, with an incidence of approximately 4.1 per million population each year [1]. Dermatofibrosarcoma protuberans is most frequently seen in young to middle-aged adults 30-44 years old [2]. The rate of DFSP is greatest among African Americans (8.3/1,000,000), occurring nearly twice as frequently when compared to Caucasians (4.3/1,000,000), [2]. It is rarer among other ethnic/racial groups: Hispanic (3.4/1,000,000), Asian/Pacific Islander (3.3/1,000,000), and Native American (2.9/1,000,000).

Case Synopsis

An 81-year-old immunocompetent man presented to our dermatology clinic for evaluation of a shiny irritated nodule and a scar-like indurated plaque on his mid-left chest that had been evolving over the course of several months (**Figure 1**). He noted inconsistent use of sunscreen and denied any history

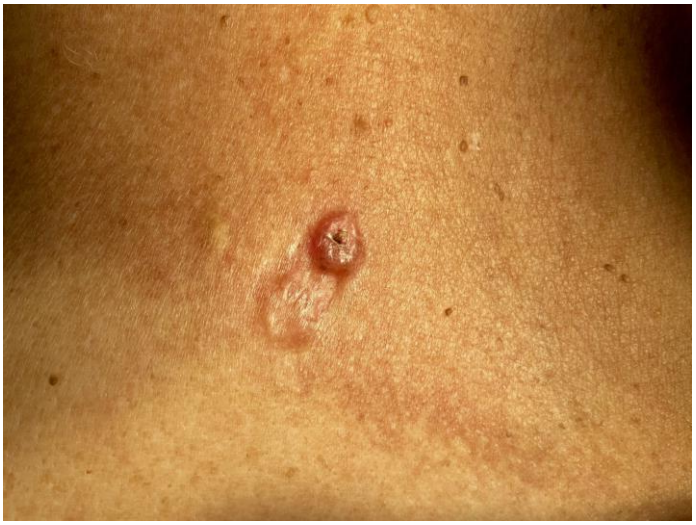

Figure 1. *A singular, exophytic, pearly, skin-red-brown colored nodule with associated telangiectasias and central ulceration adjacent scar-like morpheaform plaque, within a background of hyperpigmented skin in a V neck distribution.*

of trauma, recreational drug use, or personal or family history of skin cancer.

Skin examination demonstrated a singular, exophytic, pearly, skin-red-brown colored 14mm nodule with associated telangiectasias and central ulceration within a background of hyperpigmented skin in a V-neck distribution. A linear 21mm scar-like morpheaform plaque contiguous with the nodule was also noted. There was no evidence of similar lesions elsewhere. Given its characteristic pearly appearance, basal cell carcinoma was suspected. A diagnostic shave biopsy was performed.

Histopathologic sections showed dermal spindle cell proliferation arranged in a storiform/whorled fashion with intermixed sclerotic collagen at the periphery and some areas with myxoid stroma (**Figure 2A, B**). The lesional cells were positive for CD34 (**Figure 2C**), and negative for S100, supporting the diagnosis of DFSP. MRI confirmed the presence of a small, superficial DFSP without subcutaneous extension. The patient underwent Mohs micrographic surgery (MMS) excision of both the nodule and plaque with clear margins.

Case Discussion

Dermatofibrosarcoma protuberans is thought to originate from a dermal stem cell or undifferentiated mesenchymal cell with fibroblastic, muscular, and neurologic features [3]. Dermatofibrosarcoma protuberans tumorigenesis is associated with a translocation between chromosomes 17 and 22, leading to the fusion of *collagen type one alpha 1* (*COL1A1*) and *platelet-derived growth factor subunit beta* (*PDGFB*) genes [3,4]. This fusion results in upregulation of PDGFB protein which promotes DFSP growth through continuous activation of the PDGF receptor beta, a receptor in the tyrosine kinase family [3]. This mechanism justifies the use of tyrosine kinase inhibitors in treatment [4]. Platelet-derived growth factor subunit beta protein also plays an important role in diagnosis of DFSP. Given its essential role in tumorigenesis, it is a highly specific marker for DFSP [4]. Platelet-derived growth factor subunit beta overexpression can be assessed via light microscopy and fluorescent/chromogenic in situ hybridization [5]. However, CD34 and nestin are

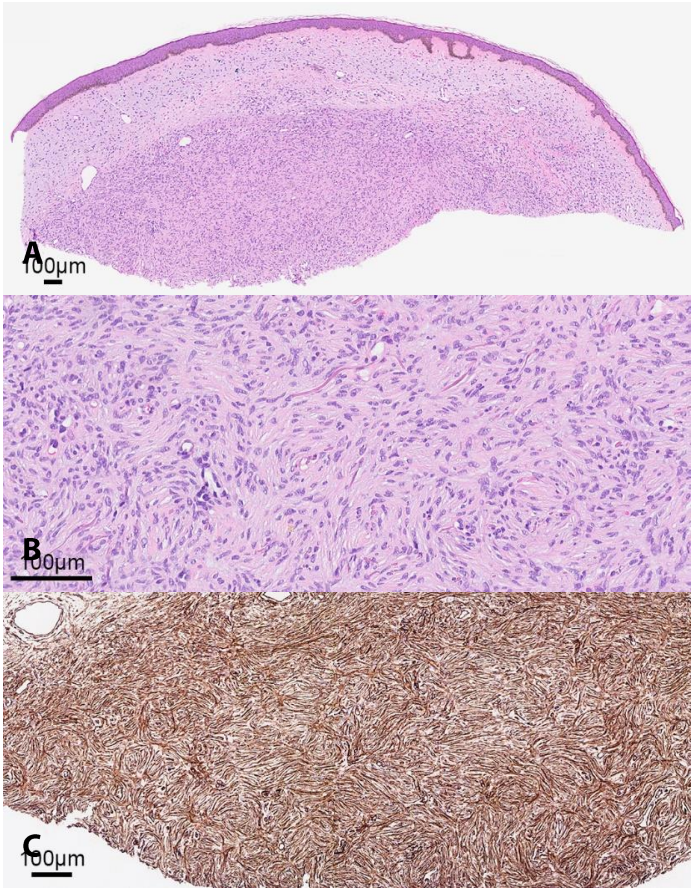

Figure 2. ***A)*** *Histopathologic sections revealed a dermal spindle cell proliferation. H&E, 4×.* ***B)*** *Lesional cells were arranged in a storiform/whorled fashion with intermixed sclerotic collagen at the periphery. H&E, 20×.* ***C)*** *An immunohistochemical study revealed the lesional cells to be positive for CD34, 10×.*

more commonly used DFSP markers for tissue sample screening [4,6,7].

Dermatofibrosarcoma protuberans typically begins as a painless flat, violet or pink plaque that usually progresses to incorporate a nodular component [8]. It is slow-growing and nodule development often follows long periods during which the plaque appears stable in size. Over time lesions may ulcerate and bleed. Its indolent course is often characterized by a high rate of local recurrence because of its infiltrative behavior, but low metastatic potential. Complications like metastasis are rare (less than 5% of cases) but most commonly involve regional lymph nodes and the lungs [4]. Aside from race/ethnicity and age, skin trauma is a risk factor for DFSP. A study of 364 patients with DFSP revealed that 10% of tumors developed at sites of previous punch biopsies, shave biopsies, minor excisions, or major surgical scars [9]. Furthermore, DFSP has been noted to occur at sites of burns, immunizations, and tattoos [9]. It is reasonable to hypothesize chronic inflammation could lead to the malignant transformation of scars in DFSP [9]. Dermatofibrosarcoma protuberans has an anatomic proclivity for the trunk, where it occurs in almost 50% of cases [2]. Less frequently it occurs on the upper/lower limbs, face, head, and neck. In rare circumstances, it has been reported on the genitals and oral mucosa.

The differential diagnosis for DFSP is broad depending on the morphology, location, and size of the tumor. Nodular DFSP may resemble nodular basal cell carcinoma, dermatofibroma, neurofibroma, leiomyoma, epidermal cyst, malignant melanoma, keloid, desmoid tumor, lipoma, nodular fasciitis, sarcoidosis, and other cutaneous soft tissue sarcomas [10,11]. Plaque or atrophic DFSP may clinically resemble morphea, morpheaform basal cell carcinoma, anetoderma, or a hypertrophic scar [12]. It is difficult to distinguish between these diagnoses on examination. Therefore, diagnosis is made via histology.

Treatment most commonly involves wide local excision; however, given its storiform growth pattern, incomplete excisions are common. Mohs micrographic surgery has a lower rate of recurrence when compared to conventional surgery given its micrographic advantage of viewing all tumor margins. When surgical resection with adequate margins is not possible, radiation therapy can be used in combination with resection, or as adjuvant therapy after resection in recurrent DFSP [4]. Additionally, for metastatic disease or large tumors in cosmetically sensitive locations, systemic therapy can be utilized [4]. Tyrosine kinase inhibitors have demonstrated efficacy in treating refractory DFPS [13].

Proposed frequency of follow-up may depend on characteristics like location, size, and margin clearance to stratify DFSP patients from low-high risk groups [14]. In general, the risk of recurrence is greatest within three years of excision [3]. Thus, patients should be evaluated every three to 6 months during this period and at least annually thereafter.

Our case demonstrates a typical example of DFSP. It occurred as a slow-growing plaque on the trunk and was diagnosed after skin biopsy with histological staining. MRI was used to assess for extension and plan for excision of our patient's DFSP. Although this is not routinely done, MRI, CT, and ultrasound are imaging modalities that can be used to assess for obvious extension especially when metastasis is suspected, which can facilitate staging and surgical planning. Given no signs of spread on imaging, MMS was performed with clear margins on our patient. Frequent monitoring visits were scheduled every 6 months to ensure no recurrence.

Conclusion

Dermatofibrosarcoma protuberans is an interesting and rare diagnosis, yet important given its potentially harmful characteristics. However, clinical diagnosis is often a challenge given the non-specific features and infrequent occurrence. This case highlights the importance of maintaining a low threshold for skin biopsy among providers encountering irregular lesions and maintaining DFSP on the differential of slowly progressive nodules and indurated plaques, especially in African Americans.

Potential conflicts of interest

The authors declare no conflicts of interest.

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