Dermatofibrosarcoma Protuberans with Fibrosarcomatous Change: A Case Report of Rare Entity

Chinathip Theerawattanawit MD, Rachot Wongjirattikarn MD, Jade Wititsuwannakul MD.

Division of Dermatology, Department of Medicine, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand.

ABSTRACT:

Dermatofibrosarcoma protuberans (DFSP) is a rare malignancy affecting the skin and underlying soft tissues. Typically characterized by a favorable prognosis with low metastatic and recurrence rates, DFSP can manifest as a subtype with an unfavorable outcome known as DFSP with fibrosarcomatous changes (DFSP-FS). Histologic examination reveals DFSP-FS to exhibit more atypical cellular features and increased mitotic activity compared to the classic DFSP variant. In this report, we detail the case of a 48-year-old Thai female patient presenting with a progressively enlarging mass on the lumbosacral area. Pathological and immunohistochemical analyses confirmed the diagnosis of DFSP-FS. This case presents the rarity of DFSP-FS and emphasizes its distinctive pathological and immunohistochemical characteristics.

Key words: Dermatofibrosarcoma protuberans,
Dermatofibrosarcoma protuberans with fibrosarcomatous changes

Introduction

Dermatofibrosarcoma protuberans (DFSP) is a rare malignancy affecting soft tissues, with an annual incidence ranging from 0.8 to 4.2 cases per million[1]. This condition primarily affects the dermis and subcutaneous tissue, potentially extending to the underlying fascia, muscles, and bones in advanced stages. DFSP typically manifests as a painless, gradually enlarging, erythematous plaque during the third to fifth decades of life. Commonly found on the trunk, proximal extremities, or the head and neck area[2]. The majority of DFSP cases exhibit a chromosomal translocation involving chromosomes 17 and 22, believed to be central to its pathogenesis. This translocation results in the fusion of the platelet-derived growth factor-beta (PDGFB) gene and the collagen type 1A1 (COL1A1) gene, leading to the upregulation of PDGFB. Consequently, this activation of the PDGFB receptor triggers cellular proliferation and the formation of tumors[3].

Corresponding Author: Jade Wititsuwannakul MD., email: jademdcu@gmail.com

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The diagnosis of DFSP primarily relies on histopathology, revealing densely packed and uniform spindle-shaped tumor cells arranged in a storiform pattern, with low mitotic activity and cytologic atypia in the dermis. This pattern may extend into the subcutaneous tissue or underlying structures. Immunohistochemistry of DFSP typically indicates positivity for CD34[1]. However, there exists a rare variant with CD34 negativity known as DFSP with fibrosarcomatous changes (DFSP-FS) [4].

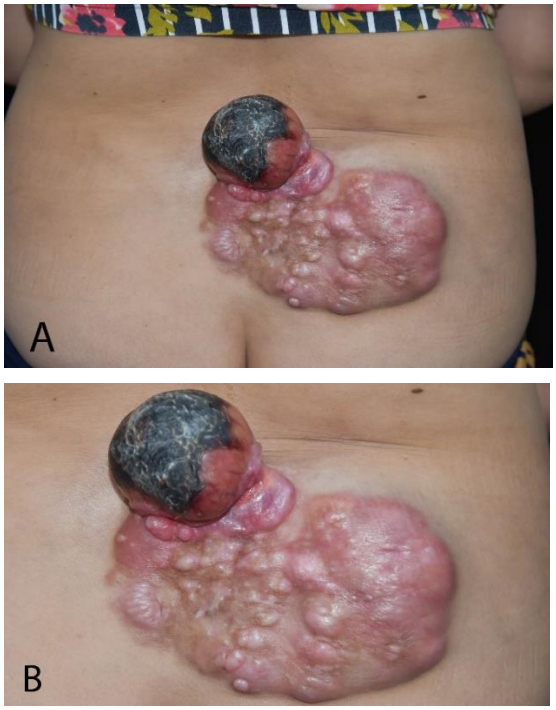

Figure 1 An erythematous to hyperpigmented infiltrative plaque was observed, accompanied by erythematous and some blackish pedunculate nodules on the lumbosacral area of the patient (A,B)

DFSP-FS represents an uncommon histologic subtype of DFSP. Despite its clinical presentation similarities to conventional DFSP, the histopathology of DFSP-FS is characterized by increased cellularity, cytologic atypia, and heightened mitotic activity. Notably, DFSP-FS exhibits areas of CD34-negative spindle cells arranged in a fascicular (herringbone) pattern, interspersed with classic DFSP areas[2]. In this report, we present a case of DFSP-FS, emphasizing its distinctive histopathological characteristics, immunohistochemistry features, and unfavorable clinical outcomes.

Case presentation

A 48-year-old Thai female patient presented with a painless, progressively enlarging mass on the lumbosacral area over the course of 10 years. She denied any history of bowel habit changes or abnormal menstruation. Upon physical examination, an erythematous to hyperpigmented infiltrative plaque was observed, accompanied by erythematous and some blackish pedunculate nodules on the lumbosacral area (Figure 1A and 1B). No hepatosplenomegaly or lymphadenopathy was detected.

Incisional biopsies were performed on both the pedunculate nodule and plaque components. Histological examination from both sites revealed a proliferation of wavy and spindle-shaped cells arranged in a storiform pattern in the dermis interspersing with fascicular and herringbone patterns, extending into the subcutaneous tissue and forming a honeycomb pattern (Figure 2A-F). Immunohistochemistry demonstrated diffuse positivity for CD34 with decreased intensity in the herringbone area (Figure 3A-B). Other stains, including AE1/AE3, EMA, S-100, SOX10, TLE1, Desmin, and SMA, were negative. The diagnosis of DFSP-FS was established.

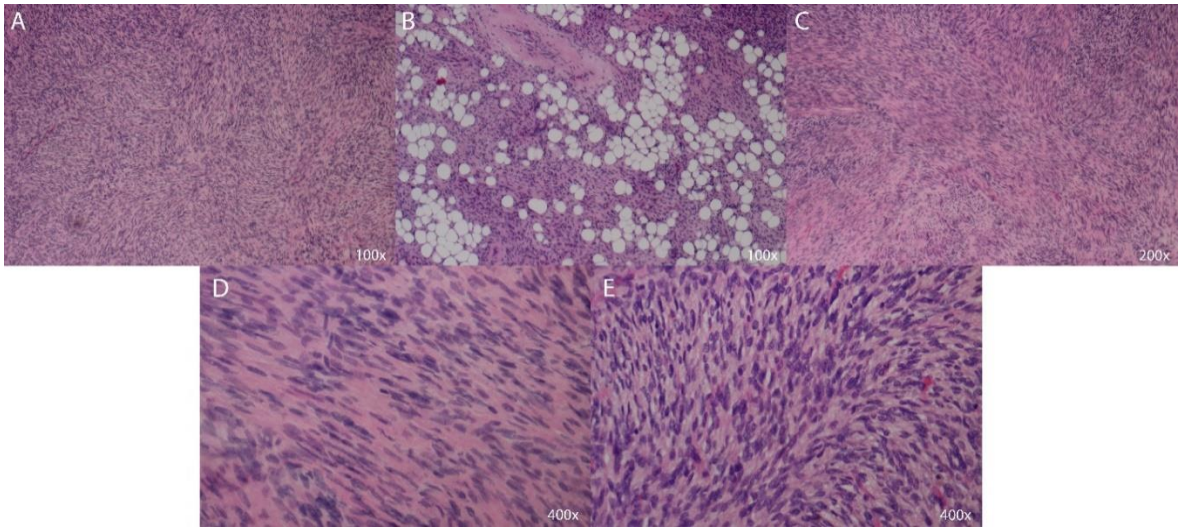

Figure 2 Histopathology from nodule lesion showed proliferation of wavy and spindle-shaped cells arranged in a storiform pattern (A), honeycomb pattern (B) and herringbone pattern (C) on lower magnification. On higher magnification, proliferation of uniform spindle-shaped tumor cells (D) in the storiform area and polymorphic spindle cell with nuclear polymorphism and mitosis in the herringbone area (E)

Figure 3 CD34 positive spindle cell admixed with area of CD34 negative spindle cell in herringbone pattern (A). Higher magnification showed CD34 positive spindle cell (B) and CD34 negative spindle cell (C)

A contrast-enhanced computerized tomography scan of the abdomen revealed a 14 cm lobulated heterogeneous enhancing mass involving the skin and subcutaneous layer on the right side to the mid part of the lower back (Figure 4). No internal organ metastasis or enlargement of lymph nodes was identified. Chest X-ray also demonstrated no abnormal findings. Consequently, the patient was referred to the plastic and reconstructive surgery department for a wide excision procedure.

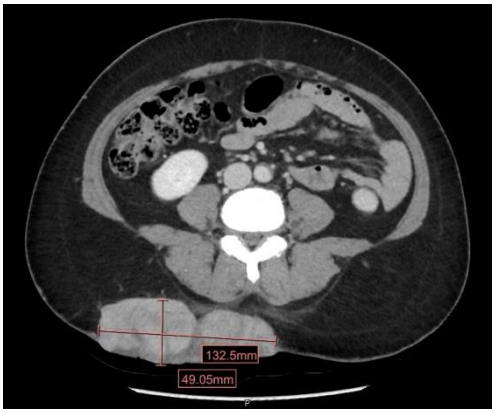

Figure 4 A contrast-enhanced computerized tomography scan revealed a 14x5 cm lobulated heterogeneous enhancing mass involving the skin and subcutaneous layer on the right side to the mid part of the lower back

Discussion

DFSP, a rare mesenchymal neoplasm originating in the dermis, typically manifests as an asymptomatic, slow-growing condition. It presents as skin-colored to red-brown indurated plaques or nodules, predominantly affecting the trunk and proximal extremities in young to middle-aged adults[2,3]. Despite its local invasiveness, DFSP generally carries a favorable prognosis due to a low rate of metastasis.

Histologically, classic DFSP exhibits spindle cells arranged in a storiform pattern, featuring monomorphic nuclei, minimal cytologic atypia, and low mitotic activity. Immunohistochemical analysis commonly reveals positivity for CD34. In cases where fibromatous changes occur, leading to the diagnosis of DFSP-FS, the histologic features include monotonously fusiform cells arranged in a fascicular or herringbone pattern. DFSP-FS is characterized by enlarged atypical nuclei, increased cytologic atypia, a high mitotic count, and CD34 negativity within the DFSP framework[4].

DFSP-FS accounts for approximately 7% to 15% of all DFSP cases[5]. Clinically, DFSP-FS is indistinguishable from classic DFSP[6]. Nevertheless, compared to classic DFSP, DFSP-FS is associated with a poorer prognosis, carrying a higher risk of local recurrence, metastasis, and mortality. However, the correlation between the degree of fibrosarcomatous change and disease outcomes is still a subject of debate. Erdem et al. reported that a higher degree of fibrosarcomatous change is linked to an increased risk of metastasis and local recurrence[7]. On the other hand, studies by Liang et al. and Abbott et al. suggested no significant correlation between the degree of FS change and prognosis[4,8]. Consequently, further research is needed to elucidate the relationship between the degree of FS change and the prognosis of DFSP-FS.

DFSP-FS poses a heightened risk of metastasis, particularly in cases of multiple local recurrences, when compared to classic DFSP[7]. Liang et al. identified the lung as the most common site of metastasis, occurring in approximately 15% of DFSP-FS cases[4]. As a result, radiologic examinations, including chest computerized tomography or magnetic resonance imaging, are recommended in cases of DFSP-FS to assess the potential for metastasis especially in the first 3 years after treatment where the risk of recurrence is at the highest[2].

The preferred treatment for DFSP-FS is surgical excision with negative margins, employing standard wide excision with 2-to-4-centimeter margins or Mohs micrographic surgery which is a preferred treatment due to its low recurrence rate compared to standard wide excision[2,5]. In cases of locally advanced or metastatic disease, systemic chemotherapy is the preferred treatment, although its efficacy is limited. Another viable option for DFSP, specifically in patients positive for the COL1A1-PDGFB fusion gene, is Imatinib-a tyrosine kinase inhibitor. Imatinib has demonstrated efficacy as an adjuvant to surgery or in advanced disease[9]. However, it is crucial

to conduct a molecular assessment of the COL1A1-PDGFB fusion gene before initiating imatinib treatment, as the drug has proven to be ineffective in the absence of this fusion gene[10].

In this report, we present a case of DFSP-FS, emphasizing its uniqueness within the spectrum of DFSP. This case underscores the importance of recognizing the diagnosis of DFSP-FS and the necessity for more aggressive management strategies.

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