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Case Report

A histopathological surprise of dermatofibrosarcoma protuberance: a case report and review of the literature

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ABSTRACT

Dermatofibrosarcoma protuberans (DFSP) represents around 1% to 6% of all soft tissue malignancies, with an annual incidence rate of 4.2 per million individuals. This tumor is most commonly found in the proximal regions of the limbs (10% to 15%), followed by the trunk (40-50%) and chest/shoulders (30% to 40%). Clinically, DFSP often presents as a gradually growing nodule or plaque, initially asymptomatic, which later enters a phase of rapid growth. While DFSP has a significant likelihood of local recurrence, it rarely metastasizes. Children are more likely to have DFSP in distant locations. A study of 27 cases found that 14.8% of childhood DFSP cases involved the hands and feet. DFSP typically presents in individuals between the ages of 20 and 50 years. In this report, we present a case of a young female patient who was incidentally diagnosed with DFSP on the right hallux, confirmed through histopathological analysis, along with its management.

Keywords: Dermatofibrosarcoma protuberans, DFSP, Soft tissue sarcoma, Toe, Lump, CD34, Pathology

INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) was first described by Sherwell and Taylor in 1890. In 1925, Hoffman recognized it as a cutaneous malignancy with a high recurrence rate and a tendency to form projecting nodules, later naming it DFSP.[1,2] DFSP is a rare cutaneous sarcoma originating from dermal fibroblasts. It is locally aggressive with a low likelihood of metastasis but has a strong propensity for local recurrence. The tumor often invades surrounding tissues such as bone, muscle, periosteum, and fascia.[2] Clinically, DFSP typically presents as a slowly growing, asymptomatic plaque or nodule that eventually accelerates in growth. Although the tumor rarely metastasizes, it frequently recurs locally, especially in cases of fibrosarcomatous transformation. The diagnosis of DFSP is often delayed, as its subtle, slow-growing nature makes it difficult to identify without biopsy and histological examination. As our understanding of DFSP's clinical and histological features has advanced, new surgical techniques and targeted pharmaceutical treatments have been developed.[2,3]

In this article, we review current surgical treatment options, the epidemiology, pathogenesis, clinical features, diagnosis, staging, and prognosis of DFSP, using a case from our practice as an example.

The incidence of DFSP ranges from 0.8 to 5 cases per million people annually, representing less than 0.1% of all malignant tumors worldwide.[1-3] Cases of DFSP in the foot and toe are relatively rare in the literature.[3] DFSP

can affect individuals of any age, from infants to those over 90 years old, although it most commonly affects young to middle-aged adults.[2,3] Although the tumor has been documented in infants as young as a few months, the occurrence of DFSP in children is relatively low. The pediatric subtype of DFSP is generally more aggressive and associated with a higher recurrence rate compared to the adult-onset form.

Due to the slow-growing nature of DFSP, it is believed that the tumor may begin in childhood but not exhibit symptoms until later in life. Although the gender ratio is roughly equal, some studies suggest that females may be slightly more affected than males. Studies also indicate that African Americans have a higher incidence of DFSP than Caucasians, although this finding varies across different research sources. Recent population-based studies show five-year relative survival rates for DFSP ranging from 98% to 100%, with no significant differences in survival based on gender or race.[3,4]

Pathogenesis

Due to its rarity, the exact cause of dermatofibrosarcoma protuberans (DFSP) remains unclear. However, the chromosomal translocation t(17;22)(q22;q13), which involves the COL1A1 and PDGFB genes, is the primary driver of tumor pathogenesis and is present in more than 90% of cases.[4] This translocation results in the fusion of the platelet-derived growth factor beta (PDGFB) gene at 22q13 with the type I collagen (COL1A1) gene at 17q22, which leads to the overexpression of PDGFB, stimulating the formation of fibroblasts and myofibroblasts. This process is thought to play a significant role in the development and progression of DFSP.[2-4]

DFSP has several histologically distinct variants, including the classic subtype, pigmented (Bednar tumor), myxoid, granular cell, atrophic, and sclerosing/sclerotic DFSPs and those with fibrosarcomatous transformation, areas of giant cell fibroblastoma, and myoid/myofibroblastic differentiation.[3,4]

The most common subtype, the classic form, is characterized by a storiform or honeycomb pattern of spindle cell proliferation. The other less common variants exhibit distinct morphological features, for example, the pigmented form contains melanin pigment within the tumor cells while fibrosarcomatous DFSP features increased cellularity and mitotic activity, with a more disorganized structure.

Several factors, including tumor suppressor genes, immunodeficiencies, and oncogenes, have been linked to DFSP development, though further research is needed to fully understand the interaction of these risk factors.

DFSP typically presents as a firm, painless, progressively growing skin lesion that is usually flat or slightly elevated. The lesion may have an irregular shape and can appear pink, brown, or skin-colored. Microscopically, DFSP often exhibits a "honeycomb" or "storiform" appearance. While DFSP can occur anywhere on the body, it most commonly affects the extremities (30-40%), particularly the proximal portions, and the trunk (40-50%), typically on the chest and shoulders. Approximately 10-15% of cases involve the head and neck, particularly the face, scalp, and supraclavicular region. On the scalp, DFSP may be misdiagnosed as benign cysts or tumors.

In children, DFSP has a higher likelihood of occurring in acral locations. The tumor may spread deeply into the subcutaneous tissue and form satellite nodules in the surrounding skin.[2-4]

Although DFSP rarely metastasizes, local recurrence is common, particularly if the tumor is not completely excised with free margins. In some cases, DFSP may transform into fibrosarcoma, a more aggressive form of sarcoma. The diagnosis of DFSP is typically confirmed through a combination of clinical evaluation, imaging, and histological examination. While a clinical suspicion can be made, definitive diagnosis requires an incisional or excisional biopsy.

CASE REPORT

A 19-year-old female patient presented to the surgical clinic in Ibra hospital, Ibra, Oman with a history of painless swelling at the medial aseptic of the left big toe for 12 years. She had no history of trauma or previous surgery this area. No history of fever or other associated symptoms. She has no any co-morbids or other warning or constitutional symptoms.

Further, she no family history of such lesions or cancer. On assessment, there was a palpable mass about one by 1.5 cm in the medial aspect of her left big toe. The mass was non-tender, mobile and non-fluctuant. It was soft to firm in consistency, having well-defined edges, normothermic skin, non-compressible, non-pulsatile with no local signs of inflammation. No joint movement restriction or neurovascular deficit was noted. Draining lymph nodes were free.

Her blood tests and x-ray foot revealed no abnormality. An excisional biopsy as a benign lesion under local anesthesia was done. Her postoperative recovery was uneventful. Further, histopathology reported as a DFSP.

After releasing the report, the patient was referred to a tertiary care hospital for further workup and management. Fortunately, she did not require a re-do or wide local excision (WLE), because of initial surgery showed WLE margins. On follow-up, she was doing well without recurrence and she is on active surveillance for the coming one year. Figure 1 represent the histologic findings of the case.

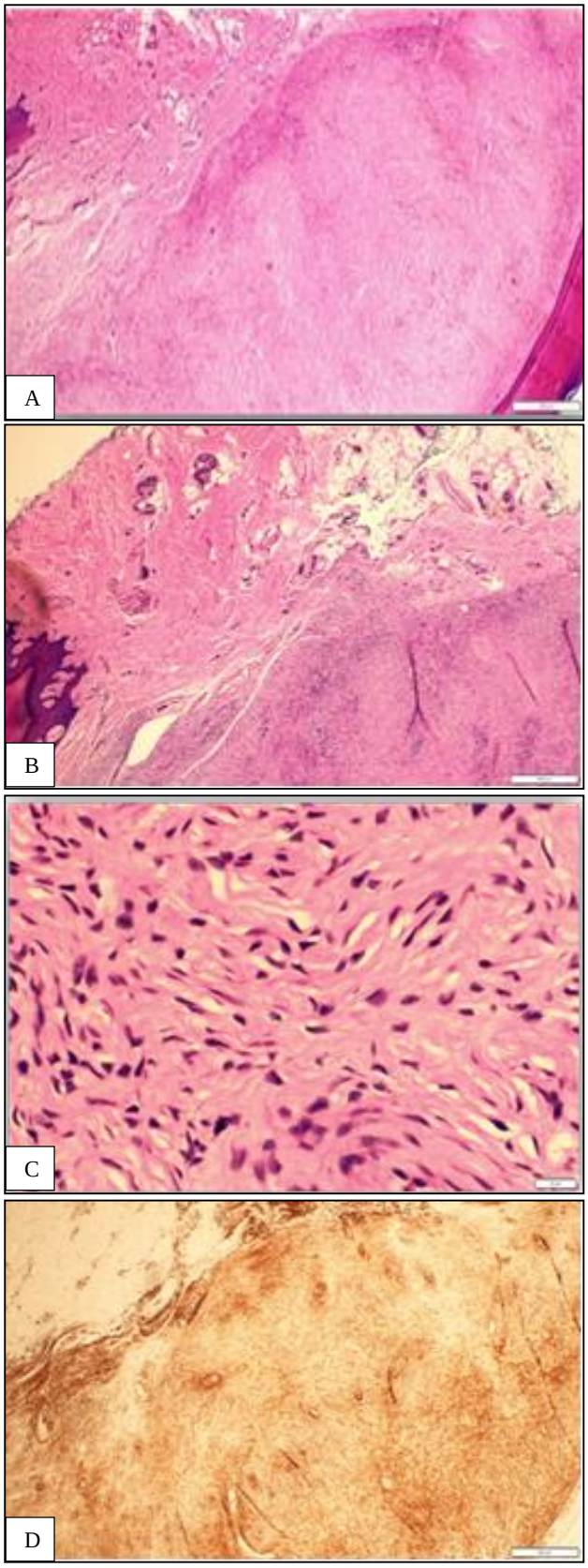

Figure 1: Dermal spindle cell neoplasm HE X40 (A), it is seen in the deep dermis but completely excised HE X100 (B), the proliferating fibroblasts are arranged in storiform pattern HE X400 (C), the spindle cells show diffuse CD34 positivity X200 (D).

DISCUSSION

Although rare, DFSP is considered the most common sarcoma of the skin. It typically presents as a firm, painless skin nodule with variable coloration and can develop slowly over several decades. DFSP may extend beyond the subcutaneous tissue and, in some cases, even invade bone.[3,4] The histopathological features of DFSP on the toes include a storiform, monomorphous proliferation of spindle-shaped cells, which usually lack significant nuclear atypia or mitotic activity. The tumor often penetrates the surrounding tissue in a characteristic honeycomb pattern.

A substantial number of DFSP cases demonstrate diffuse and strong CD34 expression, while lacking α-SMA, factor XIIIa, S-100, and melan-A.[4,5] α-SMA-positive areas, indicating myoid differentiation, are found in up to 5% of cases. It is important to note that CD34 expression is not exclusive to DFSP as it can also be seen in solitary fibrous tumors, epithelioid sarcomas, spindle cell lipomas, and CD34-positive cellular digital fibromas. In addition, approximately 19% of DFSP cases are EMA-positive, with varying expression levels depending on the tumor subtype and location.[5]

Given the overlapping marker expressions, a comprehensive differential diagnosis is essential. These findings should be considered alongside further diagnostic techniques, such as electron microscopy. Moreover, a characteristic recurrent molecular gene fusion has been identified in almost 90% of DFSP cases, which can aid in confirming the diagnosis.

The fusion of the collagen type 1α1 gene (COL1A1) and the platelet-derived growth factor β chain (PDGFB) gene, resulting from the chromosomal translocation t(17;22)(q22;q13), plays a pivotal role in the development of DFSP.[5,6]

Over time, recommendations for the diagnostic evaluation and differentiation between fibrous histiocytoma and DFSP have evolved. Modern diagnostic guidelines incorporate both morphological criteria and a panel of antibodies to facilitate an accurate diagnosis. CD34 remains a key marker, but it was later discovered that CD10 is also significant, being predominantly expressed in the dermatofibroma and rarely in DFSP.[4,6] Around fifteen years ago, West et al introduced Apolipoprotein D (Apo D) as a high-confirmatory marker, which has since gained importance in the diagnosis of DFSP. The role of factor XIIIa in the differential diagnosis has also been validated.[6,7] Additional markers such as HMGA1, HMGA2, and Stromelysin-3 have been used by various diagnostic centers and groups.[8] The histological differential diagnosis for DFSP includes dermatofibroma, peripheral nerve sheath tumors (e.g., neurofibroma or schwannoma), fibrosarcoma, leiomyosarcoma, spindle cell or

desmoplastic malignant melanoma, and morpheaform basal cell carcinoma.[6-8]

The European consensus-based guidelines recommend a three-centimeter margin for WLE.[8,9] The high recurrence rate of DFSP is attributed to its microscopic finger-like extensions into surrounding tissues, which is why Mohs micrographic surgery (MMS), slow MMS, or WLE with surgical margins as wide as five centimeters are advocated. MMS has been shown to reduce recurrence rates by a factor of ten and result in less surgical morbidity compared to WLE.[9,10] Conventional chemotherapy is considered ineffective in treating inoperable cases, such as those with prior excisions; however, radiation therapy can be used for local treatment.[9-11] The tyrosine-kinase inhibitor imatinib has been recommended as a neo-adjuvant therapy for five to six months in patients with unresectable primary or recurrent DFSP, as well as for those with metastatic disease.[11,12] A 24-month course of Imatinib has been shown to reduce tumor size by 50%.[13] Although most recurrences occur within the first three years after surgery, follow-up should extend for up to ten years due to the slow growth of DFSP.[13,14] While longer durations of untreated disease are theoretically linked to a higher likelihood of fibrosarcomatous transformation, to the best of the authors' knowledge, this correlation has yet to be statistically substantiated.

CONCLUSION

DFSP is a low- to intermediate-grade malignancy that primarily affects individuals in their early to middle years. Histologically, it is characterized by a storiform pattern of bland spindle cells with several variations. It is essential to differentiate DFSP from other benign and malignant lesions. Most DFSP cases carry the chromosomal translocation t(17;22)(q22;q13), which results in the formation of the COL1A1-PDGFB fusion gene transcript, a crucial factor for both diagnostic and therapeutic purposes.

When considering WLE for DFSPs on the trunk and extremities, it is critical to ensure a safe surgical margin. MMS can be an effective option for medium to large DFSPs in these areas, offering high cure rates. MMS should also be considered for smaller DFSPs in cosmetically sensitive regions to achieve optimal tissue preservation while ensuring better aesthetic and functional outcomes. In cases where complete surgical excision is not feasible, DFSPs located on the digits may require subtotal or full digital amputation.

For patients who are not candidates for surgery, adjuvant treatments, such as radiation therapy and targeted therapies, should be considered. Regular follow-up is essential to monitor for metastasis and recurrence, with surveillance beginning six months post-surgery and continuing annually thereafter.

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