Dermatofibrosarcoma protuberans

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Abstract

Introduction: dermatofibrosarcoma protuberans (DFSP) is a rare superficial sarcoma arising from dermal fibroblasts, that affects the dermis and subcutaneous tissue, with the potential for muscle and bone invasion, and is locally aggressive. We present a case of DFSP in a young patient, highlighting the importance of appropriate diagnosis and treatment.

Clinical case: an 18-year-old male, with no history of toxic ingestion or chronic diseases developed an asymptomatic lesion in the right scapular area over three months. It began as an indurated plaque which grew, with the subsequent appearance of a nodular lesion. An incisional biopsy was done, along with histopathological and immunohistochemical studies, confirming the diagnosis of DFSP. A computed tomography scan was done to rule out metastasis, as well as a molecular study. The physical exam showed an oval 3 x 4 cm plaque with a 2 x 2 cm central nodule. The biopsy confirmed intermediate-grade DFSP. The immunohistochemical studies were positive for CD34, C163, FXIII-a and 5% Ki67. The tomography showed no metastasis. The molecular study did not show the disease's characteristic translocation. A wide excision of the tumor was done with Z-flap reconstruction. The surgical site healed properly with no recurrence.

Discussion: DFSP, while rare and frequently misdiagnosed, should be considered when slow-growing skin lesions are encountered. The main treatment is wide surgical excision, due to its infiltrative nature. The case we have presented shows the importance of appropriate diagnosis and treatment to avoid recurrence and preserve the functionality of the affected area. **(Acta Med Colomb 2025; 50. DOI: https://doi.org/10.36104/amc.2024.3789).**

Keywords: *dermatofibrosarcoma, sarcoma, skin cancer, biopsy, tumor markers*.

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Introduction

Dermatofibrosarcoma protuberans (DFSP) is a very rare low-grade soft-tissue sarcoma, although it is the most common of its type affecting the dermis and subcutaneous tissue (1-6). It has an incidence of 4/1,000,000 and was originally described by Duer and Ferrand in 1924 (7, 8).

These cancerous tumors are slow-growing, with low metastatic potential and a high recurrence rate (2, 8, 9). They occur mainly in middle-aged adults, although congenital cases are also reported (4, 10–12).

From a clinical perspective, DFSP may occur as a robust plaque or firm nodule with a pink or purple hue. Its most frequent location is on the trunk or proximal extremities. Most cases are associated with a t(17;22) (q22;q13) translocation that produces the COL1A1-PDGFB fusion protein (1, 3, 4, 9, 13).

Although metastases are uncommon, DFSP is a very locally aggressive tumor with a high tendency to recur. The presence of fibrosarcomatous areas within the tumor has been reported to increase the risk of recurrence and more aggressive behavior (14, 15).

Case presentation

This was an 18-year-old male student with no significant medical history who was seen by the dermatology service due to a three-year history of an asymptomatic lesion in the right scapular region.

It initially presented as a small indurated pinkish plaque which grew slowly, with surrounding redness and the subsequent onset of a nonpainful nodular lesion.

Physical exam showed an approximately 3 x 4 cm oval plaque in the right scapular region, with an approximately 2 x 2 cm central nodule. No other lesions were found.

An incisional biopsy was done due to clinical suspicion of DSFP (Figure 1). The histopathological study confirmed the diagnosis of intermediate grade dermatofibrosarcoma with histological malignancy.

The immunohistochemical study was positive for CD34, C163, and FXIII-a markers and 5% Ki67. A computed tomography with contrast of the chest, abdomen and pelvis showed no evidence of metastasis.

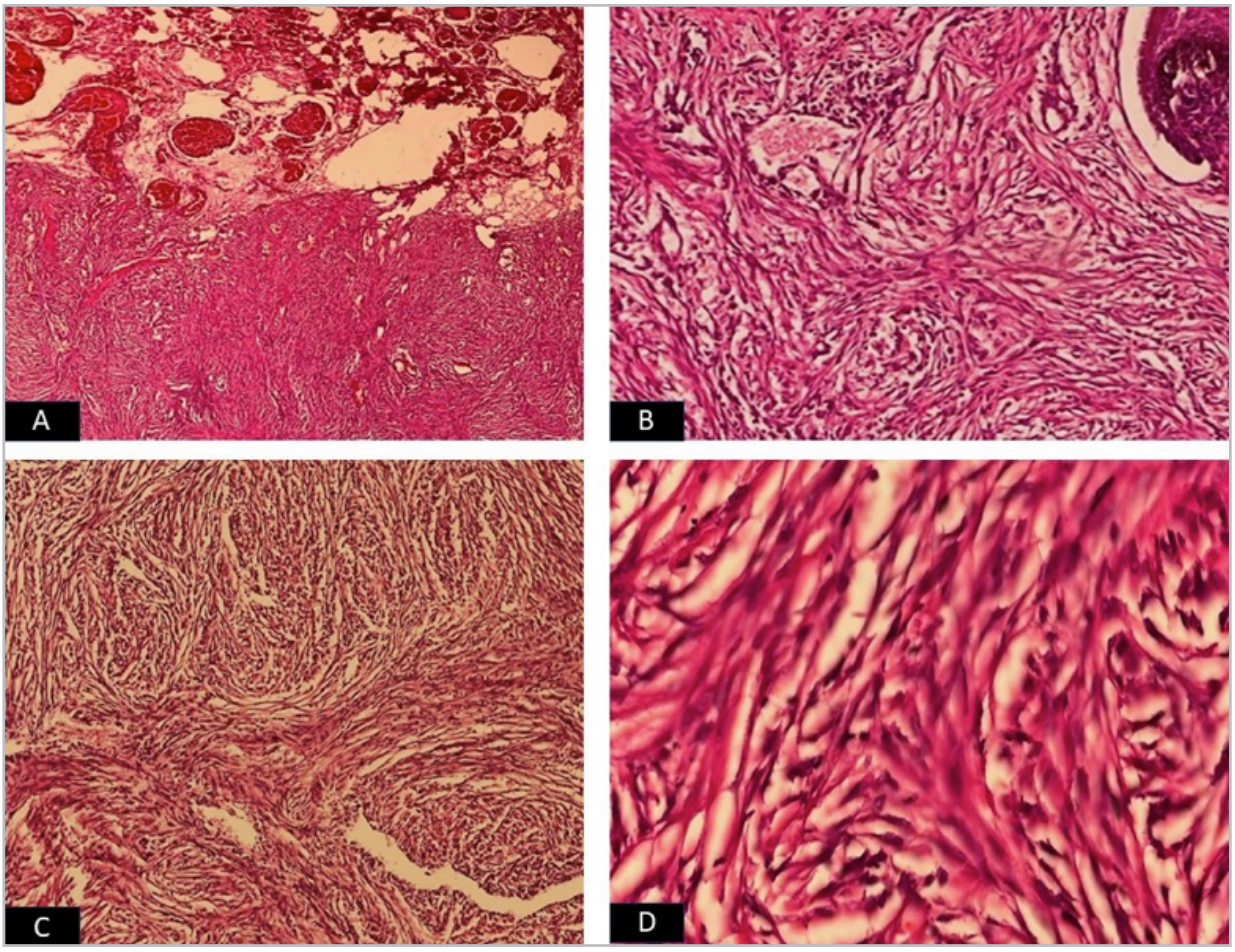

Figure 1. A: *dermis and subcutaneous cell tissue with a spindle-shaped cell neoplasm*. **B-C:** *storiform pattern in the dermis*. **D:** *spindle-shaped cells in a desmoplastic stroma*.

A molecular test was also done, which was negative, and neither the disease's characteristic translocation nor other genetic variants were documented.

Subsequently, a wide excision of the tumor was performed, with Z flap reconstruction. The surgical site has healed well with no recurrence reported to date (Figure 2).

Discussion

Dermatofibrosarcoma protuberans is a tumor arising from dermal fibroblasts. It is a superficial, poorly circumscribed sarcoma that usually affects the dermis and subcutaneous tissue, with some cases potentially invading the muscles and bone (1, 7, 15, 16). It usually occurs in people between the ages of 20 and 60, equally distributed between males and females. However, some studies say that female patients are slightly more affected, but the proportion is still classified as one to one (5, 9, 10, 16).

Due to its metastatic potential, it is considered an intermediate tumor, between a benign dermatofibroma and a full-blown fibrosarcoma. Transformation to a high-grade sarcoma is extremely rare, with an incidence of 1-3/1,000,000

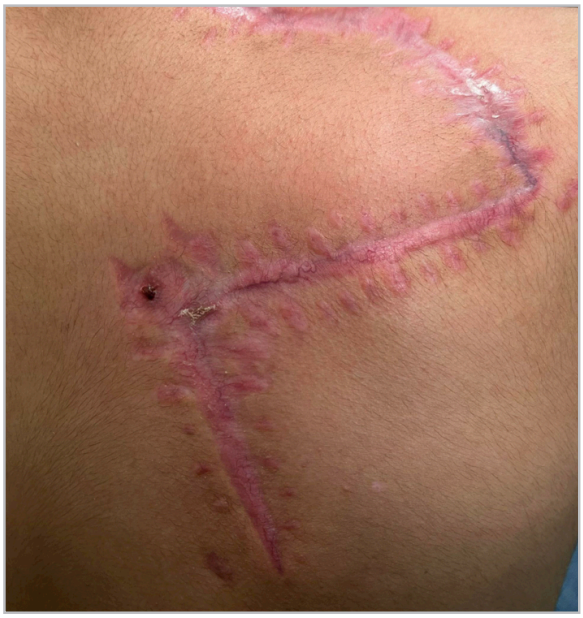

Figure 2. Healing of the surgical excision and Z-plasty reconstruction.

(1, 7). As a rare condition, it is usually diagnosed erroneously or late, due to its mostly benign nature (2).

Dermatofibrosarcoma protuberans usually manifests as a firm, asymptomatic flesh-colored to reddish-brown plaque. Over time, it progresses to multiple raised nodules with a purplish to reddish-brown hue. It grows slowly, spreading over months or years, at times resembling dermatofibromas or cheloids (1, 8, 9, 17). Given its similar appearance, it is advisable to rule out other soft tissue tumors like fibrolipomas, solitary fibrous tumors, spindle cell lipomas, schwannomas, angiosarcomas and fibrosarcomas (1, 3, 9–11).

In the case we have presented, the patient had an oval plaque measuring approximately 3 x 4 cm in the right scapular region that had developed over approximately three years, with a 2 x 2 cm central nodule.

As DFSP grows, it can reach several centimeters in diameter. Some cases may develop telangiectasias in the surrounding or overlying skin. As the lesions enlarge, some may ulcerate and become painful (1, 8).

More than 90% of lesions are said to be due to t(17;22) (q22;q13) translocation. Other variants include translocation of the platelet-derived growth factor subunit B gene, although on chromosome 22. It may occur in some patients without t(17;22) translocation, with evidence of microsatellite instability, which is a characteristic due to abnormalities in the number of repeated deoxyribonucleic acid bases within the microsatellites or sequences. The t(17;22) (q22;q13) translocation causes the formation of supernumerary ring chromosomes from chromosome 22, and contains low-level amplified sequences from 17q22-qter and 22q10-q13.1 (1, 4, 9, 18).

Spindle tumor cells are arranged in a storiform pattern, parallel to the epidermal surface, and have pleomorphism. These cells are surrounded by collagenous stroma, normally associated with hyaline or myxoid changes. The predominant characteristic is a honeycomb appearance, resulting from irregular tentacle-like projections infiltrating the underlying subcutaneous tissue that traverse the septa and fat, trapping it (1, 4, 8, 12, 19). The epidermis is generally not affected.

The pathology study images showed the characteristic honeycomb shape and irregular tentacle-shaped projections (Figure 1).

Histological variants have been described, including the classic, pigmented, myxoid and giant cell variants, atrophic DFSP, fibrosarcomatous DFSP, DFSP with areas of giant cell fibroblastoma, and DFSP with myoid differentiation foci (6, 9, 14, 16).

Hematoxylin-eosin staining is used to evaluate and detect DFSP, using optic microscopy. When DFSP is suspected, immunohistochemistry confirmation tests should be done, because DFSP stains positive for CD34 in 80 to 100% of cases. Hyaluronate and vimentin are also typically positive. Dermatofibrosarcoma protuberans has negative staining for factor XIIIa, smooth muscle actin, desmin, S100 proteins and keratins (1, 4).

The immunohistochemistry study in our patient was positive for CD34, C163, and FXIII-a markers and 5% Ki67.

It is primarily treated with surgical excision with wide surgical borders, as it is locally aggressive and infiltrative (2, 8, 15, 19, 20). Treatment after surgery may employ imatinib mesylate, which is the treatment for primary/recurrent tumors and metastatic DFSP, although radiation therapy may be used if the tumor is radiation sensitive (7, 9, 10, 20).

The patient underwent local broad surgical excision with Z-flap reconstruction. He was not a candidate for a wider incision due to the presence of muscle and bone structures inherent to arm function.

Conclusion

The case we have presented of an 18-year-old male patient with dermatofibrosarcoma protuberans (DFSP) in the right scapular region highlights the importance of early diagnosis and multidisciplinary management to treat this rare tumor.

Despite the insidious and asymptomatic nature of the lesion, the clinical assessment and diagnostic tests, including the histopathological and immunohistochemical tests, helped confirm the diagnosis and plan an appropriate surgical intervention.

Wide excision of the tumor, followed by Z-plasty flap reconstruction resulted in adequate healing, with no evidence of recurrence to date.

This case underscores the importance of continuous surveillance and postoperative follow-up, both to ensure no recurrence as well as to evaluate the need for additional interventions.

It also highlights the relevance of considering DFSP within the differential diagnosis of chronic skin lesions, especially in young patients with no history of chronic or infectious diseases.

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