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Dermatofibrosarcoma Protuberans of the Cheek: A Case Report and Review of the Literature

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Abstract | **Case Report**

***Background*:** Dermatofibrosarcoma protuberans (DFSP) is a rare mesenchymal skin tumor characterized by slow growth, local aggressiveness, and a low metastatic potential. Facial involvement is uncommon, posing diagnostic and therapeutic challenges. ***Case Presentation*:** We report the case of a 38-year-old male presenting with a progressively enlarging mass on the right cheek. Imaging and biopsy confirmed DFSP, and molecular testing identified the COL1A1-PDGFB fusion. Given the lesion's size, muscular infiltration, and potential functional and aesthetic morbidity, neoadjuvant imatinib (400 mg/day) was administered for three cycles, achieving a 40% volumetric reduction. This allowed a wide local excision with clear margins (R0) and local flap reconstruction, preserving facial function and aesthetics. At 12 months follow-up, the patient remained in complete remission without local recurrence or metastasis. ***Conclusion*:** This case illustrates the efficacy of neoadjuvant imatinib in locally advanced DFSP of the head and neck, facilitating conservative surgical management. Molecular confirmation of COL1A1-PDGFB fusion is essential to guide targeted therapy. Lifelong surveillance remains crucial due to the risk of late local recurrence.

Keywords: Dermatofibrosarcoma Protuberans, Cheek, Neoadjuvant Imatinib, COL1A1-PDGFB Fusion, Surgical Management, Case Report.

INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a rare mesenchymal skin tumor, accounting for about 1% of soft tissue sarcomas and less than 0.1% of all malignant skin tumors [1,2]. First described by Darier and Ferrand in 1924, DFSP is characterized by slow but infiltrative growth, low metastatic potential, and a high risk of local recurrence in the absence of complete excision [3]. Facial involvement is exceptional, representing less than 15% of cases [4]. Histologically, DFSP is composed of spindle cells arranged in a storiform pattern and strongly expressing CD34 [5].

In 90% of cases, it is associated with the COL1A1-PDGFB fusion, which activates the PDGF pathway and serves as a therapeutic target for imatinib mesylate, a tyrosine kinase inhibitor [6]. The standard treatment for localized DFSP is wide surgical excision with 2–3 cm margins, or Mohs micrographic surgery in functionally or cosmetically sensitive areas [10]. However, in locally advanced, unresectable, or mutilating cases, neoadjuvant imatinib has proven effective, enabling significant tumor reduction and subsequent surgery in more than half of cases [11,12].

We report a rare case of DFSP located on the right cheek, treated with neoadjuvant imatinib followed by complete surgical resection, illustrating the value of a multimodal therapeutic strategy.

CASE PRESENTATION

A 38-year-old male, single, with a history of chronic urticaria and psoriasis, presented with a right cheek mass of two years' duration. The lesion demonstrated progressive enlargement and was associated with systemic symptoms including unquantified weight loss, asthenia, and anorexia. The patient was otherwise healthy, with no relevant family history and no use of oral contraception.

Clinical examination revealed a firm, deep, non-fluctuant mass on the right cheek, mobile relative to

underlying structures, without regional lymphadenopathy. ECOG performance status was 1.

Ultrasound imaging showed a heterogeneous, vascularized subcutaneous mass measuring 15 × 22 mm without involvement of underlying muscle or bone.

A cutaneous biopsy revealed a spindle-cell mesenchymal proliferation. Immunohistochemistry supported a diagnosis of DFSP, and molecular testing confirmed the COL1A1-PDGFB fusion.

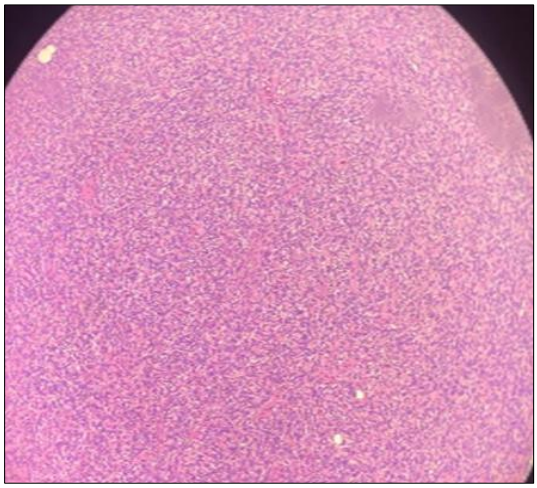

Figure 1: HES ×4: Mesenchymal proliferation with moderate to high cellularity, showing interlacing short fascicles and focal storiform pattern

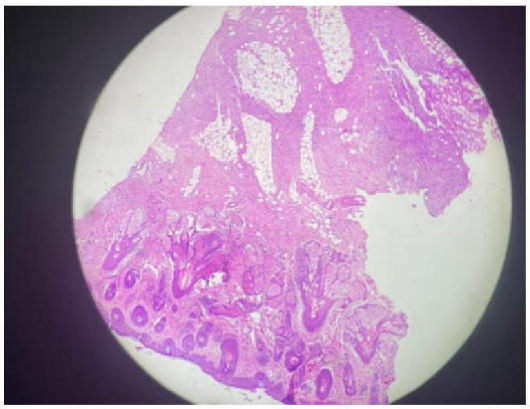

Figure 2: HES×10: spindle-cell proliferation infiltrating subcutaneous fat in a "honeycomb" pattern.

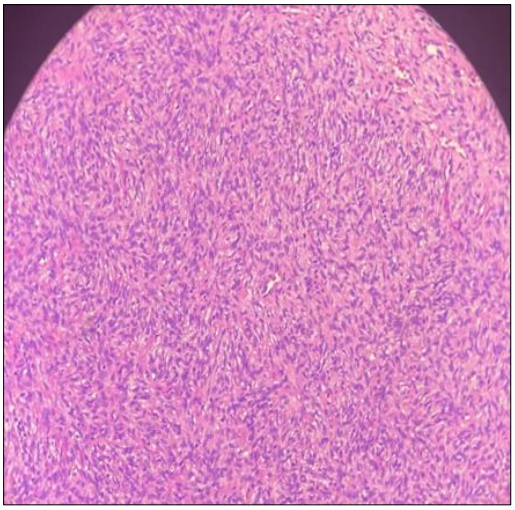

Figure 3: HES×10: Dermal spindle cell proliferation with storiform pattern

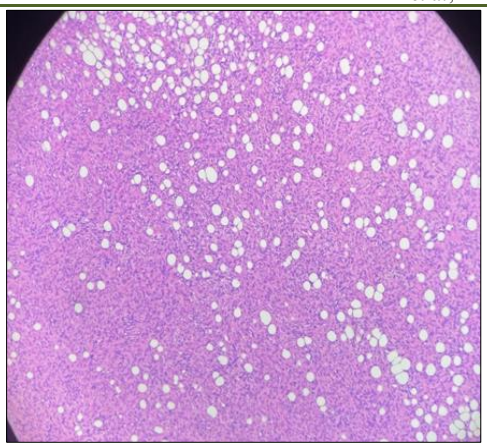

Figure 4: HES×10: infiltration of subcutaneous adipose tissue by spindle cell proliferation

Facial MRI identified a 26 × 18 mm dermo-hypodermal lesion infiltrating the underlying masseter muscle fascia, with no bone involvement. Staging thoraco-abdomino-pelvic CT scan showed no distant metastases.

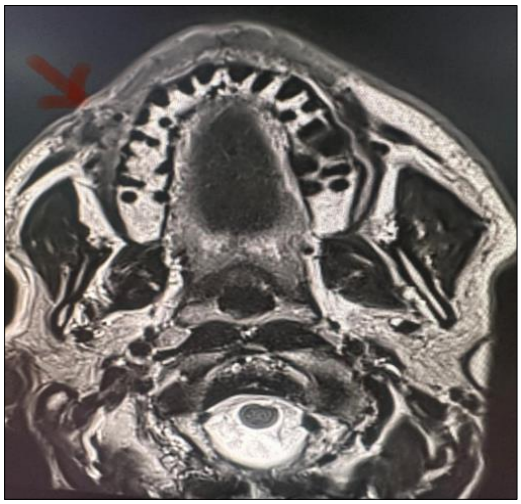

Figure 5: Axial MRI in T1 sequence with gadolinium injection, showing a heterogeneous tissue mass in the right cheek infiltrating the surrounding soft tissues and fat

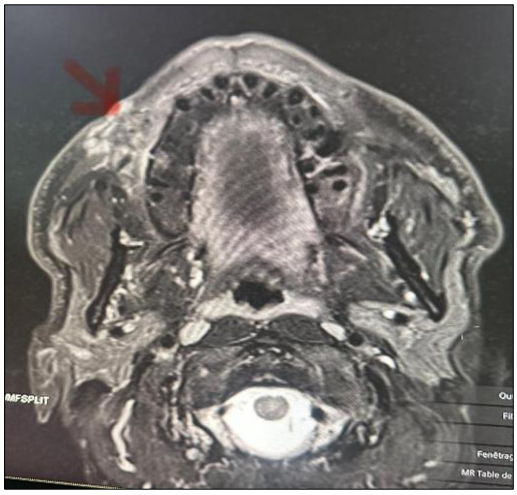

Figure 6: Axial MRI in T2 sequence, showing a tissue lesion with irregular contours and relative hyperintensity in the right cheek region

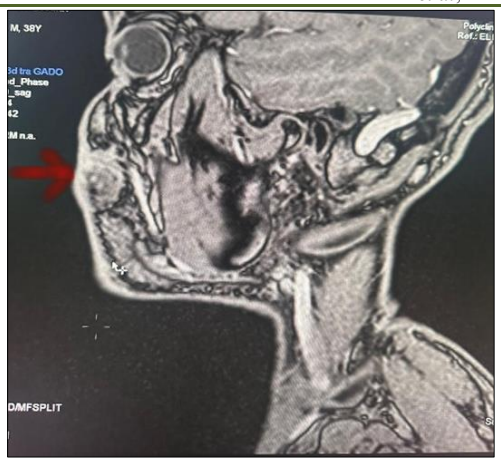

Figure 7: MRI sagittal view in T1 sequence with gadolinium injection, showing a heterogeneous tissue process in the cheek region enhanced after contrast injection

The case was discussed in a multidisciplinary tumor board. Due to the tumor's location, muscular infiltration, and the fusion gene target, neoadjuvant imatinib (400 mg/day) was initiated for three cycles.

Post-treatment MRI demonstrated a 40% reduction in tumor volume (15.5 × 13 mm), facilitating a more conservative surgical approach.

Wide local excision with 2 cm lateral margins and depth including the fascia of the masseter muscle was performed under general anesthesia, followed by local flap reconstruction. Postoperative recovery was uneventful. Histopathology confirmed residual DFSP with reduced cellularity and fibrotic changes post-treatment. Margins were negative (R0), with the closest margin measuring 1.8 cm, and no high-grade sarcomatous transformation was noted.

The patient underwent semi-annual clinical evaluations and annual MRI surveillance. At 12 months postoperatively, he remained in complete remission, with excellent functional and cosmetic outcomes. Lifetime annual follow-up is planned due to the risk of late recurrence.

DISCUSSION

Dermatofibrosarcoma protuberans (DFSP) is a rare mesenchymal tumor, accounting for less than 0.1% of all malignant neoplasms and approximately 1% of soft tissue sarcomas [1]. It is characterized by slow growth, significant local aggressiveness, and a low metastatic potential (<5%) [13]. The clinical presentation is often misleading, typically manifesting as a firm nodule or a gradually enlarging subcutaneous mass, as observed in our patient [14].

Diagnosis relies on histopathological and immunohistochemical examination, which reveals a spindle-cell proliferation arranged in a storiform pattern and strong CD34 expression, allowing DFSP to be distinguished from other cutaneous spindle-cell tumors [10]. Molecular biology plays a fundamental role: the t(17;22) (q22;q13) translocation, responsible for the COL1A1-PDGFB fusion, is detected in over 90% of cases and confers particular sensitivity to imatinib [15]. In our case, molecular confirmation justified the use of targeted therapy.

The standard treatment remains wide local excision with safety margins of at least 2–3 cm, including deep anatomical structures when necessary, or Mohs micrographic surgery when available [16]. However, in complex anatomical sites such as the head and neck region, upfront surgery may lead to significant aesthetic and functional sequelae. In this context, neoadjuvant treatment with imatinib mesylate is a validated option, particularly for locally advanced or unresectable tumors [17,18].

Several studies have demonstrated the efficacy of imatinib in tumor reduction for DFSP harboring the COL1A1-PDGFB fusion, with objective response rates ranging from 50 to 65%, thus optimizing surgical conditions [19]. In our observation, administration of imatinib at 400 mg/day for three cycles resulted in a volumetric reduction of 40%, enabling a conservative surgical approach with clear margins (R0).

The prognosis of DFSP is generally favorable when complete excision is achieved. The risk of local recurrence ranges from 10 to 20%, primarily in cases of inadequate margins [20]. High-grade sarcomatous transformation is rare but possible. Therefore, prolonged and lifelong surveillance is recommended, with closer follow-up during the first five years [21]. In our case, after 12 months of follow-up, the patient remains in complete remission, without local or metastatic recurrence, and demonstrates excellent aesthetic outcome.

In summary, this case illustrates the value of neoadjuvant imatinib in locally advanced DFSP, enabling more conservative surgery, and underscores the

importance of molecular biology in the personalized management of this rare tumor.

CONCLUSION

Dermatofibrosarcoma protuberans is a rare, locally aggressive cutaneous tumor, whose management primarily relies on complete surgical excision. The identification of the COL1A1-PDGFB fusion has enabled the introduction of targeted therapy with imatinib as an effective option for locally advanced or surgically challenging cases, thereby improving operative conditions as well as functional and aesthetic outcomes. The reported case illustrates the value of a multidisciplinary approach integrating molecular diagnostics, targeted therapy, and tailored surgery, achieving complete remission with satisfactory aesthetic results. Prolonged surveillance remains essential due to the risk of late local recurrence.

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