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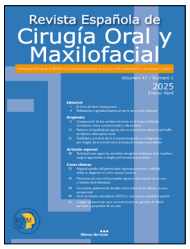

Caso Clínico

Atypical spindle cell/pleomorphic lipomatous tumor: a difficult entity to diagnose in a very unusual location

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ABSTRACT

Nowadays, lipomatous tumors are difficult entities to identify. In some cases, this process requires experience and a high level of knowledge. Such is the case of atypical spindle cell/pleomorphic lipomatous tumors (ASPLT), known for their similarity with numerous pathologies, in addition to the great histopathologic variability that they can present. This article aims to further investigate ASPLTs and their clinical and microscopic features, as well as provide key points that can help detect them more easily. It is of vital importance to be able to distinguish ASPLTs from malignant entities, such as liposarcomas. Therefore, we present the case of a 68-year-old woman who was diagnosed with an ASPLT on the left cheek and underwent surgical enucleation. We furthermore describe the clinical, histopathological, and immunohistochemical features of the lesion.

Tumor lipomatoso atípico de células fusiformes/pleomórfico: una entidad difícil de diagnosticar en una localización muy poco habitual

Palabras clave:

Célula fusiforme atípica/tumor lipomatoso pleomórfico, liposarcoma, deleción RB1, amplificación MDM2, inmunohistoquímica.

RESUMEN

Hoy en día, los tumores lipomatosos son entidades difíciles de identificar. En algunos casos, este proceso requiere experiencia y un alto nivel de conocimientos. Tal es el caso de los tumores lipomatosos atípicos de células fusiformes/pleomórficos (ASPLT), conocidos por su similitud con numerosas patologías, además de la gran variabilidad histopatológica que pueden presentar. Este artículo pretende profundizar en los ASPLT y en sus características clínicas y

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microscópicas, así como proporcionar puntos clave que pueden ayudar a detectarlos con mayor facilidad. Es de vital importancia poder distinguir los ASPLT de entidades malignas, como los liposarcomas. Por ello, presentamos el caso de una mujer de 68 años a la que se le diagnosticó un ASPLT en la mejilla izquierda y fue sometida a enucleación quirúrgica. Además, describimos las características clínicas, histopatológicas e inmunohistoquímicas de la lesión.

INTRODUCTION

The most common soft tissue tumors are those of lipomatous origin, more specifically, lipoma. Although most times it is an easy lesion to identify, certain types of tumors, such as atypical spindle cell/pleomorphic lipomatous tumors (ASPLT), are known to be particularly difficult to diagnose[1]. Dei Tos et al. were the first to describe spindle cell lipomatous tumors with atypia and further described their histopathologic similarity to spindle cell liposarcoma, well-differentiated liposarcoma, myxoid liposarcoma, and atypical spindle cell lipoma[2]. Subsequent genetic and molecular analyses determined that ASPLT can be classified into atypical spindle cell lipomatous tumors and atypical pleomorphic lipomatous tumors; thus, the term ASPLT is employed due to the possibility of finding spindle and/or pleomorphic cells[3]. It was not until 2020 that the term ASPLT was included in the World Health Organization (WHO) classification of soft tissue tumors[4]. By definition, ASPLT is a benign lipomatous neoplasm characterized by the presence of atypical spindle cells, adipocytes, lipoblasts, pleomorphic cells, and multinucleated giant cells in an extracellular matrix formed by collagen or myxoid tissue. In addition, it is characterized by poorly defined borders, no capacity for differentiation, and low local recurrence if completely removed[4]. Histologically, it can vary depending on the proportion of different cell types that can be found. In most cases, adipocytes are mature with low to moderate atypia and multinucleation. Sometimes, it is possible to find scattered pleomorphic cells in the adipocytic or spindle cell components. ASPLT shows variable expression of CD34 (64 % positive), Rb (43 % positive), and S100 (40 % positive)[4,5]. The most frequent locations of ASPLT are the body's extremities—with hands and feet being the most common—followed by shoulders, buttocks, forearms, knees, and legs. Less common areas include the head and neck, genital area, trunk, and back. Although its etiology is not fully understood, numerous studies have found 13q14 deletions, including RB1 and its flanking genes RCBTB2, DLEU1, and ITM2B [4,6]. The present study aimed to investigate the clinical and microscopic features of ASPLT and to inquire about treatment options and prognosis. In the following, we present a case of ASPLT, its treatment, and the subsequent follow-up appointments over nineteen months.

CASE REPORT

A 68-year-old female presented to the Oral and Maxillofacial Surgery Clinic of HGZ 57 IMSS Estado de México in March 2023 with an asymptomatic enlargement of the left cheek that had continued to grow for a year. The patient had hypertension and diabetes diagnosed and controlled with irbesartan and hydrochlorothiazide for hypertension and metformin for diabetes. There was no history of any similar tumor in the family. Extraoral examination revealed a soft, fluctuant enlargement of 6 cm in length in the left buccal region, presenting an intact surface and similar color to the surrounding skin. In addition, no abnormal lymph nodes were palpable (Figures 1A, B). A CT scan revealed a 5 cm mass anteroposteriorly, extending superficially to the mandibular ramus (Figure 1C). Given the data collected, a presumptive diagnosis of lipoma was made, and the patient was scheduled for surgical enucleation under general anesthesia. Two cartridges of 2 % lidocaine with epinephrine 1:100,000 were injected into the left cheek at the level of the first upper molar. Then, an incision was made in the mucosa

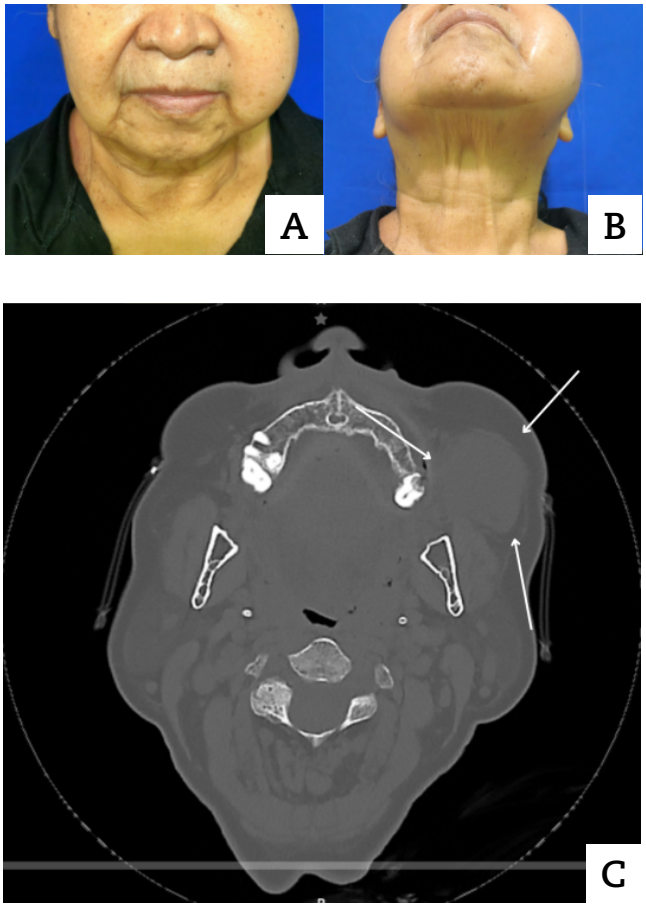

Figure. 1. Pre-operative pictures of the patient showing an increase in volume in the left buccal region (A, B), Maxillofacial CT scan of the lesion showing a 5 cm tumor in the left buccal region (white arrows) (C).

of the cheek 5 mm below Stensen's duct. A blunt dissection was performed until the entire lesion was enucleated. During surgery, the deep adherence of the lesion to the buccal fat pad was very evident, as was the lack of a fibrous capsule around the tumor, which made the removal of the lesion significantly more complex (Figures 2A, B). After surgery, the patient healed without complications. The 9 cm long specimen was sent for further study, which reported a lipomatous neoplasm with proliferation of spindle cells, lipoblasts, and adipose cells with mild atypia in a myxoid tissue stroma. The macroscopic characteristics of the tumor included myxoid components and little fat tissue (Figures 3A, B, C). Immunohistochemistry

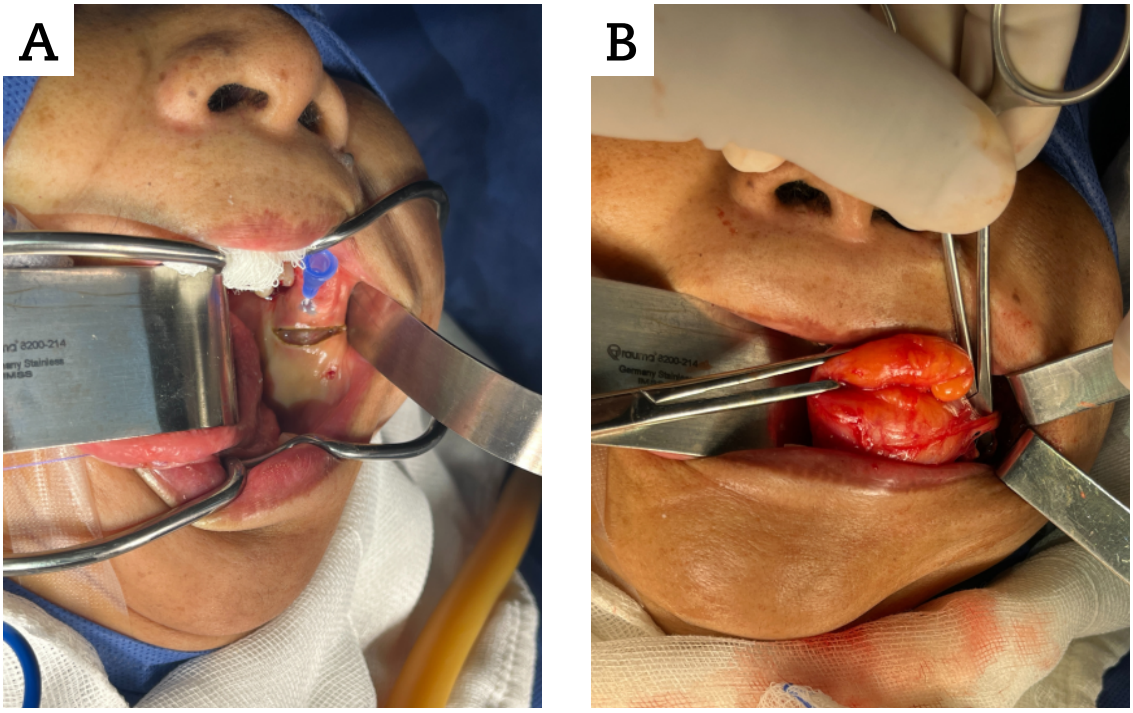

Figure 2. Incision in the left cheek 5 mm below Stensen´s duct (A), Lesion as it is coming off through the incision (B).

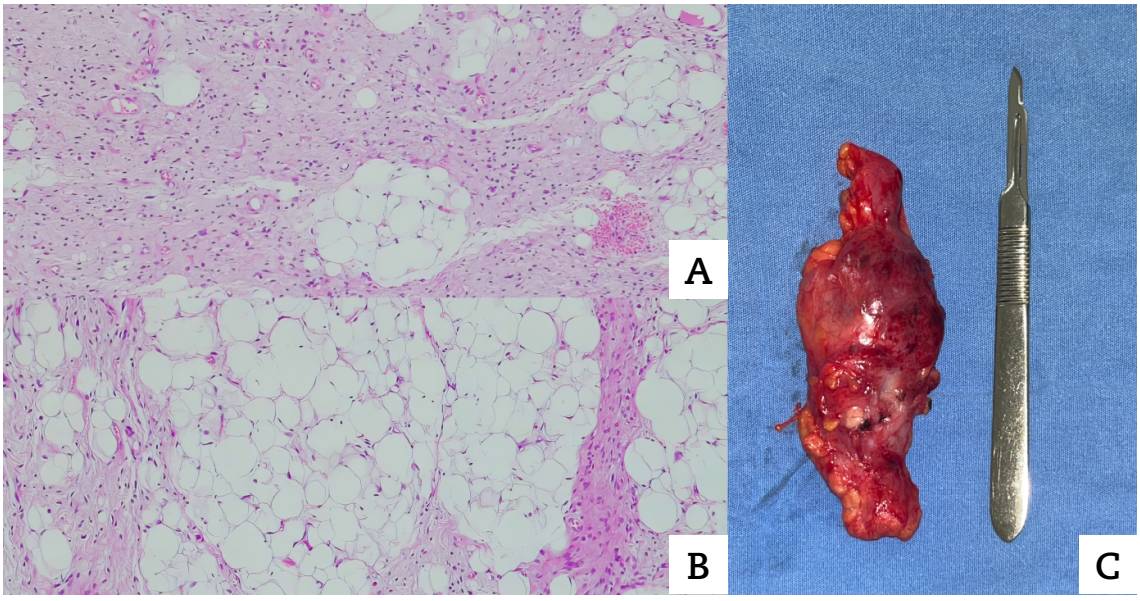

Figure 3. Histological findings, H&E staining showing proliferation of spindle cells, lipoblasts with hyperchromatic nuclei, and adipose cells with mild atypia in a myxoid tissue stroma (A, B), 9 cm long specimen containing lipomatous tissue from the buccal fat pad attached to it (C).

indicated positivity for CD34 and was negative for S100. Given the histopathologic and immunohistochemical findings correlated with the patient's symptomatology, a diagnosis of ASPLT was made. The patient was sent to the Department of Head and Neck Oncology UMAE Siglo XXI IMMS for further investigations. Initially, follow-up appointments were made at two weeks and subsequently every month. After two months, total healing of the cheek mucosa and a significant reduction of volume in the area of the lesion could be noted. In the imaging studies and control appointments conducted nineteen months after surgery, there was no evidence of recurrence of the lesion or of a significant increase in volume (Figures 4A, B, C, D).

DISCUSSION

The management of these lesions can be complex, as they do not present a single defined histopathologic pattern. Furthermore, they show variable immunohistochemistry. Ichikawa et al. reported a case of ASPLT with features very similar to myxoid liposarcoma and intramuscular myxoma[3]. Given the significant similarity with malignant entities, it is imperative to perform an excellent differential diagnosis and to include histopathology, immunohistochemistry, and fluorescence in situ hybridization (FISH) analysis[3]. The WHO currently describes essential and desirable features of an ASPLT in terms of histopathology and immunohistochemistry in order to make an accurate diagnosis. It should be noted that the lesion may contain essential but not desirable features, in which the pathologist's judgment and expertise play a fundamental role in an accurate diagnosis[4]. Mariño-Enriquez et al., in their study of 232 cases, demonstrated to an even greater extent how variable tumors of lipomatous origin can be[6].

In terms of histopathological and immunohistochemical variability, ASPLTs can differ in the number of adipocytes, lipoblasts, and spindle cells and the degree of atypia; furthermore,

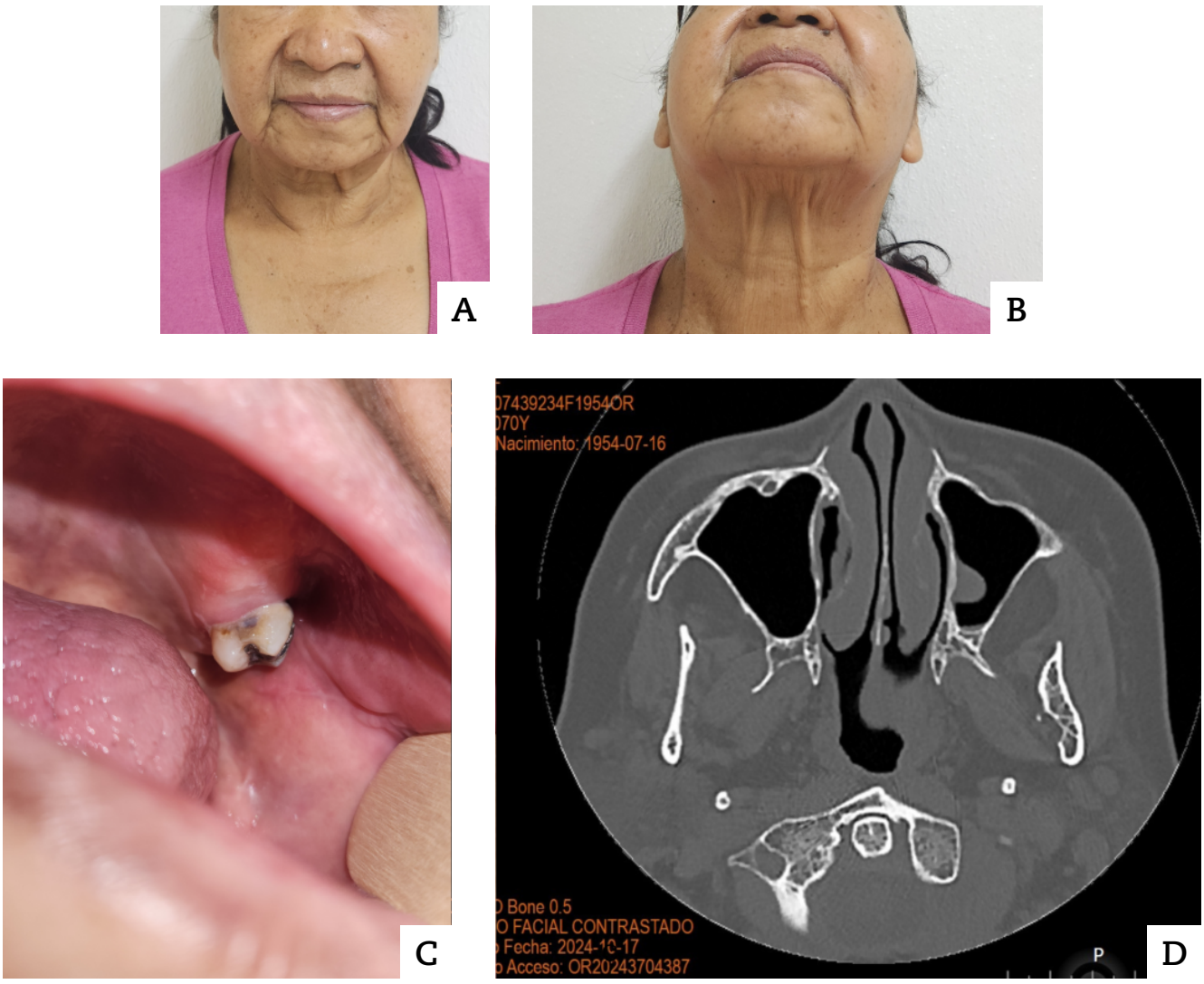

Figure. 4. Post operative pictures of the patient showing a significant decrease in the volume of the left buccal region (A, B), left buccal mucosa completely healed after 2 months (C), Maxillofacial CT scan image after nineteen months showing no data of recurrence (D).

they can be scattered in a collagen tissue stroma or a myxoid stroma to a lesser extent[7]. ASPLTs are most commonly found in the body's extremities and less commonly in the head and neck. Although infrequent, cases have also been reported in the cheek, tongue, and orbit[5,8,9].

Macroscopically, the tumor usually has myxoid components with small portions of fat. They are usually unencapsulated and show ill-defined tumor margins. Tumor size is variable (range: 0.5–28 cm; median: 5–8.5 cm)[3,4].

The treatment proposed by the WHO is surgical resection with negative margins. However, treatment proposals have been made that include radiotherapy for patients with lesion recurrence that cannot be operated on a second time[10]. Although this proposal could be effective, it is highly controversial, as cases have been published in which resection with positive margins was performed and no recurrence was reported after thirteen months[10]. As the definitive diagnosis was unknown prior to surgery in our case, the surgery was not specifically planned to leave negative surgical margins, so we immediately referred the patient to the head and neck oncology service for follow-up. Although there is no recurrence data at nineteen months, we strongly recommend a histopathologic diagnosis prior to surgical excision when the lesion is a lipomatous tumor in the deep spaces of the face.

The differential diagnosis of ASPLT includes a wide variety of lesions, the most critical factor being to confirm or rule out a malignant lesion. Because ASPLT has no metastatic capacity, has low local recurrence, and has been described as a benign lesion, it is of utmost importance to make a thorough diagnosis. Immunohistochemical tests are highly recommended, as ASPLT is negative for MDM2 with Rb loss (50-70 %), while in atypical lipomatous tumor/well-differentiated liposarcoma (ALT/WDLPS), the MDM2 factor is positive. Furthermore, in FISH analysis, ASPLT does not have MDM2 amplification and shows RB1 deletion, while ALT/WDLPS has marked MDM2 amplification without RB1 deletion[11]. Perhaps the most important differential diagnosis in this case is liposarcoma, given that different variants of it are similar to ASPLT. In this situation, because the lesion occurred in the cheek, the clinical differential diagnosis must be made considering pathologies such as liposarcoma (the most common locations of liposarcomas are the head and neck region, particularly the tongue and cheeks), lipoma, synovial sarcoma, schwannoma, and non-Hodgkin lymphoma[4,12]. Other differential diagnoses, such as pleomorphic adenoma or Warthin's tumor, were not considered due to the anterior presentation of the lesion, and typically, salivary gland lesions tend to appear in the parotid region[4].

The most common clinical presentation of ASPLT is an enlargement in the form of a soft tissue mass, nodule, or swelling, sometimes presenting with tenderness, that grows slowly and is typically painless. As previously described, the most common locations for ASPLT are the body's extremities, followed by shoulders, buttocks, forearms, knees, and legs. Less common areas include the head and neck, genital area, trunk, and back. It mainly affects men and usually appears in the sixth decade of life, measuring 5 cm or more[3,4,9].

The prognosis of ASPLT is generally good, yet it largely depends on the quality of excision and surgical edges. Nevertheless, more scientifically valid studies are needed to confirm the good prognosis of ASPLT[13].

Lipomatous tumors should not be underestimated as they can challenge physicians and pathologists in terms of diagnosis and treatment of the lesions. For any lipomatous tumor presenting diagnostic uncertainty, we suggest that an expert pathologist with extensive experience with similar lesions make the definitive diagnosis.

ETHICAL APPROVAL

Verbal and written informed consent was obtained regarding publishing the patient's data in this case report.

CONFLICT OF INTEREST

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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