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A RARE CASE OF DESMOID TUMOR IN THE LOWER MANDIBULAR SECTOR OF AN ADOLESCENT: SURGICAL INTERVENTION AND OUTCOMES

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ABSTRACT

Introduction: Desmoid tumors are rare, benign neoplasms known for their locally aggressive behavior and high recurrence rates. They arise from fibroblasts and are characterized by their infiltrative growth, making surgical management challenging. This report presents a case of a desmoid tumor in the lower mandibular sector of a 16-year-old male, focusing on surgical intervention and molecular insights. **Case Report:** A 16-year-old male presented with a growing mass in the lower right mandible, causing pain and mobility of tooth 47. Imaging revealed an infiltrative lesion eroding the mandibular bone. Histopathological analysis confirmed a desmoid tumor with spindle-shaped fibroblasts and nuclear β-caténine expression. Molecular testing identified a CTNNB1 gene mutation, confirming the diagnosis. **Discussion:** Desmoid tumors, despite being benign, are challenging due to their aggressive local behavior. Surgical excision with clear margins is the primary treatment, as these tumors are resistant to chemotherapy and radiotherapy. This case highlights the importance of precise surgical planning and the role of β-caténine and CTNNB1 mutations in diagnosis and prognosis. **Conclusion:** Effective management of desmoid tumors in adolescents requires thorough surgical intervention and comprehensive diagnostic analysis. Long-term follow-up is crucial to monitor for recurrence. The molecular characteristics of the tumor, such as CTNNB1 mutations, play a significant role in guiding treatment and predicting outcomes.

KEYWORDS: Desmoid tumor, mandibular neoplasm, fibromatosis, β-caténine, CTNNB1 mutation.

INTRODUCTION

Desmoid tumors, also known as aggressive fibromatoses, are benign mesenchymal neoplasms that arise from fibroblasts. Despite their benign nature, these tumors are locally aggressive, with a high potential for local recurrence following surgical excision [4]. Unlike other benign neoplasms, desmoid tumors do

not metastasize but have a propensity to invade adjacent tissues, making complete surgical resection difficult [5][6].

The molecular pathogenesis of desmoid tumors is closely associated with mutations in the CTNNB1 gene, leading to aberrant activation of the Wnt/β-caténine signaling pathway. This results in nuclear accumulation of β-caténine, which drives tumor growth [7]. Immunohistochemical identification of β-caténine is crucial for diagnosis, and molecular testing for CTNNB1 mutations can further confirm the condition [8][9].

This article presents a case of a desmoid tumor in the lower mandibular sector in a 16-year-old patient, with an emphasis on the surgical approach, histological findings, immunohistochemical analysis, and molecular biology results.

CASE REPORT

A 16-year-old male was referred to our Maxillofacial Surgery Department at CHU Hassan II, Fes, Morocco, for evaluation of a progressively enlarging mass in the lower right mandibular region (Figure 1), associated with pain and mobility of tooth 47. The mass had been growing over six months. Clinical examination revealed a firm, non-tender mass in the lower mandibular area, with no lymphadenopathy or systemic symptoms.

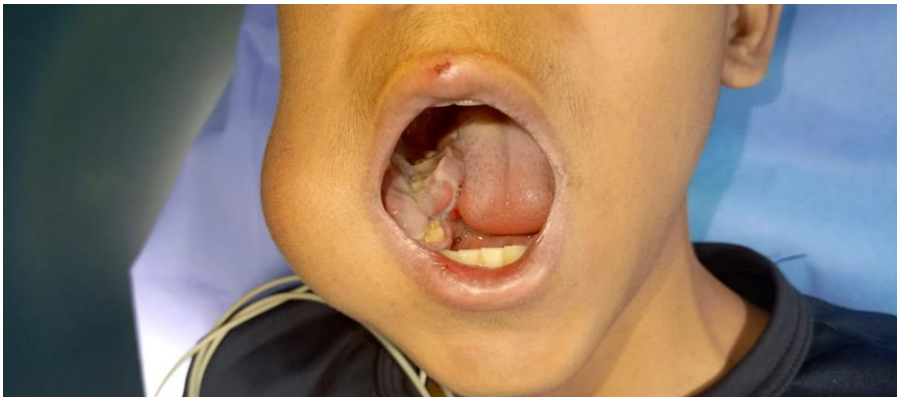

Figure 1 *(Clinical image showing a tissue-like intraoral mass pushing against the cheek.)*

Radiological imaging (CT scan) showed a homogeneously dense, infiltrative lesion eroding the cortical bone of the lower mandible. The tumor was located near the right molar region, displacing tooth 47 (Figure 2). There was no evidence of metastatic disease [10].

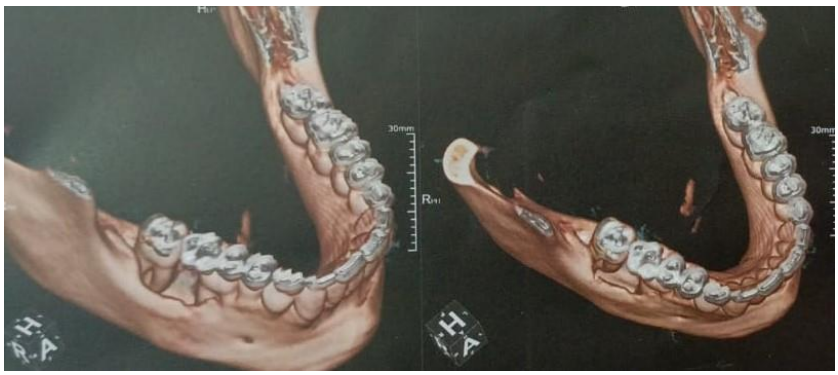

Figure 2 *(CT scan 3D reconstruction showed an infiltrative lesion eroding the cortical bone of the lower mandible.)*

A biopsy was performed, and histopathological analysis revealed spindle-shaped fibroblasts embedded in a dense collagen matrix, consistent with a desmoid tumor (Figure 3). Immunohistochemistry confirmed nuclear expression of β-caténine. Molecular testing identified a mutation in the CTNNB1 gene, confirming the diagnosis of desmoid fibromatosis [11] [12].

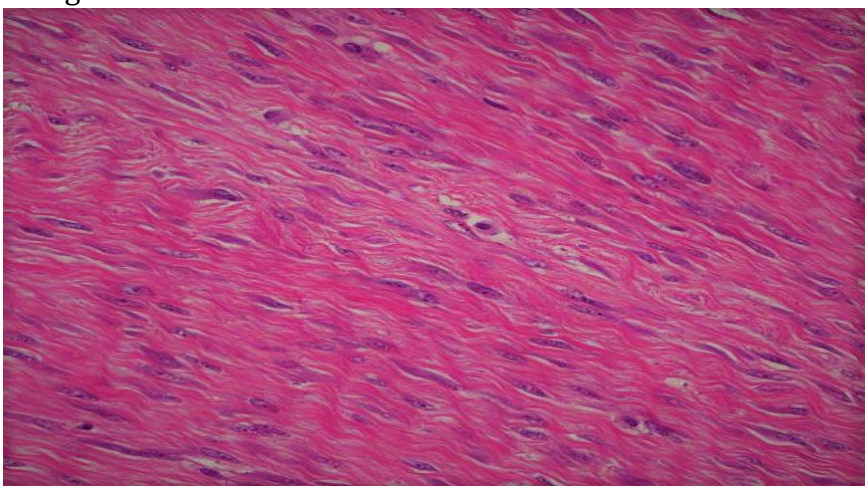

Figure 3 *(histopathological image showing spindle-shaped fibroblasts embedded in a dense collagen matrix.)*

Surgical resection was planned, and under general anesthesia, the tumor was excised with negative margins. Tooth 47, which was mobile due to tumor infiltration, was also extracted (Figure 4). The mandibular bone in proximity to the tumor was preserved, and the soft tissues were carefully approximated to ensure complete healing.

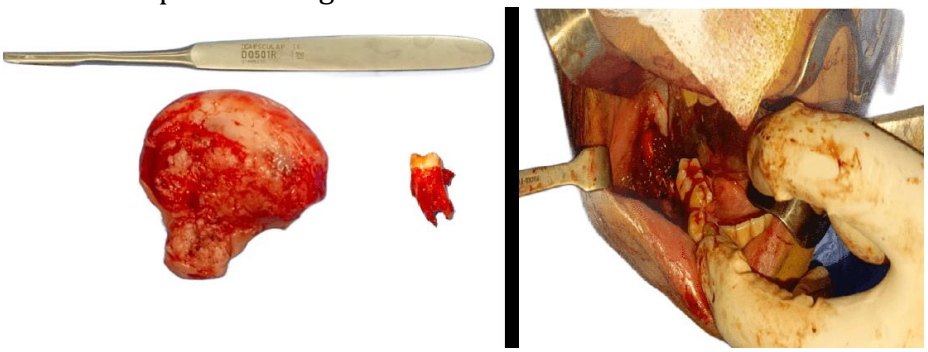

Figure 4 *(Image showing surgical excision of the tumor located near the right molar region and extraction of the second molar.)*

Postoperatively, the patient's recovery was uneventful, and follow-up imaging over 18 months showed no signs of recurrence. The patient reported full recovery of mandibular function [13] [14].

DISCUSSION

Desmoid tumors are notorious for their locally aggressive behavior, which can complicate surgical treatment. Although benign, these tumors have a high recurrence rate, often requiring a multidisciplinary approach involving surgery, radiotherapy, or systemic therapies in cases where complete excision is not feasible [1][4] [15]. In this case, surgical excision with negative margins was successfully achieved without the need for adjuvant therapy, highlighting the importance of early detection and thorough surgical planning [16].

Histologically, desmoid tumors are composed of spindle-shaped fibroblasts with low mitotic activity, lacking the features of malignancy such as pleomorphism or necrosis [3][6]. The tumor's infiltrative

nature is reflected in its ability to invade surrounding tissues, which underscores the importance of achieving clear surgical margins [7].

The role of β-caténine in desmoid tumor pathogenesis has been extensively studied, with mutations in the CTNNB1 gene identified in up to 85% of cases [8]. These mutations lead to stabilization of β-caténine, resulting in its nuclear accumulation, which drives cell proliferation [17]. Immunohistochemical detection of β-caténine is a critical diagnostic tool in distinguishing desmoid tumors from other spindle cell neoplasms [18].

In our patient, the immunohistochemical analysis showed strong nuclear positivity for β-caténine, and molecular testing confirmed the presence of a CTNNB1 mutation. This molecular insight not only aids in diagnosis but also has prognostic implications, as certain CTNNB1 mutations may be associated with higher recurrence rates [12] [19].

Long-term follow-up is essential in cases of desmoid tumors, given the risk of recurrence even after complete surgical excision. In this case, the patient was monitored for 18 months postoperatively with no signs of recurrence, demonstrating the effectiveness of early surgical intervention with appropriate margin control [10].

Conclusion

This case highlights the challenges associated with managing desmoid tumors in the mandibular region, particularly in adolescents. The combination of thorough surgical excision, histopathological confirmation, immunohistochemistry, and molecular analysis provided a comprehensive approach to diagnosis and treatment. Long-term follow-up is necessary to detect potential recurrences early. The role of β-caténine and CTNNB1 mutations in the pathogenesis and behavior of desmoid tumors underscores the importance of molecular diagnostics in guiding treatment decisions [9] [16].

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