DIAGNOSTIC AND THERAPEUTIC CHALLENGES OF CERVICAL DESMOID TUMOR: ABOUT A RARE LOCALIZATION

DÉFIS DIAGNOSTIQUE ET THÉRAPEUTIQUE DE LA TUMEUR DESMOÏDE CERVICALE: À PROPOS D'UNE LOCALISATION RARE

G. Yousfi, S. Kallel, C. Ben Amar, N. Selem, S. Graja, T. Sallemi, R. Kharrat ,I. Charfeddine
Service ORL et CCF CHU Habib Bourguiba Sfax, Université de Sfax
Laboratoire de recherche LR23ES01

ABSTRACT

Introduction: Desmoid tumor, also known as aggressive fibromatosis, resulting from fibroblastic proliferation. Although it's a benign lesion, it has a locally invasive potential leading to compressive effects on adjacent vital structures and a risk of recurrence. Surgical treatment is the main option for resecable tumors whereas primary radiotherapy could be proposed for unresectable forms.
Case report: We report the case of a 46-year-old woman presenting with an extensive desmoid tumor of the trapezius muscle. The lesion was deemed unresectable; the patient underwent partial surgical excision followed by adjuvant radiotherapy. At a one-year follow-up, no recurrence was observed.
Conclusion: This case highlights the importance of a multidisciplinary approach in the management of complex desmoid tumors. Tumor recurrence has been associated with extra-abdominal location, bone involvement, large tumor size, and younger patient age.
Key words: Desmoid tumor, Cervical desmoid tumor, Local invasion, Surgical treatment, Targeted therapy, Local recurrence.

RÉSUMÉ

Introduction: La tumeur desmoïde, également appelée fibromatose agressive, correspond à une prolifération fibroblastique. Bien qu'il s'agisse d'une lésion bénigne, elle présente un potentiel invasif local, responsable d'effets compressifs sur les structures vitales adjacentes, ainsi qu'un risque élevé de récidive. Le traitement chirurgical représente l'option de première intention pour les formes résécables, tandis qu'une radiothérapie primaire peut être proposée dans les formes non résécables.
Cas clinique: Nous rapportons le cas d'une patiente âgée de 46 ans présentant une tumeur desmoïde étendue du muscle trapèze. La lésion, jugée non résécable, a bénéficié d'une exérèse partielle suivie d'une radiothérapie adjuvante. Après un recul d'un an, aucune récidive n'a été observée.
Conclusion: Ce cas souligne l'importance d'une prise en charge multidisciplinaire dans le traitement des tumeurs desmoïdes complexes.
La récidive tumorale est favorisée par une localisation extra-abdominale, une atteinte osseuse, une taille tumorale importante et un âge jeune des patients.
Mots-clés: Tumeur desmoïde, Tumeur desmoïde cervicale, Invasion locale, Traitement chirurgical, Thérapies ciblées, Récidive locale.

INTRODUCTION

Desmoid tumors are rare benign lesions with no metastatic potential resulting from the abnormal infiltrating proliferation of fibroblasts. It is a locally invasive tumor that has a high risk of recurrence. Desmoid tumors are reported in nearly every part of the body. The localization of desmoid tumors in the trapezius muscle is extremely rare.

Auteur correspondant: chahdha Ben Ammar
E-mail: benamarchadha@gmail.com

Surgical excision is the main pillar of treatment, but complete excision of the tumor remains challenging due to its infiltrating behavior. We report a case of desmoid tumour of the head and neck diagnosed and treated in our department.

Case report

A 46-year-old diabetic woman, with no family history of cancer or familial adenomatous polyposis (FAP), presented with a painless, progressively enlarging mass on the left side of the neck that had developed over the past five months. No limitation on head rotation or dyspnea was reported, nor episodes of infection.

Physical examination revealed a postero-lateral mass of the left neck measuring 6 cm in diameter, painless, of firm consistency, adherent to the deep plane with palpable lower limit. There was no palpable cervical lymphadenopathy or inflammatory signs of the adjacent skin.
Magnetic resonance imaging (MRI) showed a left posterior cervical hypervascular mass measuring 48 × 32 × 63 mm, that has close contact with the internal carotid artery and with C2 C3 vertebrates without signs of invasion (Figure 1).

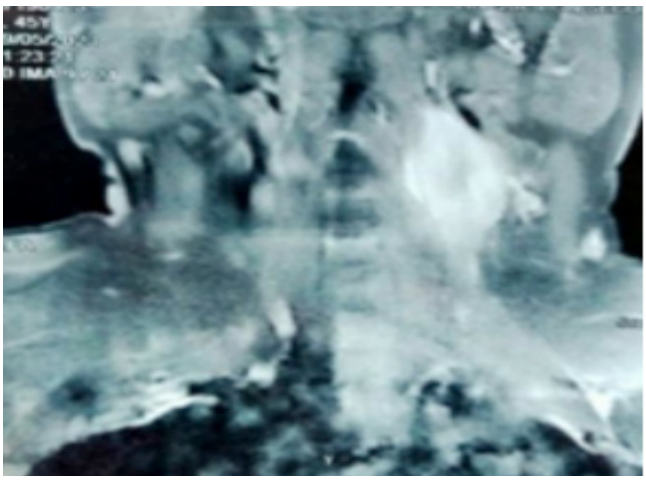

Figure 1: Coronal T1-weighted post gadolinium magnetic resonance imaging (MRI) showing an isosignal intensity tumor developed from left trapezius muscle in contact with cervical vertebral column

A biopsy of the mass confirmed a desmoid-type fibromatosis.
Based on these findings, a wide surgical resection was planned.
Intraoperatively, the tumor originated from the trapezius muscle and infiltrated the prevertebral and lateravertebral muscles. Sharp dissection separating the tumor from the surrounding muscle was not feasible. An incomplete excision involving part of the trapezius muscle was performed (Figure 2).

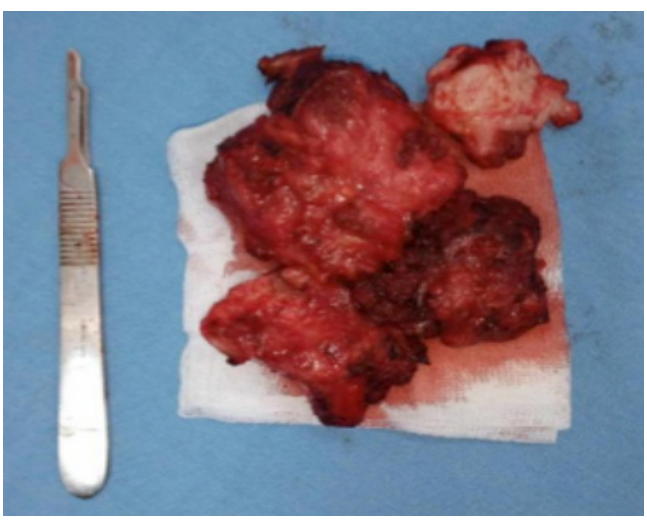

Figure 2: Perioperative view of the tumor removal specimen with the surrounding resected muscle tissue.

Macroscopic residual tumor was left in the parapharyngeal and prevertebral areas, which were marked with surgical clips.
Final histopathology showed a low-cellularity fibroblastic proliferation infiltrating fibro-adipose and striated muscle tissue. The spindle-shaped tumor cells were arranged in collagen bundles and encased adjacent nerves. Immunohistochemical staining revealed nuclear positivity for β-catenin.
As the patient had an R2 resection, adjuvant radiotherapy was administered shortly after surgery to control residual disease. The patient remained stable without recurrence at the two-year follow-up.

DISCUSSION:

Desmoid tumors originate from the musculoaponeurotic structures and are classified as deep-type fibromatoses, characterized by benign, locally aggressive fibroblastic proliferation without metastatic potential. It's a distinctly rare tumor with incidence of 5–6 cases per 1 million of the population per annum (1). Based on their localization, desmoid tumors are categorized into three forms: intra-abdominal, abdominal wall, and extra-abdominal types. Head and neck desmoid tumors are estimated to constitute only 7% to 15% of all desmoid tumors throughout the body (2).
Desmoid-type fibromatosis typically occurs as a sporadic lesion, as illustrated in our case. However, a hereditary form may also be observed, particularly in association with familial adenomatous polyposis (FAP) as part of Gardner syndrome. The etiology of desmoid tumors remains poorly understood. However, certain factors, such as a history of local trauma and previous operations, have been strongly associated with their development (3)
Additionally, hormonal influences have been suggested, with higher incidence reported during pregnancy, in the postpartum period, and after the use of oral contraceptives (4).
The clinical presentation of desmoid tumors can be misleading. Initial symptoms are often nonspecific, including an unexplained lump, pain, or fatigue. In the study by Lucas et al., the average time from symptom onset to definitive diagnosis was approximately four years (5).
Computed tomography CT scan and MRI imaging of the lesion is important to determine the locoregional extension and resectability of the tumor. On a CT scan imaging the tumor is well-limited isodense, homogeneously enhanced after injection of contrast product (4). MRI is preferred for an adequate assessment as it's the mainstay of imaging in the desmoid tumors. The lesion is homogeneous, hypointense or isointense to muscles on T1-weighted images. In T2-weighted sequence it appears more heterogeneous and mainly hyperintense, in the event of high cellularity and water content of extracellular spaces. T2 weighted hypointense bands may be seen representing excessive collagen accumulation (4).

A desmoid tumor could be diagnosed only by a biopsy for histological examination. It is important to use nuclear positivity for beta-catenin to eliminate a true sarcoma.

However, Nuclear staining for β-catenin is not definitive of the diagnosis of desmoid fibromatosis (6).

The management of desmoid tumors is not standardized. Therefore, treatment decisions should be made in a sarcoma reference center or within a multidisciplinary tumor board experienced in desmoid tumors (7).

A watch-and-wait approach is recommended for asymptomatic patients or those with mild symptoms . Ongoing surveillance should include a first MRI within 1 to 2 months, and subsequent evaluations every 3 to 6 months thereafter (1).

However, surgical resection remains the treatment of choice for symptomatic or progressively enlarging tumors (4). The importance of resection margins remains unclear. Since recurrence and postoperative morbidity are high, the emphasis should be on functional preservation rather than aggressive pursuit of wide margins (7). In that positive margins are acceptable if needed to maintain organ functions (4).

The role of adjuvant radiotherapy in desmoid-type fibromatosis remains debated. A retrospective meta-analysis including 1,295 cases showed improved local control after incomplete resection of primary extra-abdominal tumors when radiotherapy was added (6). Consequently, owing to the high local recurrence rate (30–40%), adjuvant radiotherapy is frequently employed alongside surgery to enhance local control (2).

Radiotherapy is an effective primary treatment for patients with high risk for surgery or who refuse it (7).

Primary radiotherapy (50–60 Gy) could reach 70–80% of long-term local control of desmoid tumors (6). According to the global consensus-based guideline from the Desmoid Tumor Working Group, the absolute risk of disease progression following radiotherapy alone (19%) was lower than after surgery alone (29%) (1).

Chemotherapy could be used especially for patients unsuitable for surgery. Bhati et al. reported that methotrexate and vinblastine could make a significant tumor regression (3).

In recent years, targeted therapies have emerged as promising treatment options for desmoid-type fibromatosis (DTF), particularly for unresectable or progressive disease.

Gounder et al. conducted a phase III, international, double-blind, randomized, placebo-controlled trial evaluating nirogacestat in adults with progressive desmoid tumors. Nirogacestat demonstrated significant improvements in progression-free survival, objective response rate, pain, symptom burden, physical and role functioning, and health-related quality of life compared with placebo (8).

Predictive factors of recurrence include the bony involvement. Tumors arising from bone structures—such as the mandible, maxilla, or cranial base—are more prone to recurrence (24%of all recurrences). Extra-abdominal tumors had a higher recurrence risk compared to abdominal ones (9).

A high PCNA index has been reported in recurrent tumors in some series. McLean et al. reported that tumors with abnormal β-catenin nuclear accumulation exhibited a significantly higher PCNA labeling index compared with those showing normal staining patterns (10). However, PCNA has not been reliably established as an independent predictor of recurrence in desmoid-type fibromatosis.

The size of the tumor, particularly > 7 cm, is associated with higher recurrence rate.

Sex women have higher rates of local recurrence), and age (patients under 37 years old) (6).

CONCLUSION

Desmoid tumors originating from the trapezius muscle are extremely rare. Although these lesions are benign and lack metastatic potential, their locally infiltrative behavior can lead to significant morbidity and incomplete surgical resection.

Management of DT should rely on a multidisciplinary approach, involving surgeons, radiotherapists, and oncologists, to balance disease control with functional preservation.

Ethical Considerations: Written informed consent was obtained from the patient.

Conflict of Interest Statement: The authors declare that they have no conflicts of interest related to this article.

REFERENCES:

1. Alman B, Attia S, Baumgarten C, Benson C, Blay JY, Bonvalot S, et al. The management of desmoid tumours: A joint global consensus-based guideline approach for adult and paediatric patients. Eur J Cancer. 2020;127:96-107.
2. Kant S, Charan BD, Goel V, Das S, Sahu S, Sharma R, et al. Desmoid-type fibromatosis of neck masquerading as nerve sheath tumors: two case reports. Egypt J Radiol Nucl Med. 2023;54(1):176.
3. Bhati M, Balakrishna G, Joshi K, Bhattacharya K, Bal M, Ghosh Laskar S, et al. Fibromatoses of Head and Neck: Case Series and Literature Review. Rambam Maimonides Med J. 2021;12(3):e0022.
4. Harouachi A, Ramdani A, Kharkhach A, Akouh N, Bouhout T, Bennani A, et al. Desmoid tumor of trapezius muscle: A case report. Ann Med Surg. 2021;72:103127.
5. Lucas A, Zhou S, Hernandez L, Bell T, Oton AB, Mercier K. The impact of desmoid tumors: insights from the Desmoid Tumor Research Foundation Natural History Study, 2017–2023. Orphanet J Rare Dis. 2025;20(1):515.
6. Bautista-Pérez IJ, Luna-Peteuil Z, Garcia-Ortega DY, Luna-Ortiz K, Caro-Sanchez CHS. Desmoid Tumor of the Posterior Neck: A Case Report. Indian J Otolaryngol Head Neck Surg. 2023;75(2):1157-61.
7. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®): Soft Tissue Sarcoma. Version 2.2025.
8. Fort Washington, PA: National Comprehensive Cancer Network, Inc.
9. Available from: https://www.nccn.org
10. Gounder M, Ratan R, Alcindor T, Schöffski P, Graaf WT van der, Wilky BA, et al. Nirogacestat, a γ-Secretase Inhibitor for Desmoid Tumors. N Engl J Med. 2023;388(10):898-912.
11. Lazcano CS, Gronchi A. Surgical Management of Desmoid Tumors-Patient Selection, Timing, and Approach. Curr Oncol Tor Ont. 2025;32(7):408.
12. McLean TD, Duchi S, Di Bella C. Molecular Pathogenesis of Sporadic Desmoid Tumours and Its Implications for Novel Therapies: A Systematised Narrative Review. Target Oncol. 2022;17(3):223-52.