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Cervical Desmoid-Type Fibromatosis in an Adolescent: A Rare Imaging Case

J. Ait Si Abdessadeq[1*], F. Akhatar[1], S. Ouassil[1], Hc. Ahmanna[1], B. Zouita[1], D. Basraoui[1], H. Jalal[1]

[1]Department of Radiology, Mother-and-Child Hospital, Mohamed VI University Hospital, Faculty of Medicine and Pharmacy, Cadi Ayyad University, Marrakech, Morocco

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***Corresponding author:** J. Ait Si Abdessadeq
Department of Radiology, Mother-and-Child Hospital, Mohamed VI University Hospital, Faculty of Medicine and Pharmacy, Cadi Ayyad University, Marrakech, Morocco

Abstract **Case Report**

Desmoid-type fibromatosis is a locally aggressive tumor characterized by a well-differentiated proliferation and a high risk of recurrence. It is classified into extra-abdominal, abdominal, and intra-abdominal subtypes based on its location. Although rare, cervical desmoid fibromatosis is an even less common manifestation. Furthermore, its radiological features are seldom reported in the literature, making differentiation from other cervical masses particularly challenging. We report an unusual case of a 16-year-old female patient, who presented with a progressively enlarging, painless mass on the left side of her neck over the past six months. Further imaging and histopathological evaluation were performed, leading to the diagnosis of cervical desmoid fibromatosis.

Key words: DESMOID – FIBROMATOSIS – NECK – CT – IMAGING.

INTRODUCTION

Desmoid-type fibromatosis or musculoaponeurotic fibromatosis is a locally aggressive, non-metastasizing tumor characterized by a well-differentiated, unencapsulated monoclonal proliferation of myofibroblasts, with a strong tendency for local invasion and recurrence [1]. It is considered an intermediate entity between fibroma and fibrosarcoma [1]. The etiology is multifactorial, involving traumatic, hormonal, and genetic factors particularly mutations in the *CTNNB1* gene [2,3].

The annual incidence of desmoid fibromatosis is estimated to be 0.2 to 0.4 per 100,000 individuals [4]. Of all cases, 7 to 15% occur in the head and neck region [5-7].

Due to their deep location, infiltrative growth into adjacent subcutaneous tissue or muscle, and myxoid or fibrotic composition, they can resemble malignant soft tissue tumors, making differentiation based solely on imaging findings challenging. We report a 16-year-old female with a painless, enlarging neck mass over six months, diagnosed as cervical desmoid fibromatosis after imaging and histopathological evaluation.

OBSERVATION

A 16-year-old female patient presented in our department with a progressively enlarging, painless mass on the left side of her neck over the past six months. There was no history of trauma, fever, weight loss, or systemic symptoms. The mass had gradually increased in size, causing mild discomfort but no significant pain or neurological deficits.

On physical examination, a firm, non-tender, poorly mobile mass was palpated in the left lateral cervical region. There were no signs of skin discoloration, or overlying inflammation. Neurological examination was unremarkable, and there was no evidence of airway compression or dysphagia.

The patient had no significant medical or family history of similar conditions. Routine lab workup was unremarkable.

An initial ultrasound was performed, revealing a heterogeneous echogenic lesion with color Doppler vascularization, displacing the vascular axis of the neck, and associated with nearby lymphadenopathy. Based on these findings, a contrast-enhanced cervical CT scan was performed.

The CT scan showed a left lateral cervical mass extending into the anterosuperior mediastinum, well-defined, with heterogeneous density on spontaneous contrast, containing punctate calcifications, and mildly heterogeneously enhanced with contrast. The mass measured approximately 10.6 x 6 x 5 cm (CC x T x AP). (FIGURE 1)

Topographically, it was in contact with the left sternocleidomastoid muscles anteriorly and extended downward to the inner surface of the first, second, and third left intercostal spaces, with partial loss of the fat interface separating them, without any adjacent bone lysis.

Medially, it displaces the left thyroid lobe and the aerodigestive tract, without any detectable endoluminal mass. Posteriorly, it causes scalloping of the anterior surface of the posterior arch of the left first rib without any adjacent bone lysis. Laterally, it reached the subcutaneous cervical adipose tissue and enveloped the internal jugular vein, which was thrombosed.

Inferiorly, it is in contact with the aortic arch, encased the left common carotid artery over a 360-degree circumference, and also enveloped the left subclavian artery, both of which remained patent.

There were also left jugulocarotid cervical lymphadenopathies associated, with the largest one on the right measuring 14 x 8 mm.

The patient underwent an additional abdominal ultrasound to screen for an associated abdominal localization, which revealed no abnormalities.

A surgical biopsy was performed, confirming the diagnosis of cervical desmoid fibromatosis. Given the complex anatomical relationships of the mass and its high recurrence potential, a conservative treatment approach was chosen. Over a one-year period, the evolution was without any notable complications.

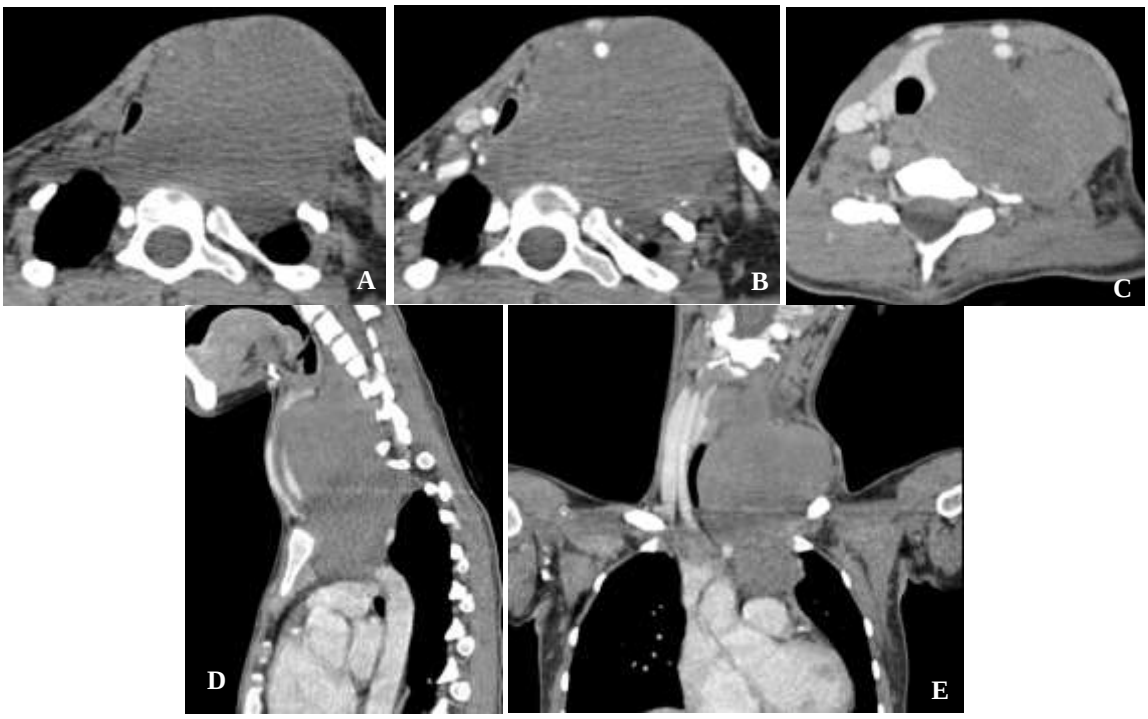

Figure 1: Cervicothoracic CT scan with axial section without contrast injection (A), axial section with contrast injection in the arterial phase (B), axial section in the venous phase (C), sagittal section in the venous phase (D), and coronal section in the venous phase (E), illustrating a left cervical lesion extending into the superior mediastinum, locally infiltrative, and heterogeneously enhanced by contrast, consistent with desmoid-type fibromatosis

DISCUSSION

According to the World Health Organization's classification of head and neck tumors [5], desmoid-type fibromatosis is classified as a borderline soft tissue tumor with low malignant potential. It exhibits local aggressiveness, with an estimated 20% recurrence rate, but does not metastasize.

Desmoid tumors are found in the head and neck region in 7% to 15% of cases [5-7], with 57% of these occurring in the pediatric population. The incidence of pediatric desmoid tumors peaks around the age of 8 [8]. No gender predilection has been reported. The most common presenting symptom of head and neck fibromatosis is a painless, enlarging mass [9]. The mandible is the most frequently affected site, followed

by the submandibular region, neck, tongue, and paranasal sinuses, respectively [10,11].

The exact cause of desmoid fibromatosis is unknown, but it is likely multifactorial such as traumatic and genetic factors, particularly mutations in the *CTNNB1* gene [2,3]. Spontaneous tumor regression in females during menarche and menopause, along with reports of responses to antiestrogen therapy, suggest that hormonal factors may influence tumor growth [9].

Regarding the diagnostic approach in imaging, the radiological characteristics of desmoid fibromatosis have been rarely reported in the literature, with the exception of a few case reports, particularly the criteria for differentiating this entity from other cervical masses, as well as the radiological and pathological correlation. However, it has been stated that CT and MRI are the preferred imaging techniques, providing crucial information on the extent of the disease and involvement of vital structures. Imaging findings can vary depending on the cellularity and collagen content of the lesions [12], with lesions rich in collagen deposits revealing a higher attenuation on both precontrast and postcontrast CT-images. On MRI, the lesion typically displayed high signal intensity on T2-weighted images and iso signal intensity on T1-weighted images, with significant enhancement [13,14]. However, varying amounts of cellular tissue, myxoid changes, and collagenous stroma may exhibit different signal intensities. Low signal intensity on T2-weighted images is associated with lower cellularity and myxoid changes, as well as increased collagenization.

Among the radiological characteristics of cervical desmoid fibromatosis reported in the study by Rhim *et al*. [12], it was noted that a rim of surrounding fat is a common feature of desmoid-type fibromatosis in the head and neck, particularly in cases involving the peri-vertebral space. This characteristic occurs because the tumor arises from musculoaponeurotic or fascial structures, which are typically surrounded by fat, even as the mass infiltrates the underlying muscles.

The differential diagnosis of desmoid-type fibromatosis in the head and neck region includes malignant tumors such as malignant fibrous histiocytoma, fibrosarcoma, rhabdomyosarcoma, and metastases, due to the occasionally aggressive features of this condition. It also includes solitary fibrous tumors, as they may exhibit the fibrotic low signal intensity seen on T2-weighted images [15]. Additionally, desmoid-type fibromatosis can resemble neurogenic tumors, like neurofibromas and schwannomas, because of the elongated appearance present in some lesions [12].

In addition to its rare incidence and diagnostic difficulties, the structural complexity, proximity to vital structures, and infiltrative growth characteristics of desmoid-type fibromatosis in the head and neck region pose challenges for effective treatment.

A paper on the European Organisation for Research and Treatment of Cancer (EORTC)/Soft Tissue and Bone Sarcoma Group's stance on DF stated that the "watch and wait strategy" is the preferred initial approach for treating desmoid fibromatosis in all populations. It also suggested that resection with clear margins may be considered as a treatment option if postoperative morbidity is deemed acceptable [16].

CONCLUSION

In conclusion, desmoid-type fibromatosis remains a diagnostic challenge due to its deep location, infiltrative nature, and imaging similarities to malignant soft tissue tumors. Although rare in the head and neck region, its potential for local recurrence necessitates careful evaluation and long-term follow-up. This case highlights the importance of imaging analysis for accurate diagnosis, allowing for appropriate management strategies.

Conflicts of interest: Authors declare no conflict of interest.

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