CASE REPORTS

GIANT INTRATHORACIC DESMOID TUMOR – A CASE REPORT

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Abstract

Desmoid tumors are soft tissue neoplasms arising from fascial and muscle-aponeurotic structure. These tumors are locally aggressive and have a high recurrence rate, even after complete resection. We present the case of a female with a giant intrathoracic desmoid tumor. She underwent complete surgical resection with no disease recurrence. Desmoid tumors' natural history is not well defined and is often enigmatic, making these tumors difficult to manage. Currently, for intrathoracic desmoid tumors, medical treatment is the recommended approach, nevertheless, surgery can be considered in selected patients.

Keywords: *Desmoid Tumor, Desmoid Fibromatosis, Thoracic Surgery*

INTRODUCTION

Desmoid tumors are soft tissue neoplasms arising from fascial and muscle-aponeurotic structure. These tumors are slow growing and histologically benign. However, they are locally aggressive with a tendency to recur[1], making the treatment of these lesions challenging.

CLINICAL CASE

A 19-year-old female presented with thoracic pain and fatigue, with progressive worsening in recent weeks, and declining exercise tolerance. No relevant previous medical history. Initial investigation with chest x-ray showed an opacity occupying two-thirds of the left hemithorax, followed by a CT scan, revealing a mass measuring 17.5x10.5x10.0 cm, occupying most of the left hemithorax, compressing the ipsilateral lung and causing mediastinal shift. A CT-guided biopsy was attempted however with inconclusive results.

A left thoracotomy was performed with excision of the mass, which was demanding due to size and presence of adherences to the parietal pleura, however, no vascular structures were involved. After, there was a complete lung expansion. Postoperative period was uneventful, and patient was discharged 9 days after the surgery.

On gross examination, the lesion was partially delimitated by a capsule with irregular surface areas; the cut surface was firm, white, and coarsely trabeculated. Microscopically, the tumor was formed by long sweeping fascicles of spindle cells on a collagenous stroma with thin-walled blood vessels with perivascular edema and scattered mast cells. On immunohistochemistry, the tumor cells diffusively expressed vimentin and beta-catenin, with focal positivity for the S100 protein. These results were compatible with desmoid tumor.

During the follow-up of 48 months there was no recurrence registered.

She maintains regular surveillance with the oncology team. No adjuvant radiotherapy was performed.

DISCUSSION

Desmoid tumors (DT) are rare, slow-growing tumors, accounting for 0.03% of all neoplasm.[2] They are known by many names, such as desmoid fibromatosis, aggressive fibromatosis, and desmoplastic fibroma. These tumors are defined by World Health Organization (WHO) as "clonal fibroblastic proliferation that arises in the deep soft tissues and is characterized by infiltrative growth and tendency towards local recurrence but an inability to metastasize".

Despite their benign appearance, they are locally aggressive and invade surrounding structures, sometimes being classified as low-grade fibrosarcoma.[3,4]

DT might arise in any location and are divided into three groups based on their anatomical location: abdominal, extra-abdominal, and intra-abdominal.[1]

They might present with an unpredictable clinical course and their natural history remains poorly understood.[3] In fact, it remains unclear why some tumors will continue to grow while others will remain stable or even regress with no treatment at all.[3,5]

DT occurs mainly between the age of 15 and 60 years and most occur sporadically.[6] Nevertheless, an association has been reported between DT and inherited syndromes, familial adenomatous polyposis (FAP) and Gardner's Syndrome.[7,8] There have been reports that pregnancy, trauma and surgery could act as predisposing factors for developing desmoid tumors.[1,2,5,7]

The clinical presentation depends on the location the tumor arises, most being asymptomatic when they grow in the trunk or extremities, and symptomatic when they grow into cavities with mass effect symptoms.1 Hence, intrathoracic tumors are expected to cause symptoms of lung compression, like in this case's patient.

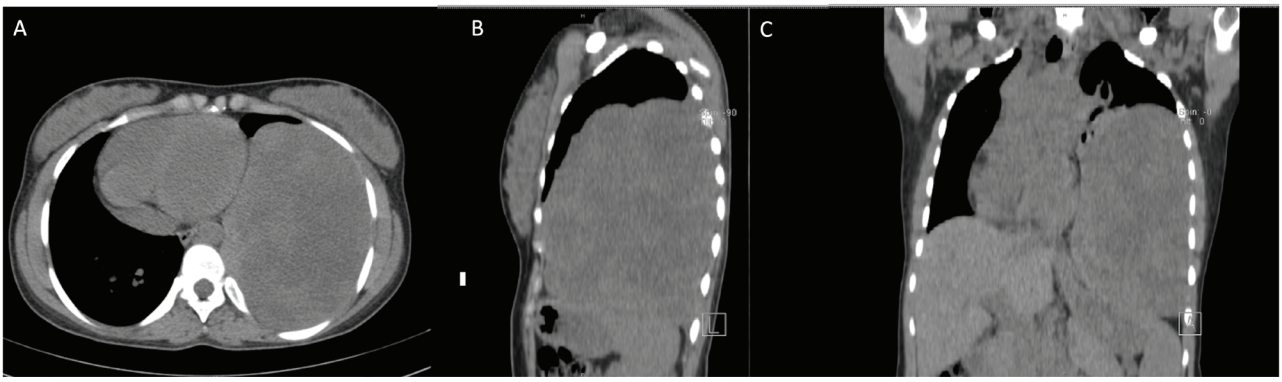

Figure 1 *Thoracic CT scan. A: Axial CT revealing a giant heterogeneous mass occupying almost completely the left hemithorax, with lung compression and slight mediastinal shift. Sagittal(B)and Coronal (C)CT scan.*

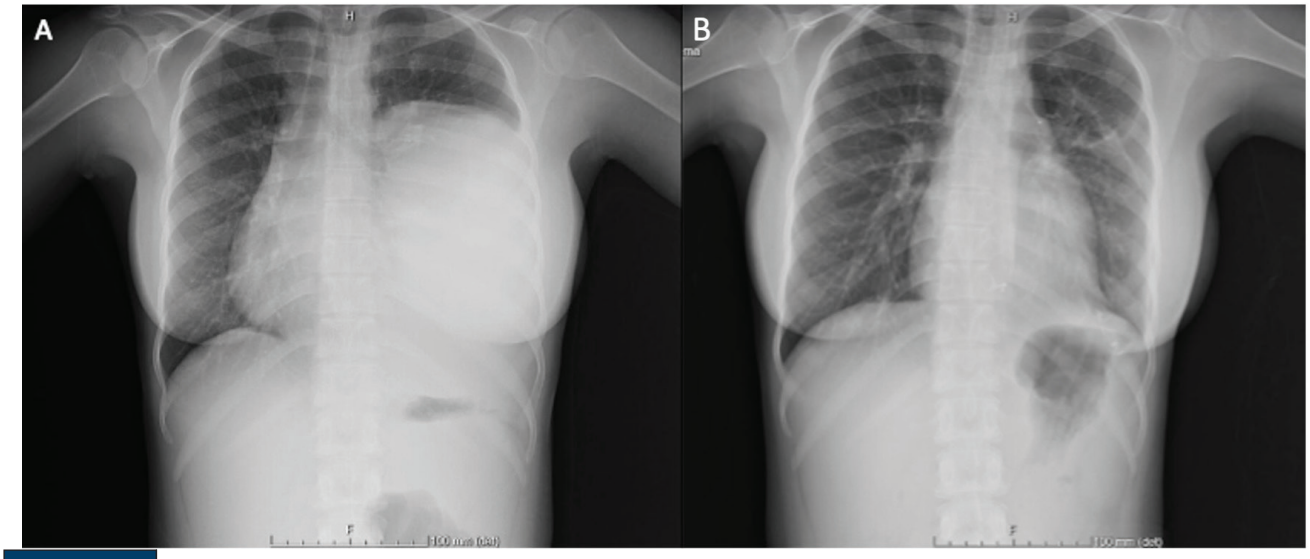

Figure 2 *Chest x-ray: A. Preoperative x-ray show opacity occupying 2/3 of left hemithorax. B. Post-operative x-ray demonstrating a complete left lung expansion.*

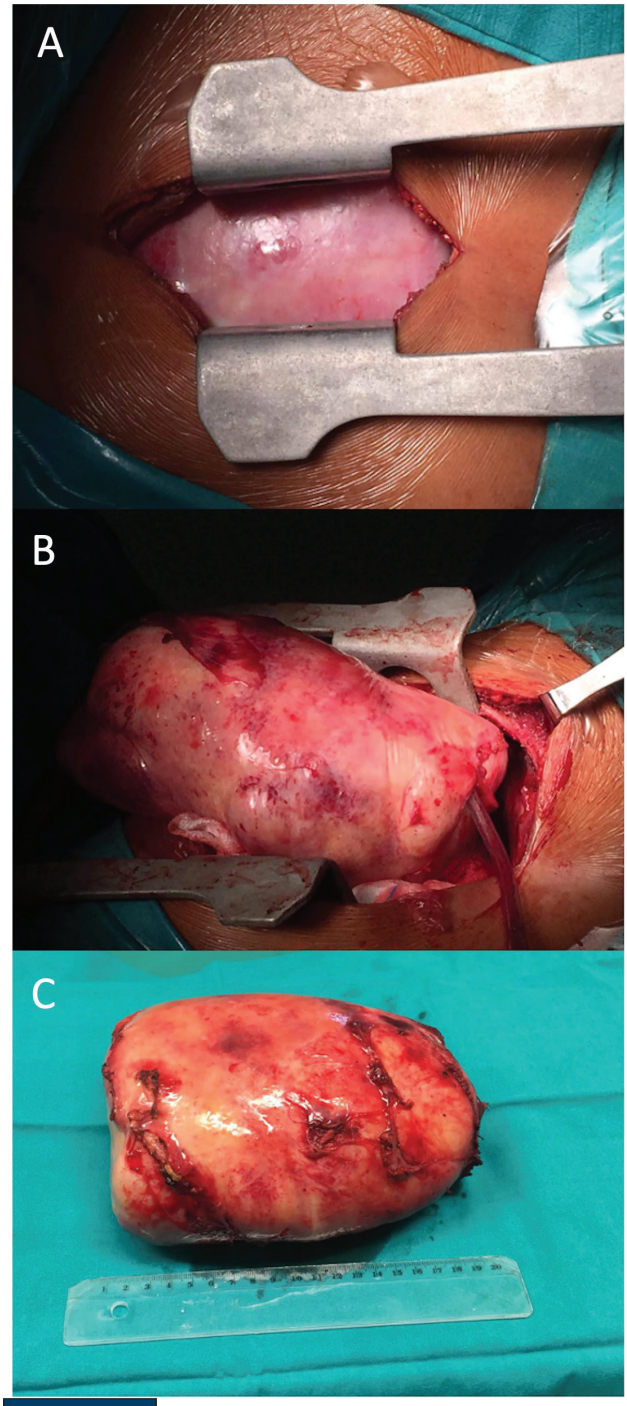

Figure 3 *A. Posterolateral thoracothomy with mass. B: Desmoid tumor during excision. C: Excised tumor.*

For imaging investigations, MRI is the mainstay choice7, however, CT scan might also be used.[1] In MRI, DT may have an heterogeneous signal and inhomogeneous enhancement, with key diagnostic feature being hypointense bands identifiable on T2-Weighted images.[1,7] CT scan shows a soft tissue mass of variable attenuation and enhancement, and margins might be difficult to distinguish given the infiltrative tendency of this tumors.[1]

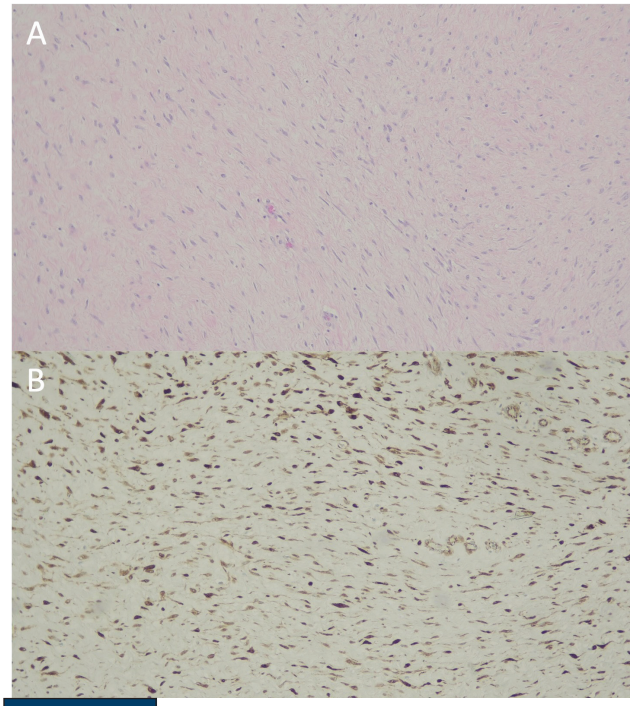

Figure 4 *A.Tumor composed by spindle cells with clear cytoplasm on a collagenous stroma. Cell atypia is minimal, and no mitoses are observed. B. Nuclear positivity for beta-catenine.*

Surgery with negative margins was considered the mainstay of treatment in the past. However, the consensus initiative between the Sarcoma Patients Euronet (SPAEN) and the European Organization for Research and Treatment of Cancer/Soft Tissue and Bone Sarcoma Group (EORTC/STBSG)[6,7] proposed that the first approach should be a conservative watch and wait for patients with histologic diagnosis of DT, to further understand the behavior of the disease and better define next treatment steps. There are reports that these tumors might stop growing and even regress.[3,6] Hence, it is recommended a watchful waiting for asymptomatic tumors for a period up to 1 to 2 years, with patients maintaining a close follow-up with MRI or CT scan.[1,7,8] The shift for active treatment depends on many factors, such as tumor size, growth rate, anatomical location, compression and worsening of function, but mostly is based on tumor size progression in multiple consecutive images.[7]

For intrathoracic tumors, medical therapy stands as the standard approach.[1,7] Systemic treatment options include non-steroidal anti-inflammatory drugs, antihormonal therapies, tyrosine kinase inhibitors, and chemotherapy.[1,3,7] When progression occurs and vital organs are at risk, surgery or radiotherapy are both options.

For patients with symptomatic resectable DT, surgery can be a valid option.[2,7,8]

The significance of positive margins following DT excision is limited, as various studies have presented conflicting data on their prognostic value.[7] Hence, the

primary objective should be achieving R0 margins, while also prioritizing the preservation of organ function and the patient's quality of life. In cases where achieving R0 margins would result in function loss or excessive morbidity, R1 margins can be deemed acceptable.[7,8] However, it is crucial to note that an aggressive approach should be avoided, and systemic therapy may be considered in such situations.[6] Additionally, postoperative radiotherapy can enhance local control and decrease the risk of recurrence after incomplete resections.[7,8]

CONCLUSION

Intrathoracic desmoid tumors remain challenging to manage. Starting with watchful waiting and upon progression or symptoms, a multimodal therapy should be tailored for each patient, with surgery, radiotherapy and medical therapy as valid options, never losing the focus of organ function preservation and patients' quality of life.

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