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The challenging management of a giant intrathoracic desmoid tumour: a case report

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Abstract

Desmoid tumours are rare, locally aggressive neoplasms exhibiting high tendency for recurrence, even after complete resection. Only 1 in 5 of them originates from the chest wall, usually measuring less than 10 cm at diagnosis. Herein, we report the case of a woman presenting with symptoms of gradual lung compression by a giant desmoid tumour occupying the entire hemithorax. She underwent complete surgical resection of the tumour and chest wall reconstruction. She had disease recurrence 15 months later and currently remains under regular follow-up. The management of intrathoracic desmoid tumours is challenging because they are usually not diagnosed until they become large enough to cause compression symptoms. While medical management is the primary modality of treatment, surgery could be considered in selected cases where significant symptoms arise, and the functional status is impaired secondary to the tumour. Adjuvant radiotherapy to minimise the risk of local recurrence should also be considered.

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Introduction

Desmoid tumours are monoclonal fibroblastic neoplasms arising from musculoaponeurotic structures. They account for only 0.03% of all neoplasms [1]; of these, less than 20% originate from the chest wall [2]. Although being considered benign, desmoid tumours are usually locally aggressive and have a high recurrence rate, even after complete resection. These features make the treatment of these lesions challenging. Herein, we present an interesting case of a woman who developed a giant intrathoracic desmoid tumour occupying an entire hemithorax, and we review the management of these neoplasms.

Case Report

A 33-year-old African-Caribbean woman presented with progressively increasing dyspnoea, deteriorating fatigue and declining exercise tolerance over the last month. She had an unremarkable past medical history. She was initially investigated with a chest radiograph showing complete opacification of the left hemithorax, followed by computed tomography (CT) scan of the thorax, which revealed a giant mass traversing the left chest wall and invading the hemithorax, resulting in compression of the entire ipsilateral lung and significant midline shift.

A CT-guided biopsy of the tumour demonstrated features suggestive of pleural fibroma; however, a sarcomatoid component could not be overruled. Subsequently, positron emission tomography with fluorodeoxyglucose integrated with CT (PET-CT), from

the cranial vertex through the mid thighs, showed mild, peripheral tracer uptake by the lesion (Figure 1), without any evidence of lymphadenopathy or metastatic foci.

The patient underwent lateral thoracotomy and excision of the tumour, along with resection of 4 ribs and involved intercostal tissues. Immediately after the removal of the mass, the left lung fully expanded. Chest wall reconstruction was performed with Strattice™ tissue matrix (Allergan, Dublin, Ireland) (Figure 2A). She was discharged 5 days later after an uncomplicated postoperative course.

The specimen measured 24.5cm x 18.3cm x 9.7cm and weighed 1852g (Figure 2B). On macroscopic examination, it had a light tan, firm, homogenous surface covered by thin fibrous membrane. It was partially encasing four consecutive ribs without, however, infiltrating the osseous tissue. Microscopic examination revealed long sweeping fascicles of bland spindle cells. The spindle cells showed pale eosinophilic cytoplasm with tapered nuclei and were distributed in a collagenous stroma with many thin-walled blood vessels (Figure 3). Mitotic activity, cytological atypia and necrosis were not present. The tumour abutted the specimen ribs but did not infiltrate them. Immunohistochemistry was negative for the following markers: SMA, Desmin, S100, MUC4, CD34, STAT6, MNF116, EMA.

The patient remained free of disease for 15 months following her discharge. An interval PET- CT scan demonstrated local recurrence in the left second rib measuring 4.2cm x 2.8cm x 4.3cm, with mild tracer uptake. She currently remains under regular surveillance by the Oncology team and is being treated with non-steroidal anti-inflammatory drugs. No adjuvant radiotherapy has been administered.

Discussion

Desmoid tumours appear in the literature with various names, including desmoid-type fibromatosis, aggressive fibromatosis and musculoaponeurotic fibromatosis, but they all denote a rare type of myofibroblastic neoplasm. World Health Organisation defines them as clonal fibroblastic proliferations originating from deep soft tissues. Regarding their biological behaviour, they are classified as intermediate soft tissue tumours: they are locally invasive and exhibit high tendency to recur; they do not display metastatic potential [3]. Their incidence is estimated at 2–5 cases per million people per year, with small female preponderance and maximum occurrence between 20 and 44 years of age [4]. Most desmoid tumours arise sporadically. However, inherited syndromes, mainly familial adenomatous polyposis and Gardner's syndrome, have been closely related with desmoid-type fibromatosis, accounting

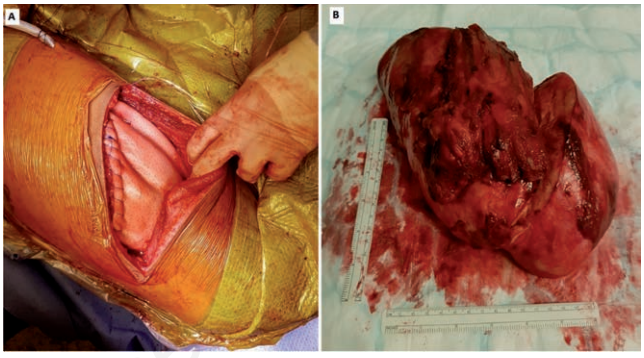

Figure 2. A) Final operative result demonstrating removal of the mass and reconstruction of the chest wall using Strattice™ tissue matrix. B) Soft tissue mass of the left hemithorax removed along with infiltrated ribs and intercostal tissues, cut in two pieces to facilitate removal.

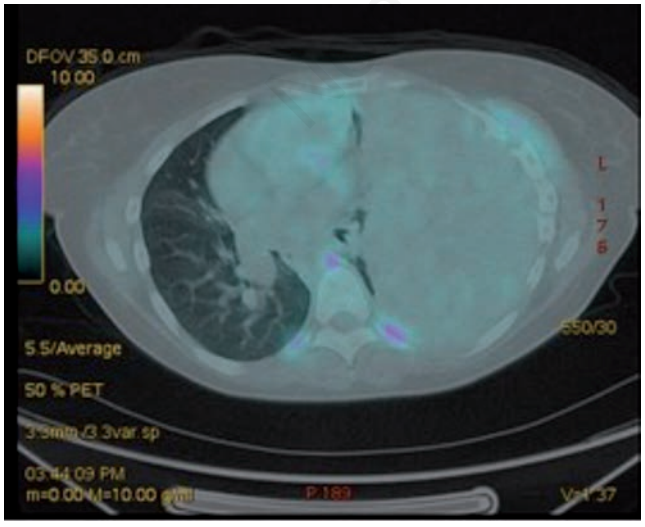

Figure 1. Positron emission tomography demonstrating the left hemithorax being occupied by a huge mass of soft-tissue density causing significant mediastinal shift that shows mild peripheral uptake of fluorodeoxyglucose.

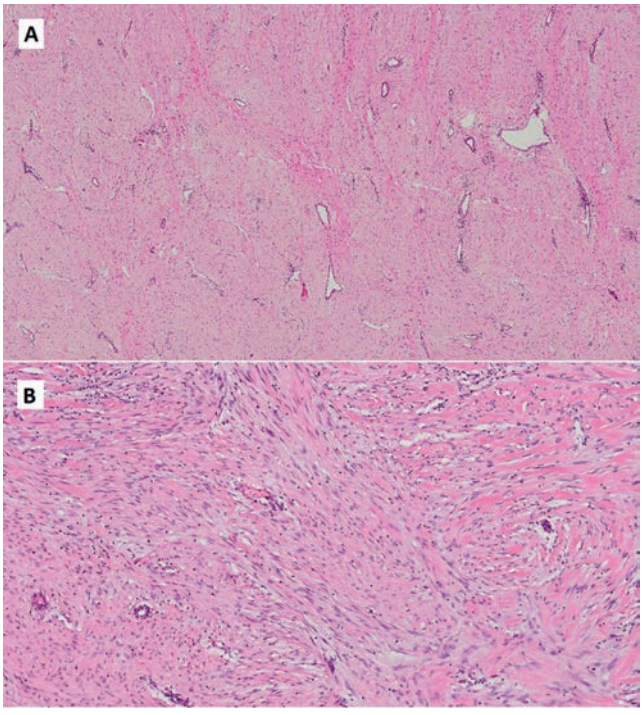

Figure 3. A) Haematoxylin & Eosin (x 20 magnification): low-power view showing lesion with collagenous stroma and interspersed thin-walled blood vessels. B) Haematoxylin & Eosin (x 80 magnification): long sweeping fascicles of bland spindle cells within collagenous stroma, with scattered inflammatory cells.

for 5–10% of all cases and leading mostly to mesenteric desmoid tumours. Endocrine factors, pregnancy, trauma and surgery have been recognised as predisposing factors for developing desmoid tumours [5,6].

The clinical presentation of desmoid tumours varies based on the region of occurrence. When they grow in the trunk or the extremities, they are usually asymptomatic. When they develop within body cavities, they can present with mass effect symptoms [3]. Therefore, intrathoracic desmoid tumours are expected to present with lung compression symptoms, such as dyspnoea, which depend on the size of the tumour and the respiratory reserve of the patient. Interestingly, our young and otherwise healthy patient did not seek medical advice until the tumour occupied her entire hemithorax.

The most useful imaging investigations to achieve diagnosis comprise CT and magnetic resonance imaging (MRI). CT usually demonstrates an enhancing soft tissue mass with variable attenuation and vague margins [4]. On MRI, the most prominent characteristics include an ovoid or irregular mass that does not respect fascial compartments and is isointense on T1-weighted images and heterogeneously hyperintense on T2-weighted images. Occasionally atypical bone changes may be present [7]. The main differential diagnosis comprises of malignant soft-tissue sarcoma, extranodal lymphoma, benign myositis ossificans and arteriovenous malformation [4].

On gross pathological examination, desmoid tumours are usually limited in the musculature, the overlying aponeurosis and fascia. Occasionally, these neoplasms tend to penetrate surrounding structures, such as adipose tissue, periosteum and bone. The usual dimensions of these neoplasms vary between 5 and 10 cm, although there are reports of larger tumours [8,9]. In our case, the tumour measured nearly 25 cm in its largest diameter, thereby representing one of the largest intrathoracic desmoid tumours ever reported. On cross section, the cut surface of desmoid tumours appears to be glistening white and coarsely trabeculated, resembling scar tissue [10]. On microscopic examination, they are usually ill-defined invading neighbouring soft-tissue structures (usually skeletal muscles). They are characterised by elongated, slender, spindle-shaped cells, arranged in fascicles, embedded in fibrous, focally hyalinised or keloidal collagenous stroma. The cells are not hyperchromatic, they do not show signs of atypia and have small, pale-staining nuclei with 1-3 nucleoli. Regarding immunohistochemical markers, the cells usually express vimentin, beta-catenin and occasionally smooth muscle actin. Rarely, cells that express desmin and S100 protein are detected [3].

In the past, the mainstay in the management of desmoid tumours was immediate surgery; similar to that of soft tissue sarcomas. The European consensus initiative between the Sarcoma Patients Euronet (SPAEN) and the European Organisation for Research and Treatment of Cancer / Soft Tissue and Bone Sarcoma Group (EORTC/STBSG), published in 2015 [11] and revised 2 years later [12], suggest a fundamental change in the management of histopathologically diagnosed desmoid tumours towards an initial period of watchful waiting in order to record potential tumour progression. Retrospective case series have demonstrated that 50% of asymptomatic individuals managed conservatively with close observation remained progression-free at 5 years from diagnosis [13-15]. Moreover, spontaneous regressions have been detected in up to 30% of cases [16]; more commonly in the abdominal wall [17] but have been observed at all sites [18]. Therefore, watchful waiting as the first approach is considered appropriate for asymptomatic tumours developing near critical structures. The timeframe for this type of approach could be 12–24 months, and patients should remain under close surveillance with contrast enhanced MRI. However, not only the lack of specific criteria to define who would require active therapy at the time of diagnosis, but also the tumour site, size and growth rate complicate the process of decision making. In cases of clearly progressing disease (e.g., in multiple consecutive images), compression of surrounding organs, risk to vital structures and deterioration of function, multimodal therapy needs to be administered on an individual basis. Location, resectability, and hormonal and molecular profile of the tumour, as well as functionality of the surrounding tissues, need to be taken into account for achieving disease control with acceptable risk of morbidity and mortality [19,20].

For desmoid tumours of the chest wall, according to recommendations by the SPAEN and EORTC/STBSG [12], the decision for the type of treatment should be directed by the expected postoperative morbidity and functional impairment. An observation strategy should be implemented until vital structures are at risk of involvement, in which case surgical resection, medical therapy or radiotherapy should be considered as equal alternative options. The aim of surgery is to obtain resection margins free of tumour, without significant functional loss and with acceptable cosmetic outcome. In the event of positive resection margins or critically located lesions, adjuvant radiotherapy may be considered for further management.

For intrathoracic desmoid tumours, medical treatment is widely considered as the standard approach [12]. Medical treatment includes antihormonal therapies, non-steroidal anti- inflammatory drugs, tyrosine kinase inhibitors and chemotherapy with methotrexate and/or vinblastine or vinorelbine or an anthracycline-based regimen. In case of rapid disease progression or threatening of vital organs, radiotherapy is an effective alternative treatment. When surgery is elected, it is advisable to administer adjuvant radiotherapy to reduce the probability of recurrence. In our case, surgical resection was performed due to the significant tumour size and related symptoms. The patient subsequently preferred to undergo surveillance and not receive radiotherapy, after being informed of the risks and benefits of such a management plan.

In conclusion, intrathoracic desmoid tumours present at a later stage compared to their counterparts in other anatomic locations where they can be palpated. This is because they will not cause symptoms until their size is significant enough to compress the ipsilateral lung causing dyspnoea, as in our case, or to locally invade the chest wall or surrounding structures causing pain. In these cases, surgery is generally recommended, with consideration of additional radiotherapy to minimise the risk of local relapse.

Conclusions

The management of intrathoracic desmoid tumours can be rather challenging due to their potentially significant size at the time of diagnosis and their tendency for local recurrence after surgical resection. Watchful waiting, surgery, radiotherapy and multimodal therapy are valid treatment options that should be individualised based on symptomatology, functional status, expected cosmetic results and patient preferences.

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