A Rare Intrathoracic Desmoid Type Fibromatosis Tumor: A Case Report

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Case Report

In August, 2013, a 76 year old African American female presented to her family physician with chest pain. A computed tomography (CT) scan identified a 4 cm, well-circumscribed mass of the left lung. Subsequently, on August 23, 2013, the patient underwent a left lung CT-directed fine needle aspiration (FNA) and core biopsy of the mass. An immediate adequacy check of the FNA revealed scant cellularity, with rare fragments of connective tissue, and as a result, the pathologist requested core biopsies. The cores showed proliferation of uniform spindle cells in a collagenous background. The pathologist rendered a diagnosis of spindle cell tumor and recommended resection for definitive subclassification. However, the patient did not return until February, 2014, at which time a follow-up CT scan showed the mass to have increased in size, to 17 cm (Figure 1). A second CT-directed needle core biopsy of the left lung was performed and the formalin fixed cores once again showed a proliferation of spindle cells in a collagenous background, with fairly uniform nuclei, no mitotic figures or necrosis. Because of the unusual clinical and pathological features of the case, it was sent for outside consultation. The consultant felt the tumor was most consistent with a desmoid type fibromatosis, based on the immunological stains, which showed the tumor cells to be positive for beta-catenin (nuclear and cytoplasmic), SMA and desmin, and negative for MAK-6, S-100, CD34 and estrogen receptor.

On April 1, 2014 the patient underwent a left pneumonectomy with tumor resection. The specimen had a total weight of 1955 grams. A small portion was processed for frozen section, which was determined to be positive for skeletal muscle involvement. The tumor had increased in size, from 4.0 cm when first identified in August, 2013, to 19.0 x 14.7 x 11.0 cm, at the time of
Resection in April, 2014. A total of 10 lymph nodes were resected with the case, all of which were negative for tumor. Grossly, the tumor was light tan-pink, dense, and rubbery in appearance, and adherent to the antihilar aspect of the 22.0 x 15.0 x 3.5 cm lung (Figure 2). The tumor was encased by a thin tan-gray capsule and the interface between the lung and tumor was well-demarcated. Per orientation by the surgeon, the superior surface was the chest wall margin and the second surgical margin was the posterior/aortic margin. Both were marked with ink.

![Figure 2: Bivalved desmoid tumor adherent to and compressing left lung parenchyma but not invading into the lung parenchyma.](image)

Histologic examination of the slides revealed an appearance similar to that of the tumor in the previous biopsies, with small uniform spindled cells (Figure 3). The tumor involved the superior chest wall margin, and inked margin of resection adjacent to the aorta, but did not invade the parenchyma of the lung.
As of today, the patient is recovering well from surgery. She is currently in discussion with her thoracic surgeon and radiation therapy team as to whether she should proceed with adjuvant therapy or “wait and see”.

DISCUSSION

Desmoid tumors were first described by John Macfarlane in 1832.\(^1\) It is a soft tissue tumor that arises from connective tissue, which provides protection to organs, such as lung, liver and heart, and strength and flexibility to bones, ligaments, muscles.\(^2\) Desmoid tumors are highly uncommon, accounting for approximately 3.5% of fibrous tumors, 0.3% of all solid tumors,\(^1\) and less than 0.03% of all neoplasms.\(^3\) The estimated incidence in the United States is 2-4 cases per million people per year,\(^4\) which is about 900 cases.\(^2\) Desmoid tumors are more commonly found intra-abdominally; however extra-abdominal sites do occur, and include chest wall, extremities, head and neck.\(^1,3\) Desmoid tumors of the chest wall account for approximately 20% of all desmoid tumors, however, only 26 case reports of intrathoracic desmoid tumors have appeared in English literature\(^1\) and only 12 cases protruding into the pleural cavity.\(^3\)

Desmoid-type fibromatosis (DTF) represents a rare tumor which is borderline between nonaggressive fibrous tumors and low-grade fibrosarcomas. They usually present as a large infiltrative mass\(^5\) with indistinct boundaries that blend imperceptibly with the surrounding soft tissue.\(^6\) The infiltration of cortex, nerves and blood vessels in DTF is not uncommon.\(^7\) These tumors have a high potential to recur locally\(^8\) without metastasis.\(^3\) Weiss and Goldblum reported the rate of local recurrence to be 40%,\(^8\) while some others as high as 65%.\(^7\)
On gross appearance, these tumors can range in diameter from 1-15cm, have a gray white gritty cut surface and are deceptively circumscribed. Upon microscopic examination, however, the plump fibroblasts are arranged in broad sweeping fascicles, which infiltrate irregularly into the adjacent tissue. DTF tumors have numerous evenly dispersed venule-sized vessels with thick walls and open and distinct lumens, rather then compressed. Mitoses, nucleur atypia and necrosis is rare. Due to the indistinct boundaries of the DTF, while surgical excision may remove all visible tumor, it rarely completely excises the lesion, leaving behind a positive microscopic margin, with frequent recurrence.

Desmoid tumors can occur at any age, but are more frequent from teens to 30s, and occur in almost any part of the body. They occur more commonly in women (2:1), with no significant racial or ethnic allotment. The cause of the desmoid tumor is uncertain. Speculated etiologies include previous trauma, such as surgical scars - as they have been reported to arise in post mastectomy sites; hormonal, due to the fact that they commonly appear in young women during or after pregnancy; and genetic factors, such as patients with Gardner syndrome, or FAP (familial adenomatous polyposis) mutations in the APC or B-catenin gene. These tumors tend to occur within the mesentery or pelvic wall. Desmoid tumors may be disfiguring, disabling, and/or painful. They can be slow growing or aggressive, producing life threatening problems or death by compressing or invading vital organs.

The differential diagnosis to the desmoids-type fibromatosis is a solitary fibrous tumor (SFT). Like the DTF, they are composed of cytologically bland spindle cells. However, unlike the DTF, SFT’s are composed of disorderly bipolar spindle cells and collagen fibers in a disorganized arrangement. They have a rich vascularity, similar to DTF, however the vessels assume an irregular branched appearance. The SFT has a uniform, white-gray firm cut surface. The number one distinguishing factor between the two lesions is that while the SFT demonstrates a positive immunohistological analysis for CD34, the DTF does not.

To date, no malignant transformation of DTF has been recorded among intrathoracic cases. Similar to extra-abdominal DTF, intrathoracic DTF have an unpredictable natural course and can vary from growth arrest, to spontaneous remission, to multiple recurrences. While some clinicians recommend surgical management of desmoid tumors, others have decided on the “wait-and see” approach in some of their patients. Adjuvant therapy may be considered due to the high recurrence rate of DTF, however a proper treatment regimen, both surgical and adjuvant, have not yet been established. While radiotherapy is more commonly used for known positive margins and inoperative cases, anti-estrogen hormone therapy and cytotoxic chemotherapy have also been utilized, however, such experiences are limited and results have been inconclusive. Due to the limited accumulation of data on intrathoracic desmoid-type fibromatosis, they remain a challenge for both the patient and clinician and should remain under long term follow-up.

References:


