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Case Report

Mediastinal Desmoid Tumor Presents as Lymphadenopathy in Patient with Lymphoma: A Case Report

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Keywords
Fibromatosis · Aggressive · Lymphadenopathy · Radiation · Protein-tyrosine kinase · Lymphoma · Non-Hodgkin

Abstract
Desmoid tumors are rare mesenchymal neoplasms that are rapidly growing but do not metastasize. We present a case of a 75-year-old man with a history of non-Hodgkin lymphoma in remission incidentally found to have an enlarging internal mammary lymph node on screening CT, subsequently diagnosed as a desmoid tumor via biopsy. The patient was deemed unfit for surgical resection and instead underwent urgent radiation and immunotherapy. This report highlights a unique case of desmoid tumor presenting as mediastinal lymphadenopathy.

Introduction

Desmoid tumors, also known as aggressive fibromatoses, are rare mesenchymal neoplasms defined as a locally fibroblastic proliferative disease that are often rapidly growing and infiltrative [1]. They are unable to metastasize; however, they are a significant source of morbidity and mortality due to their rapidly growing nature and ability to impinge on vital anatomical structures.

Desmoid tumors are split into two main subtypes: sporadic and familial [2]. Sporadic desmoid tumors are the far more common etiology and are associated with somatic beta-catenin mutations. Familial cases are associated with germline APC mutations or familial adenomatous polyposis. Both categories of desmoid tumors typically present in patients aged 15–60 and are most common in young adults, with APC-associated desmoids presenting even earlier [3].

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While desmoid tumors can arise throughout the body, they are typically extra-abdominal. Extra-abdominal desmoid tumors are typically sporadic and most commonly occur in the limbs, the trunk, or head and neck [4]. This is in contrast to intra-abdominal desmoid tumors which are classically associated with familial adenomatous polyposis. Interestingly, 25% of cases of desmoid tumor are associated with prior history of trauma or surgery at that site [1]. Patients often present with concerns of a palpable mass, with or without tenderness. They more rarely may present with symptoms secondary to compression of adjacent anatomical structures (i.e., abdominal pain, hoarseness, dyspnea). This paper illustrates a case of a uniquely presenting asymptomatic, sporadic desmoid tumor in an elderly man. Written informed consent was obtained by the patient for publication of this case and associated images. The CARE Checklist has been completed by the authors for this case report, attached as online supplementary material (for all online suppl. material, see https://doi.org/10.1159/000532097).

Case Presentation

Our case involves a 75-year-old man with a history of low-grade follicular cell lymphoma who recently completed maintenance immunotherapy. He presented to the office for asymptomatic follow-up of treatment. Surveillance non-contrast CT of the chest, abdomen, and pelvis was ordered. Findings included the development of a 2.3 by 2.2 cm enlarged lymph node along the internal mammary lymph node chain (shown in Fig. 1). The decision was made to continue surveilling the node for growth.

It is important to note the patient's prior medical history. Initial presentation of non-Hodgkin lymphoma 2 years earlier was with a left chylothorax secondary to thoracic duct injury. Subsequent left submandibular lymph node excisional biopsy revealed follicular lymphoma grades 1–2. The patient underwent left chest tube placement, which drained 2.4 L of fluid.

The pleural effusion eventually reaccumulated, and the patient agreed to undergo video-assisted pleurodesis. The patient later underwent eight cycles of maintenance immunotherapy complicated by hospitalization for severe nausea and failure to thrive. At the time of presentation, he was asymptomatic and following up for routine surveillance of malignancy in remission.

Five months after initial surveillance imaging, repeat CT scan found dramatic growth of the presumed lymph node, now measuring 8.4 by 6.4 by 11.1 cm (shown in Fig. 2). CT-guided biopsy was done to assess for recurrence of follicular lymphoma or transformation. Pathology revealed a spindle cell neoplasm consistent with desmoid fibromatosis, with no lymphoid tissue present. Antibody staining was positive only for B-catenin. Repeat CT of the chest 3 months after last imaging showed enlargement of the left internal mammary lymph node, now measuring 11.4 × 8.8 × 14.7 cm (shown in Fig. 3, 4). The mass was noted to be extending into the anterior mediastinum. Its echotexture was described as somewhat heterogenous with regions of high density and a linear region of calcification.

Due to progression of the mass and extension into the mediastinum, cardiothoracic surgery was consulted for potential resection. The patient was deemed to be a poor surgical candidate due to age, significant comorbidities, and proximity of the mass to important structures. The patient was then referred to radiation oncology for a course of urgent radiation. An MRI prior to initiation of radiation revealed stable large soft tissue mass abutting the adjacent pleural surface with extension into the left side of the superior mediastinum and

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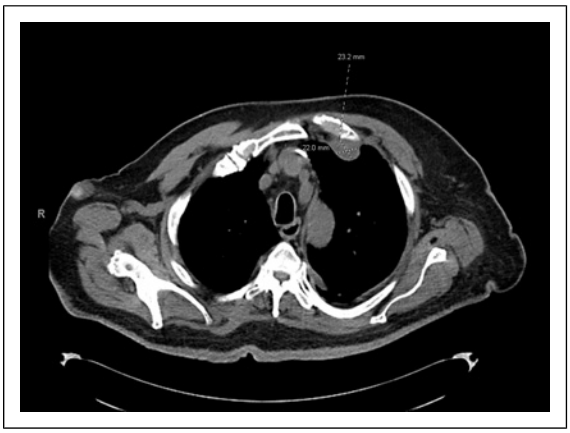

Fig. 1. Axial CT chest showing mediastinal mass measuring 22.0 by 23.2 mm, adjacent to mammary lymph nodes.

anterior left chest wall. At the time of publishing this case report, the patient is undergoing radiation therapy and is scheduled to begin tyrosine kinase inhibitor immunotherapy following completion of radiation.

Discussion

This case represents an unusual presentation of desmoid tumor appearing as lymphadenopathy on imaging. Desmoid tumors arise most commonly in young women, typically appearing in the third and fourth decades of life [2]. Santti et al. [5] found the expression of estrogen receptors in around 10% of desmoid tumors examined, correlating to the more frequent presentation in women of child-bearing age. More commonly, cyclin D1 receptors were present. Immunopositivity of each of these receptors was found to correlate with high proliferation in desmoid tumors. Our patient being an elderly man is already an uncommon circumstance of desmoids. However, his prior intrathoracic procedures predisposed him to developing desmoid tumors [1].

The clinical presentation of desmoid tumors widely varies and is determined by the rate of growth and location of tumor [6]. Tumors in the limbs are often symptomatic early, impacting neurovascular bundles. Conversely, intra-abdominal desmoids may not present until large in size, leading to intestinal obstruction or ischemia. Desmoid tumors can often mimic other malignancies, for example, breast masses [7]. However, we found no cases of desmoid tumors presenting as lymph-related malignancy.

Several images revealed the mass to be an enlarged lymph node, suspicious for recurrent lymphoproliferative disease. However, image-guided core needle biopsy accurately diagnosed it as a desmoid mass adjacent to the internal mammary lymph node chain rather than a lymph node itself. Core needle biopsy is required for formal diagnosis of desmoid tumors [2].

In contrast, fine needle biopsies can provide nonspecific, misleading results and are typically not diagnostic [8]. This finding highlights the necessity of biopsy for suspicious lesions in those with history of malignancy nonconcurrent with presentation. Likewise, this supports the use of MRI in suspicious masses. MRI is considered the gold standard imaging for diagnosing, monitoring, and assessing resectability of desmoid tumors [9]. In this case, MRI may have been useful in delineating boundaries of the mass and determining the true etiology of the mass, enlarged lymph node versus a soft tissue mass adjacent to lymph node chain.

This patient's case was confounded by his comorbidities. Primarily, both of his CT scans were done without contrast due to renal insufficiency. This provided limitation to visualization of the mass and may have contributed to mislabeling of the desmoid tumor

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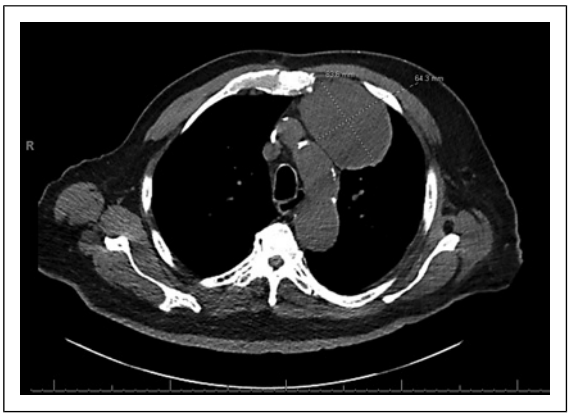

Fig. 2. Axial CT chest showing progression of mediastinal mass measuring 83.6 mm by 64.3 mm.

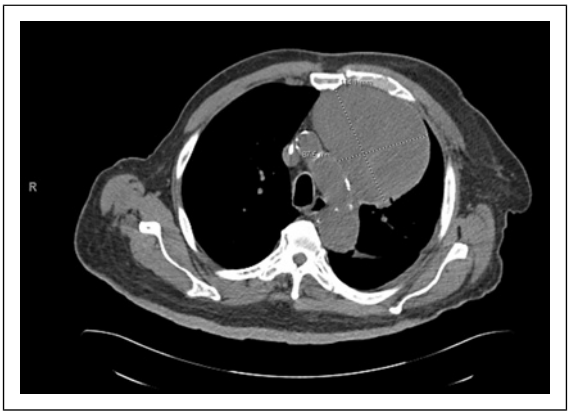

Fig. 3. Axial CT chest showing progression of mediastinal mass measuring 114.1 mm by 87.5 mm.

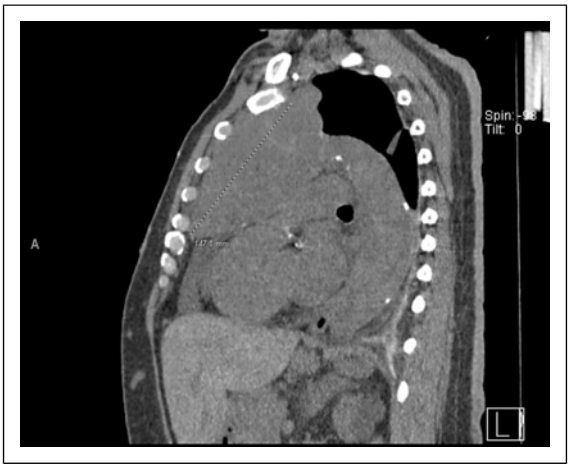

Fig. 4. Lateral CT chest showing anterior mediastinal mass measuring 147.1 mm in length.

as a lymph node. In patients that cannot receive contrast with CT imaging, there is greater indication for MRI to investigate suspicious masses. Additionally, this patient's history of non-Hodgkin lymphoma supported the clinical picture of asymptomatic lymphadenopathy due to recurrence of disease, further complicating his case and diagnosis. Malignancy and benign reactive processes like reactive lymphadenopathy had to be carefully ruled out in this case.

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Due to the rapidly growing and invasive nature of these neoplasms, historically standard treatment for desmoid tumors has been surgical resection with the goal of preserving involved structures or minimizing symptoms. However, rate of recurrence of these malignancies has been found to be over 20% [10]. Various other treatment methods have been studied. In 2019, a joint global consensus formed a guideline for desmoid management [11]. They established that the first step of management is surveillance. If the mass progresses over serial images, impinges on adjacent anatomical structures, or becomes overtly symptomatic, surgery is first-line treatment for abdominal wall masses. For all other locations of malignancy, medical management is preferred and consists of options like antihormonal therapies, NSAIDs, tyrosine kinase inhibitors, or low-dose chemotherapy. An exception is made for desmoids of the head, neck, or intrathoracic cavity, for which radiation therapy can be considered. Radiation has been shown to provide good local control of disease, especially in older adults [12].

Conclusion

This is an overall unique presentation of desmoid tumor. From our review, this is the only published case of a desmoid tumor presenting as an enlarged lymph node. This contributes to the body of evidence elucidating the difficulties in diagnosing desmoid tumors, supporting the use of MRI as diagnostic imaging method of choice and more scrutinous workup for suspicious abdominal or mediastinal lesions.

Statement of Ethics

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. No patient identifying information is included in this study. This research was conducted in accordance with the World Medical Association Declaration of Helsinki. This retrospective review of patient data did not require ethical approval in accordance with local guidelines.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

Tara L. Ballouz, BS, is the primary author of this report. Jeffrey H. Margolis, MD, is the principal investigator and was involved with data collection and editing of this report. All coauthors have seen and agree with the contents of the manuscript.

Data Availability Statement

All data generated or analyzed for this study are included in this report and its online supplementary material. Further inquiries can be directed to the corresponding author.

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