Desmoid Fibromatosis: not always benign in nature

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INTRODUCTION

- Desmoid tumors, also known as desmoid fibromatoses, are rare, locally aggressive, benign soft tissue sarcomas with an annual incidence of between 2-4 cases per million.
- The majority of cases are sporadic, often associated with somatic mutations in the beta catenin or APC genes. True intra-thoracic tumors are very rare.
- Although desmoids do not metastasize, they can cause significant morbidity and mortality due to local invasion as well as high recurrence rates.
- This case illustrates the use of chemotherapy in a patient with a large intra-thoracic desmoid tumor (see Figure).

LARGE THORACIC DESMOID TUMOR

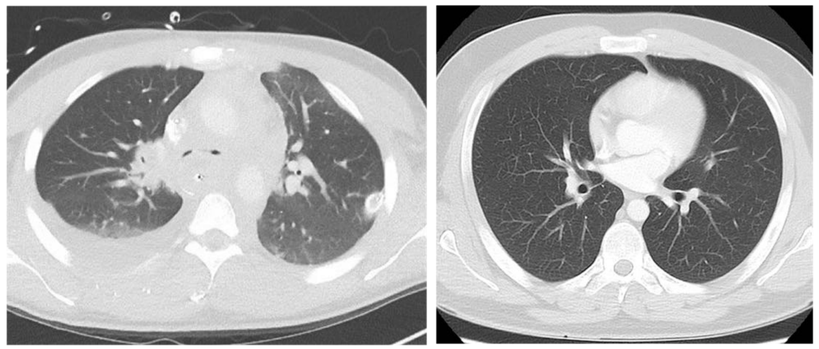
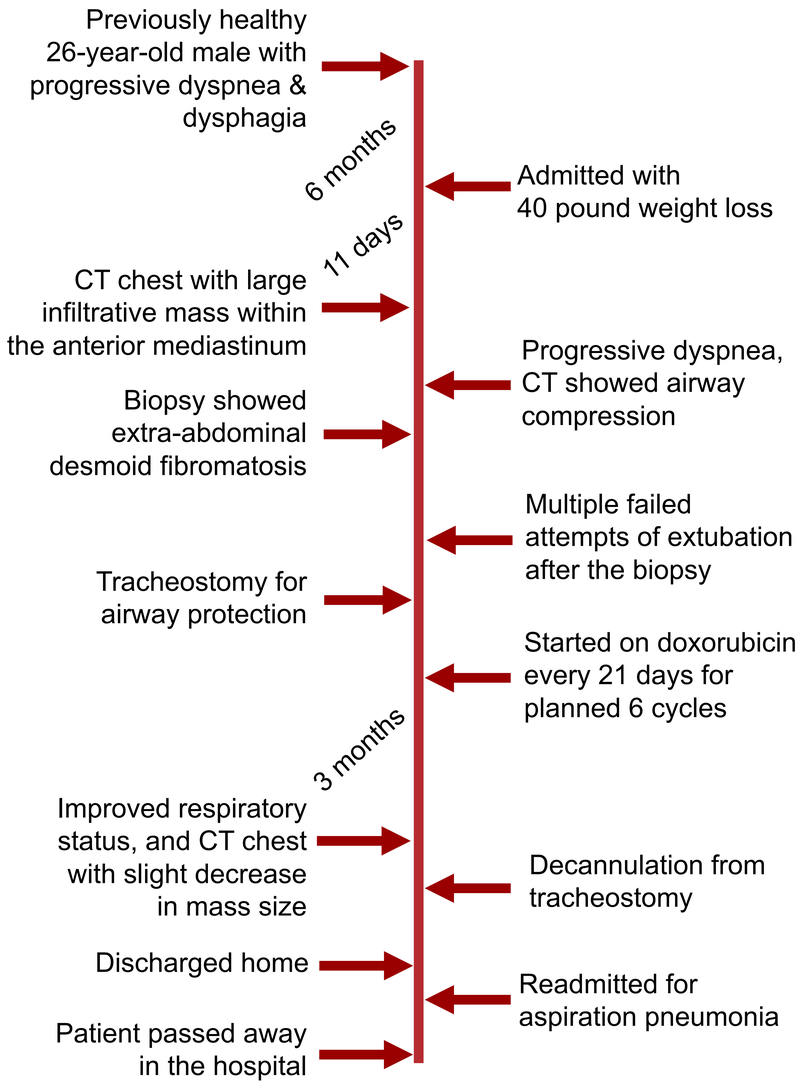


Figure: CT chest (left) revealed a large infiltrative mass within the anterior mediastinum, extending into the aortopulmonary window and encircling the aorta, pulmonary arteries, SVC and central pulmonary veins. Normal CT chest (right) for comparison.

CASE DESCRIPTION



DISCUSSION

- There is no standard first line treatment for desmoid tumors where progression is rapid. Options include:
  - radiation
  - systemic therapy
  - surgery
  - ablative treatments
- Our patient was not a candidate for surgical or ablative treatments due to the location and local involvement of mass.
- Radiation and systemic therapy have a long response time given the slow growing nature of the tumor.
- Radiation was not pursued as it has no clear advantage over systemic therapy and challenging as this patient was intubated.
- Systemic therapy was identified as the first line treatment. Given the urgency for faster response time, the following systemic therapies were considered:
  - TKI inhibitors (sorafenib, inmatinib, pazopanib)
  - Anthracycline
  - Low-dose methotrexate + vinblastine.
- Ultimately, doxorubicin was chosen given retrospective data indicating favorable outcomes for unresectable desmoid tumors, although the literature lacks RCTs on this topic due to the rarity of this disease.
- In this patient, doxorubicin achieved slight clinical improvement following three 21-day cycles as evidenced by improvement in respiratory status, with minimal mass shrinkage on CT imaging.
- This case highlights that while desmoid tumors are considered benign, they may be approached as malignant soft-tissue neoplasms that warrant aggressive therapy.