Mitral regurgitation due to a mediastinal desmoid tumor: A case report and review of the literature

Baraa Shebli1 | Ayham Alzahran1 | Ali Mansour1 | Sami Kabaweh1 | Sarab Agha2 | Bayan Shareef3 | Manar Abdullah3

1Faculty of Medicine, University of Aleppo, Aleppo, Syria
2Department of Pathology, Aleppo University Hospital, Aleppo, Syria
3Pediatric Department, Aleppo University Hospital, Aleppo, Syria

Correspondence
Ayham Alzahran, Faculty of Medicine, University of Aleppo, Aleppo, Syria.
Email: ayhamzh3@gmail.com

Abstract
Mediastinal tumors may cause a wide range of symptoms; therefore, it should be considered in the differential diagnosis, after excluding common causes, when dealing with pulmonary and cardiac symptoms such as dyspnea or even mitral regurgitation. The most common site of the desmoid tumors is the abdomen, but in very rare cases, it can be located in the mediastinum.

KEYWORDS
aggressive fibromatosis, Desmoid, mitral regurgitation

1 | INTRODUCTION

Desmoid tumors are rare benign tumors that arise most commonly in the abdomen, but can occasionally appear in extra-abdominal sites. We present a 3-year-old girl with mitral regurgitation in whom the final diagnosis was a mediastinal desmoid tumor. We managed the patient with complete surgical resection with an uneventful follow-up.

Desmoid tumors are locally aggressive and often recur even after complete resection, but they do not metastasize.2

Desmoid tumors most commonly arise in the abdomen, but can occasionally appear in extra-abdominal sites such as chest wall, shoulder, mediastinum, and other rare sites.3

We report a case of a 3-year-old girl with a mediastinal desmoid tumor and a thorough review of the literature to summarize the characteristics of mediastinal desmoid tumors.

2 | CASE PRESENTATION

A 3-year-old girl presented to the hospital with a 4-month history of dry cough and dyspnea. Her past medical and surgical history was unremarkable.

On physical examination, there was mild-to-moderate dyspnea without cyanosis or pallor. Chest examination showed diminished breath sounds on the right side with a respiratory rate of 44/min and an oxygen saturation of 95%. Heart rate was 136/min, blood pressure was 100/70 mm Hg, and there was a pansystolic murmur heard in the mitral area graded 3/6.

The abdomen was tender, and the liver was displaced inferiorly about 6 cm below the costal margin without splenomegaly or venous dilations on the abdominal wall.

Laboratory tests revealed abnormal levels of CRP (8 mg/L), ESR (31 mm/h), K+ (3.4 mmol/L), and urea (32 mg/dL). Other laboratory tests, including tumor markers, were unremarkable.

Chest X-ray (CXR) revealed a homogeneously increased density over the entire right side of the chest, shifting the mediastinum to the left, as seen in Figure 1.
Chest multislice computed tomography (MSCT) showed a soft-tissue mass filling the entire right side of the chest and shifting the mediastinum to the left, supplied by large blood vessels, without any invasion to the significant adjacent structures, as seen in Figure 2.

Cardiac ultrasound revealed significant mitral regurgitation with normal electrocardiography.

We did an ultrasound-guided biopsy, and histological diagnosis was a desmoid tumor.

We performed thoracotomy through a right-sided incision in the 5th intercostal area. There was a large mass extending from the posterior mediastinum and pressing the right lung. Therefore, we dissected and isolated the mass from the chest wall and adjacent structures. After that, the right lung expanded well, as seen in Figure 3.

We did a postsurgical assessment for the patient. All vital signs were normal, the liver went back to its normal location, and the mitral regurgitation resolved, meaning that the tumor was causing functional regurgitation due to the large mass effect.

We discharged the patient with good general condition and monthly follow-up.

3 | REVIEW OF THE LITERATURE

Desmoid tumors (also known as aggressive (deep) fibromatosis) are rare benign soft-tissue tumors that consist of proliferated, well-differentiated fibroblasts. They represent only 0.03% of all neoplasms, and the annual incidence is approximately 1-4 per million population. Most cases occur between the ages of 15 and 60 years old and more common in women than in men.

The exact etiology of desmoid tumors is not clear, but scientists suggest several theories, such as genetic factors, surgical scarring, trauma, pregnancy, and estrogenic hormones.

Desmoid tumors are locally aggressive and have a high rate of recurrence despite resection, but they do not metastasize. Desmoid tumors originate most commonly in the abdomen, but can occasionally involve extra-abdominal sites such as chest wall, shoulder, mediastinum, and other rare sites.

To diagnose desmoid tumors, CT is useful to determine the location of the tumor and its extensions to the surrounding structures, but MRI remains the main imaging test and can be used for diagnosis, local staging, and follow-up.
| Author     | Year | Age (y) | Gender | Location            | Pathogenesis   | Clinical Manifestations                               | First Management                                      | Recurrence Management               | Follow-Up                      |
|------------|------|---------|--------|---------------------|----------------|------------------------------------------------------|-------------------------------------------------------|-------------------------------------|-----------------------------------|
| Eugene     | NA   | NA      | NA     | Posterior mediastinum | NA             | NA                                                   | incomplete surgery                                   | NA                                  | Two recurrences/3 mo              |
| Eugene     | NA   | NA      | NA     | Posterior mediastinum | NA             | NA                                                   | Complete surgery                                     | -                                   | Free/ 7 y                        |
| Eugene     | NA   | NA      | NA     | Posterior mediastinum | NA             | NA                                                   | Complete surgery                                     | -                                   | Free/ 17 y                       |
| WADDELL    | 1980 | 38      | Male   | Anterior mediastinum | De novo        | -                                                   | Incomplete surgery                                   | chemotherapy then radiotherapy then Indomethacin | Recurrence after 6 mo then free after 3.5 y |
| Tsukiyama  | 1984 | 17      | Female | NA      | NA             | NA                                                   | external irradiation and interstitial irradiation    | NA                                  | NA                               |
| Krause     | 1985 | 2.5     | Male   | Posterior mediastinum | Postsurgery    | respiratory distress and deglutition problems       | Incomplete surgery                                   | -                                   | Free/ 2 mo                       |
| Kaplan     | 1986 | 18      | Male   | Posterior mediastinum | Postsurgery    | dysphagia and regurgitation                          | Incomplete surgery                                   | adjuvant radiotherapy and indomethacin | Recurrence after 4 mo             |
| Markhede   | 1986 | NA      | NA     | NA      | NA             | Suffocation                                         | NA                                                   | NA                                  | Death/ NA                        |
| Black      | 1987 | 45      | Male   | Posterior mediastinum | De novo        | Back pain                                           | Complete surgery                                     | NA                                  | NA                               |
| Easter     | 1989 | 5       | Male   | NA      | NA             | NA                                                   | Incomplete surgery and chemotherapy                  | -                                   | Alive with disease for 12 mo      |
| NEINSTEIN  | 1990 | 18      | Female | Anterior mediastinum | De novo        | Chest pain, wheezing and cough                       | Complete surgery (first recurrence), complete surgery with adjuvant hormonal therapy (second recurrence), complete surgery (third recurrence) | 3 Recurrences then Free for 1 year |
| Casillas   | 1991 | 19      | Male   | Anterior mediastinum | De novo        | Mass on the left side of the neck                    | NA                                                   | NA                                  | Death/ NA                        |
| Winer-Muram| 1994 | 14      | Female | Posterior mediastinum | De novo        | Pain, dyspnea, and cough                             | Complete surgery                                     | NA                                  | NA                               |
| Tam        | 1994 | 35      | Male   | Anterior mediastinum | De novo        | Asymptomatic                                        | Incomplete surgery                                   | NA                                  | NA                               |
| Okamura    | 1995 | 12      | Female | Anterior mediastinum | De novo        | Thoracic scoliosis and left upper limb weakness     | Complete surgery                                     | NA                                  | NA                               |

(Continues)
| Author            | Year | Age (y) | Gender  | Location         | Pathogenesis | Clinical Manifestations                                                                 | First Management                  | Recurrence Management                      | Follow-Up       |
|-------------------|------|---------|---------|------------------|--------------|----------------------------------------------------------------------------------------|-----------------------------------|--------------------------------------------|-----------------|
| Plukker           | 1995 | 13      | NA      | Anterior mediastinum | Post-surgery | NA                                                                                     | Complete surgery                  | NA                                         | NA              |
| Sheung-Fat Ko     | 1996 | 3       | female  | Posterior mediastinum | De novo      | a nontender mass on the posterior aspect of the left lower thorax                        | Complete surgery                  | NA                                         | NA              |
| Sabaté            | 1996 | 70      | male    | Middle mediastinum   | De novo      | NA                                                                                     | Incomplete surgery and external radiotherapy | Chemotherapy and hormonal therapy (first recurrence) and NA ( second recurrence) | 2 recurrences after (4 - 40) mo |
| Dosios            | 1998 | 21      | Female  | Posterior mediastinum | De novo      | Pain in left shoulder and upper arm                                                    | complete surgery                  | complete surgery (first recurrence), complete surgery with adjuvant hormonal and radiotherapy (second recurrence) | Two recurrences after (9 mo - 12 mo) then Free for 20 mo |
| Çiftçii           | 1998 | 40      | Male    | Posterior mediastinum | De novo      | non-productive cough and lower back pain                                                | Radiotherapy                      | NA                                         | NA              |
| Inase             | 1999 | 67      | female  | Middle mediastinum   | De novo      | SVC syndrome, Horner's syndrome, paralysis of bilateral vocal cords and diaphragm and HF | NA                               | NA                                         | Death/ 3 mo     |
| Wilson            | 1999 | 49      | male    | Posterior mediastinum | NA           | Chest pain and cough                                                                     | Complete surgery                  | -                                          | Free for 96 mo  |
| Kawashima         | 2000 | 12      | female  | Posterior mediastinum | De novo      | Neuropathy of lower limbs                                                               | Complete surgery                  | NA                                         | NA              |
| Hoeffel           | 2000 | 22      | female  | Posterior mediastinum | De novo      | painless dysphagia for solids and weight loss                                           | NA                               | -                                          | Death after 24 mo |
| Hoeffel           | 2000 | 61      | male    | Posterior mediastinum | De novo      | dysphagia                                                                               | NA                               | -                                          | Death after 26 mo |
| Cardoso           | 2002 | 35      | Male    | Superior mediastinum  | De novo      | Chest pain, right supraclavicular mass, and neck vein dilation                          | Incomplete surgery with adjuvant radiotherapy | -                                          | Free/ 6 y       |
| Yoko Torii        | 2005 | 36      | Female  | Posterior mediastinum | De novo      | Back pain                                                                               | Complete surgery                  | -                                          | Free/ 10 mo     |
| Borzellino        | 2006 | 48      | Male    | Anterior mediastinum  | Post-Surgery | Chest pain                                                                               | Complete Surgery                  | -                                          | Free/ 3 y       |
| Author         | Year | Age (y) | Gender | Location         | Pathogenesis | Clinical Manifestations                  | First Management                                                                 | Recurrence Management | Follow-Up               |
|----------------|------|---------|--------|------------------|--------------|------------------------------------------|----------------------------------------------------------------------------------|-----------------------|-------------------------|
| Nakagiri       | 2007 | 45      | Male   | Posterior mediastinum | De novo      | Dysphagia                                | Incomplete surgery with adjuvant radiotherapy and hormonal therapy                | -                     | Death after 6 mo        |
| Wilhelm        | 2007 | 67      | Female | Anterior mediastinum | Post-Surgery  | dyspnea on exertion                      | Incomplete surgery                                                            | NA                    | NA                      |
| Ayadi-Kaddour  | 2008 | 61      | Male   | Posterior mediastinum | De novo      | Thoracic and right scapular pain         | Complete surgery                                                               | -                     | Free/ 2 y               |
| Serraj         | 2011 | 44      | Female | Middle mediastinum   | De novo      | Chest pain and hemoptysis                | Corticosteroids then Radiotherapy                                                | -                     | Death during radiotherapy treatment |
| Fujinaga       | 2012 | 17      | Male   | NA               | NA           | NA                                       | Complete surgery                                                                | Complete surgery       | Recurrence after 2 y    |
| Bouchikh       | 2013 | 49      | Female | Posterior mediastinum | De novo      | Chest pain, orthopnea and weight loss    | Complete surgery                                                               | -                     | Free/ 6 mo              |
| Yuxin Xie      | 2014 | 50      | Female | Superior mediastinum | De novo      | a dull pain in the left scapular region and a decreased range of motion of the left upper limb | Complete surgery                                                                 | adjuvant radiotherapy | Recurrence after 12 mo  |
| Hyung Lee      | 2015 | 71      | Male   | Anterior mediastinum | Post-Surgery  | Chest pain, dyspnea, and productive cough | Complete surgery                                                               | -                     | Free/ NA                |
| Wojtyś         | 2015 | 26      | Male   | Posterior mediastinum | De novo      | dyspnea, dysphagia, and odynophagia      | Incomplete surgery with adjuvant radiotherapy                                 | NA                    | NA                      |
| Bhat           | 2015 | 3       | female | Anterior mediastinum | De novo      | strider and exertional dyspnea           | Incomplete surgery                                                             | NA                    | Recurrence after 5 mo   |
| Talha Mahmoud  | 2017 | 49      | male   | Anterior mediastinum | Post-Surgery  | a heaviness in the chest                  | Complete surgery                                                               | -                     | Free for 9 mo           |
| McAninch       | 2019 | 6       | male   | Anterior mediastinum | NA           | Left neck mass and acute hypoxemic respiratory failure | NA                                                                               | NA                    | NA                      |
| Our case       | 2019 | 3       | female | Posterior mediastinum | De novo      | Dry cough, dyspnea, mitral regurgitation, and hepatomegaly | Complete Surgery                                                              | -                     | Free/ 3 mo              |

Abbreviation: NA, not available.
The definitive diagnosis of desmoid tumors is only made by biopsy. Core biopsies are preferable, while neither incisional nor excisional biopsy is recommended as the initial diagnostic modality.

The literature reported several treatment options for desmoid tumor, but there is still limited evidence about their efficacy and the best management in mediastinal desmoid tumors since very few cases reported recurrence with a convenient follow-up period. Moreover, there is a lack of evidence about the best way to follow up patients with treated mediastinal desmoid tumors.

We conducted a thorough review of the literature to summarize the characteristics of mediastinal desmoid tumors and to explore the reported treatment choices and their follow-up.

We searched the literature databases for any case report that involved a mediastinal desmoid tumor using Medical Subject Headings (MeSH).

The inclusion criteria were case reports of tumors that originated from the mediastinum, and we did not have any restrictions on language or age.

We found 40 case reports to include in the review (except our case); then, we extracted information about age, gender, pathogenesis, clinical manifestations, management, and follow-up, which are summarized in the table below. (Table 1).

The table shows that the reported mean age for diagnosing mediastinal desmoid tumor is 32.2 years, ranging from 2.5 to 71 years. This indicates that mediastinal desmoid tumors are rare in small ages (only 6 patients were under 10 years).

There was an evident mild tendency to occur in males in the reported cases 55.5% (20 out of 36).

The most reported location in the mediastinum was the posterior mediastinum (54%) followed by the anterior mediastinum (32%), with only 3 cases in the middle mediastinum and 2 cases in the superior mediastinum.

Most of the reported tumors were de novo (78.1%), while the other cases developed after surgical procedures.

All the cases except one presented when the tumor became large in size, and most of the symptoms had the same mechanism (mass effect of the tumor) and were related to the location of the tumor.

According to the reported cases, pain (most commonly located in the chest) was the most common symptom at presentation, followed by dyspnea.

In general, respiratory symptoms were the most common; gastrointestinal symptoms were also commonly reported, especially dysphagia.

Specific cardiovascular symptoms were rare (1 with superior vena cava (SVC) syndrome, 1 heart failure symptoms, and 1 with mitral regurgitation).

For treatment, there was no clear evidence for the best method.

Several treatment options have been reported in the literature (complete surgery, incomplete surgery, chemotherapy, and radiotherapy) (Table 1).

The most commonly reported method was complete surgery (20 out of 35).

This method proved efficacy in 10 patients who remained clear during the follow-up period.

Four patients (who underwent complete surgery) had recurrences, and the other 6 patients did not report any follow-up.

Recurrence management and follow-up periods varied among studies without any evidence of the best method and period.

As seen in the review, evidence about the best treatment options, recurrence management, and follow-up methods and period remains unclear. Therefore, further studies are needed.

4  |  CONCLUSION

Mediastinum is a very rare location for desmoid tumors, and only a few cases have been reported in the literature.

We reviewed the literature on mediastinal desmoid tumors.

The mean age to diagnose mediastinal desmoid tumor was 32 years old.

Several clinical presentations have been reported, and they were mostly respiratory symptoms and related to the location of the tumor, but rare specific presentations were reported (mitral regurgitation), and such symptoms should not be overlooked.

There were several treatment options and follow-up periods and methods.

There was a lack of evidence about their management and follow-up, so more studies need to be conducted to provide strong evidence about their best management and follow-up.

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CONFLICT OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

BS and AA: conceived the content, reviewed the literature, drafted the manuscript, and critically revised the final manuscript. AM, SK, SA, BS, and MA: contributed in drafting the manuscript. MA: gave the final approval.
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CONSENT FOR PUBLICATION
Informed consent was signed by the patient.

ORCID
Ayham Alzahran https://orcid.org/0000-0002-8632-7068

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