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Large Desmoid Fibromatosis of the Breast Invading the Chest Wall

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Background	A desmoid fibromatosis is a locally invasive sarcoma subtype most commonly seen in females. Most of these lesions arise without risk factors, but some are associated with familial syndromes, hormonal imbalances, and surgical trauma. They do not frequently metastasize, but they often invade local structures and may cause significant morbidity and mortality. Due to high recurrence rates, iatrogenic injuries, and the potential for spontaneous resolution, management has shifted from surgery to active observation in most circumstances.
Summary	An 18-year-old female presented with a rapidly growing palpable medial left breast mass, representing a large desmoid fibromatosis posterior to the left medial breast parenchyma with invasion into the chest. An incisional biopsy displayed low-grade spindle cells favoring fibroma. Subsequent computed tomography (CT) and magnetic resonance imaging (MRI) showed that the tumor invaded the chest wall musculature between the medial third and sixth ribs. The patient underwent a left skin-sparing mastectomy with en bloc resection of the chest wall and fourth and fifth ribs. Methylmethacrylate mesh was used to reconstruct the chest wall, and acellular dermal matrix reinforced the closure. The breast defect was closed primarily with plans for delayed implant-based reconstruction.
Conclusion	Desmoid fibromatoses are locally invasive tumors with minimal potential for metastasis. While surgery has previously been the standard of care, recent management of desmoid fibromatoses has shifted to non-surgical options in most circumstances. We present a case that highlights the contrary, where a large and aggressive desmoid fibromatosis invaded the chest wall and ribs, making surgery necessary.
Key Words	desmoid fibromatosis; skin-sparing mastectomy; chest wall reconstruction

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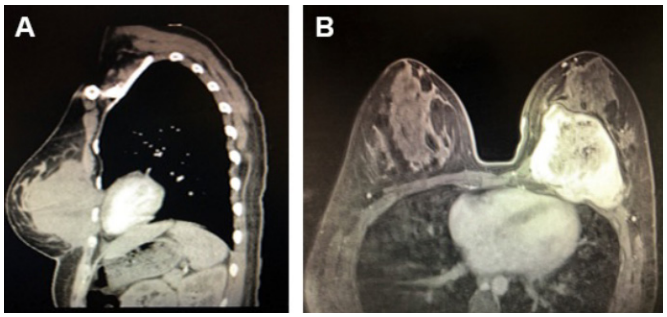
Case Description

An 18-year-old female initially presented to a local rural facility with a rapidly growing palpable medial left breast mass. Initial imaging of the mass was inconclusive, so she was referred to a higher level of care facility. Under the presumption of a fibroadenoma, the patient was taken to the operating room for complete surgical resection through an inframammary fold incision. However, it was noted that the lesion was far more extensive than suspected, and an incision biopsy was done instead. The pathology was inconclusive, but concern was raised for a low-grade spindle cell lesion. She was then referred to a tertiary/quandary care facility.

Further imaging work up at that time included magnetic resonance imaging (MRI) identifying a 10 cm × 7 cm × 9 cm solid mass arising from or adjacent to the medial chest wall, posterior to the breast parenchyma, with invasion through the pectoralis major and insinuation into the chest wall intercostal muscles between the third through sixth ribs at the costochondral junction. The mass displaced the normal fibroglandular and fatty breast tissues anteriorly (Figure 1). Comprehensive genetic counseling and testing were done and were negative for pathogenic mutation.

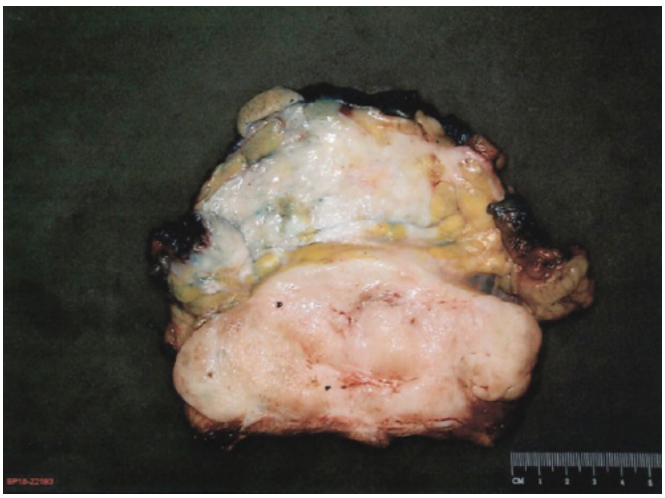
Figure 1. Solid Mass Arising From or Adjacent to the Medial Chest Wall. Published with Permission.



A) Sagittal CT scan showing 10 cm × 7 cm × 9 cm mass in left breast invading chest wall; B) Transverse MRI scan showing 10 cm × 7 cm × 9 cm mass in left breast invading chest wall

Due to the rapid growth and distortion of the left breast, the appearance of invasion into the chest wall on imaging, and the continued uncertainty of the actual pathology, surgical intervention was recommended. A left skin-sparing mastectomy (resection of the nipple-areolar complex only) with en bloc resection of the pectoralis major and minor muscles, part of the serratus anterior and fourth and fifth ribs was done in combination between breast surgery, plastic surgery, and thoracic surgery. The 15 cm × 14 cm × 12 cm surgical specimen (Figure 2) had negative resection margins and showed no evidence of bony or pleural involvement. The resultant anterior chest wall deformity was reconstructed with a piece of methylmethacrylate mesh. A 15 cm × 15 cm piece of acellular dermal matrix (AlloDerm®) reinforced the chest wall. The breast defect was closed primarily with plans for a delayed implant-based reconstruction after pathology was finalized. Final pathological diagnosis confirmed 13 cm × 13 cm × 6.7 cm desmoid fibromatosis without cytologic atypia or nuclear pleomorphism. The cells were positive for p16, BCL-2 (weak), beta-catenin (extensive nuclear), smooth muscle actin (weak), and vimentin. The cells were negative for desmin, AE1/3, CD34, and S100. Final surgical margins were negative, but it was noted there was pectoralis muscle involvement. She has recovered without complication or sign of recurrence at one year.

Figure 2. Desmoid Fibromatosis Excised from the Left Chest Wall. Published with Permission



Discussion

Extraabdominal desmoid fibromatoses are soft tissue tumors arising from musculoaponeurotic tissue with an incidence of 2.4–4.3 per million per year.[1–3] They are the clonal proliferation of cells with variable rates of progression and the ability to invade surrounding structures but not metastasize.[4–7] They exhibit no nuclear nor cytoplasmic features of malignancy.[5,7] Management of these tumors is case-specific, and treatment options include observation, hormonal therapy/nonsteroidal anti-inflammatory drugs

(NSAIDs), tyrosine kinase inhibitors (TKIs), chemotherapy, radiation, and surgical excision.[4,8] The presence of estrogen receptors on desmoid tumors and the suspected involvement of estrogen in the tumor physiology has facilitated the use of anti-estrogens, such as tamoxifen, for treatment.[13] Recent studies have shown the use of TKIs, such as sorafenib, to significantly increase disease-free survival time in patients with refractory or symptomatic desmoid tumors.[12] Historically, the mainstay of treatment was surgical excision. Still, due to high recurrence rates, iatrogenic injuries, and the possibility of spontaneous regression, active observation is recommended in most circumstances wherein significant morbidity and mortality are unlikely.[4] Management depends on location, functional impairment, invasiveness, and the potential for continued growth and local deformity. Surgery is indicated in specific circumstances.[4,7]

Desmoid fibromatosis is a locally invasive sarcoma subtype that accounts for 3 percent of soft tissue tumors and is most commonly seen in young females.[1,2,7,8] Predisposing factors include aberrations in the Wnt signaling pathway, including familial syndromes such as familial adenomatous polyposis (FAP) syndrome, specifically the Gardner syndrome subtype, hormonal exposure/pregnancy, and surgical trauma. Still, a majority of these lesions arise spontaneously.[4,6,12,13] The most common extra-abdominal sites are the limb-girdle, followed by the trunk, head, and neck.[1,4] Symptoms include pain, swelling, structural deformity, and loss of function. These tumors are known to be locally aggressive, invading surrounding structures and vital organs, causing significant morbidity in these instances.[12] However, these tumors also can have a high rate of cessation of growth and even have the potential to regress spontaneously.[4,9] Nonetheless, they can grow to massive sizes as reported by Nakayama et al. Most patients in their retrospective review had lesions that stopped growing or regressed, but one patient had a tumor that grew to 30 cm, causing significant functional impairment.[4,9]

Determining which extra-abdominal desmoid fibromatoses will grow, regress, stabilize, and/or recur can be a challenge.[4,9] Larger tumors, those located on the extremities and trunk, and those found in younger patients have been associated with a higher risk of progression.[4,10] Molecular markers predicting a lesion's course are being investigated.[4] For example, trisomy 8 and deregulation of beta-catenin are associated with recurrence after surgical excision and aggressive fibromatosis, respectively.[4,11]

Current recommendations include an initial period of observation in most cases due to high recurrence rates after surgery and high rates of arrested growth and spontaneous regression without surgery.[4,8] However, surgical intervention is warranted in cases of failed observation, local anatomy distortion and/or vital structure invasion, and instances wherein the diagnosis is in question.[1,4] Interestingly, negative resection margins do not guarantee that the tumor will not recur, and positive margins do not always lead to reappearance of the tumor.[4,5] Therefore, negative resection margins are only encouraged if they do not lead to loss of function.[4,5] Our patient highlights the current role of surgical management for extra-abdominal desmoid fibromatoses because the tumor was rapidly progressing and threatening invasion of the thoracic cavity as it grew to over 10 cm.

Conclusion

Desmoid fibromatoses are locally aggressive tumors that can lead to morbidity and mortality by destroying vital structures.[1,4,7] We present a case of an invasive desmoid fibromatosis involving the left medial chest wall with significant contour deformity and pain of the left breast and causing paresthesias of the left arm. It underscores the role of surgical management of these tumors when local structures are being invaded and when further growth will lead to morbidity.

Lessons Learned

Desmoid fibromatoses are benign tumors that can spontaneously regress and are most commonly treated with a period of active observation. However, surgeons must be aware of the signs for surgical intervention, including failed observation, local anatomic distortion, invasion of vital anatomic structures, and instances wherein the diagnosis is in question.

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