CASE REPORT
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Desmoid tumor in the male breast refractory to systemic treatment: case report

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ABSTRACT

Desmoid-type fibromatosis, an intermediate soft tissue tumor known for its local infiltrative behavior and high recurrence rate, is uncommon in the male breast. The report outlines the clinical journey of a 44-year-old male presenting with a rapidly growing breast mass, initially managed with a watchful waiting approach and subsequent systemic therapies including celecoxib, sorafenib, and a combination of methotrexate and vinblastine, all of which proved ineffective. Genetic testing revealed a catenin beta 1 (CTNNB1) mutation, indicative of a sporadic tumor. Despite various treatment attempts, significant tumor progression was observed, necessitating surgical intervention followed by radiotherapy. This case underscores the challenges in managing desmoid tumors, emphasizing the importance of multidisciplinary approaches and the potential need for surgical resection when conventional therapies fail.

KEYWORDS: breast; male; desmoid fibromatosis; diagnostic imaging; MRI.

INTRODUCTION

Desmoid-type fibromatosis is classified as an intermediate soft tissue tumor characterized by clonal fibroblastic proliferation originating from deep soft tissues[1]. This tumor is known for its local infiltrative behavior and high recurrence rate, though it rarely metastasizes[2]. These factors have led to the exploration of alternative treatment modalities beyond surgery, including radiation therapy, chemotherapy, and anti-hormonal therapies. Currently, surgical resection remains the gold standard for desmoid tumor management[3,4]. Desmoid fibromatosis is exceedingly uncommon in the breast, accounting for only 0.2% of all breast tumors, and this condition's incidence in males is sporadic[5].

CASE REPORT

A 44-year-old male patient complained of weight loss and a lump in the left breast. Percutaneous biopsy diagnosed a desmoid tumor, estrogen receptor (ER)-negative. Breast magnetic resonance imaging (MRI) demonstrated an oval mass with irregular margins, which showed heterogeneous enhancement, with a type I (persistent) kinetic curve in the left breast's lower quadrants, measuring 67 x 48 x 36 mm (estimated volume of 60 cc). It was in close contact with the skin and showed signs of infiltration into the pectoralis major muscle (Figure 1A-B). After discussion in the multidisciplinary tumor board, a watchful waiting approach was chosen with imaging control for three months after continuous use of celecoxib 200 mg/day. Upon return after this period, a significant increase in the lesion was noted, which was consistent with the results of the breast MRI performed in the same month, with the tumor volume estimated at 321 cc. A catenin beta 1 (CTNNB1) positive variant was detected at somatic genetic testing, with a sporadic origin of the desmoid tumor, without family risks. The case was meticulously reviewed during a multidisciplinary meeting, aiming at a tumor-reducing approach through targeted therapy with tyrosine kinase inhibitors. In this context, celecoxib was discontinued, and sorafenib was prescribed at an initial dose of 400 mg/day. However, due to the manifestation of cutaneous toxicity, the dose of sorafenib was adjusted to 200 mg/day. After four months, new imaging tests revealed a dimensional increase in the lesion, with the tumor volume estimated at 631 cc (Figure 1C). Given this scenario, the case was submitted to a SELNET (Sarcoma European & Latin-America Network) meeting for evaluation, resulting in a consensus to maintain systemic therapy. Methotrexate was then prescribed at a dose of 30 mg/m^2 and vinblastine at a dose of 5 mg/m^2,

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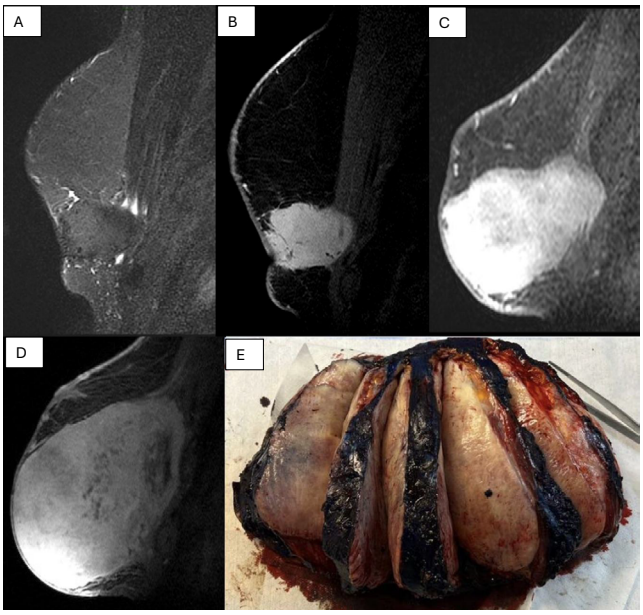

Figure 1. Contrast-enhanced sagittal T2-weighted sequence at breast MRI demonstrating an oval mass with irregular margins, presenting low signal and peri tumoral edema on T2-weighted images (A), infiltrating the pectoral muscle, and showing heterogeneous enhancement in sagittal fat-suppressed contrast--enhanced T1-weighted images (B). The lesion progressed despite the systemic treatment after seven months (C) and eleven months (D). The patient underwent a mastectomy with a partial resection of the underlying musculature (E).

administered weekly with a continuation plan for six months. After two months, another imaging examination was conducted, revealing further progression of the disease, with the tumor volume estimated at 1,146 cc (Figure 1D). Given this scenario, the decision was made to proceed with a surgical approach followed by radiotherapy. At that time, the patient experienced sporadic pain in the lower region of the tumor mass but denied dyspnea, chest pain, and other relevant symptoms. The tumor resection surgery, using an abdominal fasciomyocutaneous flap, was successful (Figure 1E). The patient remains asymptomatic at a seven-month follow-up, undergoing clinical and imaging surveillance with no signs of recurrence.

DISCUSSION

Desmoid neoplasia is characterized by its lack of metastatic potential, displaying a notable propensity for local growth, invasion of adjacent structures, and high recurrence rates[1]. The etiopathogenesis of desmoid tumors remains unknown. Although there have been reports of breast desmoid tumors following surgical procedures with breast implants and traumas, it is clear that isolated physical factors are not sufficient to trigger fibromatosis in the breast[6]. Although most cases have been observed in postmenopausal women, the issue of hormonal receptors in mammary fibromatosis remains controversial[2].

Breast imaging tests do not provide specific features to detect desmoid fibromatosis, which makes it difficult to distinguish this condition from breast malignancies[7]. However, imaging and histopathological examinations are essential for treatment planning and follow-up. Desmoid tumors often appear in mammography as high-density, irregular, non-calcified masses with spiculated margins. Chest computed tomography (CT) and MRI are valuable tools to determine the extent of the tumor's infiltration into adjacent tissues[8].

Managing desmoid tumors depends on various factors, including location, size, extent of invasion, and individual characteristics. Surgery has traditionally been the gold standard of treatment for desmoid tumors, aiming at complete resection with negative margins[3]. However, the high local recurrence rate post-surgery necessitates careful patient screening. A conservative "watch and wait" approach may be adopted in asymptomatic patients, with active surveillance employed to monitor for signs of progression, leveraging the tumor's potential for spontaneous stabilization or regression[5].

In some cases, neoadjuvant therapy may be indicated. In a study by Kaspar et al., desmoid tumors treated with imatinib demonstrated promising results. At six months, the progression arrest rate was 65%; subsequent rates were 59%, 53%, and 45% at 12, 15, and 24 months, respectively[9]. A retrospective study investigating the efficacy of apatinib, a tyrosine kinase inhibitor, revealed an objective response rate of 45.5%, with a notable disease control rate of 95.4%[10]. An additional study examined the use of tamoxifen and sulindac in treating desmoid tumors in adults. The results indicated an overall response rate of 60%, with complete response observed in 8% of cases and partial response in 52%[11].

Various chemotherapy drugs may be administered, including tyrosine kinase inhibitors (imatinib, sunitinib, and sorafenib), methotrexate and vinblastine (MTX/VBL), doxorubicin, and other agents like cyclophosphamide and pazopanib. Hormonal treatments, such as selective estrogen receptor modulators (tamoxifen, raloxifene, toremifene) and luteinizing hormone-releasing hormone analogs (leuprolide), can also be indicated in ER-positive tumors. Non-steroidal anti-inflammatory drugs like sulindac and celecoxib may also be prescribed. Radiation therapy may be used to target and shrink the tumor, particularly in cases where surgery is not feasible, or to reduce the risk of recurrence after surgery[12].

In the presented case, the patient experienced significant tumor progression over nine months, from 60 to 1,146 cc (approximately 20 times larger), despite multiple systemic treatments performed. Given this lack of efficacy, surgical resection emerged as the most appropriate method for resolving the disorder, corroborating literature data. Additionally, considering that adjuvant

radiation therapy can positively influence the disease-free survival rate, this option was also performed[4].

High-intensity focused ultrasound (HIFU) and ablation techniques are two other types of local treatment. HIFU represents an emerging, non-invasive therapeutic option, deploying targeted ultrasound waves to induce thermal ablation of tumor cells, with preliminary studies showing promising outcomes. Cryoablation and radiofrequency ablation offer minimally invasive alternatives to traditional surgery, providing benefits in selected cases, particularly for patients with small, localized tumors and those unsuitable for extensive surgical procedures[3,4].

CONCLUSION

We presented a rare case of desmoid tumor in a male breast with chest wall invasion, which showed progression after neoadjuvant systemic treatment. Imaging was important in diagnosis, staging, and response evaluation, supporting multidisciplinary meetings for proper treatment planning.

AUTHORS' CONTRIBUTION

CLLR: conceptualization, formal analysis, investigation, writing – original draft. SQD: conceptualization, formal analysis, investigation, writing – original draft. CMAS: formal analysis, investigation, writing – original draft. BML: data curation, formal analysis, writing – review & editing. JAS: conceptualization, data curation, resources, writing – review & editing. LG: data curation, resources, writing – review & editing. CSG: data curation, resources, writing – review & editing. AGVB: project administration, conceptualization, data curation, methodology, writing – review & editing.

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