Bilateral Breast Fibromatosis: Case Report and Review of the Literature

Thomas V. Taylor, MD,[*†] and Jose Sosa, MD*[†‡]

*The University of Texas Medical Branch, Houston, Texas; †St. Joseph Medical Center, Houston Texas; ‡Universidad Autonoma de Tamaulipas, Mexico

Fibromatosis or desmoid tumor is a benign, slow-growing fibroblastic neoplasm originating from musculoaponeurotic stromal elements. These tumors are characterized by an infiltrative and locally aggressive growth pattern, frequent recurrences, but no metastatic potential. The etiology is unknown, but these tumors have been associated with trauma and genetic disorders. Breast fibromatosis is exceedingly rare and often misdiagnosed, comprising only 0.2% of breast tumors. Clinically, it might mimic other breast lesions, such as carcinoma. Only approximately 6 case series and 100 case reports of this disorder affecting the breast are documented, and only 5 cases of bilateral breast fibromatosis have been reported. We describe the case of a 20-year-old woman who presented to our institution with firm, nontender masses palpable in both breasts. Prior ultrasound was suspicious for carcinoma, and breast core biopsies were suggestive of phyllodes tumor. An excisional biopsy was necessary to establish the diagnosis of breast fibromatosis for both masses. A review of articles published on desmoid tumors and breast fibromatosis was performed with emphasis on articles published in the last 10 years. Fibromatosis should be considered in the differential diagnosis of patients presenting with hard breast lumps suspicious of other diseases. (J Surg 68: 320-325. © 2011 Association of Program Directors in Surgery. Published by Elsevier Inc. All rights reserved.)

KEY WORDS: breast fibromatosis, desmoid tumors, extra-abdominal desmoid tumors, spindle cell tumors, Gardner's syndrome, familial adenomatous polyposis

COMPETENCIES: Medical Knowledge, Patient Care

INTRODUCTION

Fibromatosis, which is also known as desmoid tumors (DTs) are extremely rare, with an incidence of less than 5 individuals per million of population per year,[1] comprising 3% of soft tissue tumors.[2] These tumors usually affect the abdominal wall of parous women within 25 and 35 years of age.[3] The mammary gland is one of the rarest sites in which fibromatosis has been described, representing a 4% of extra-abdominal DTs and only 0.2% of primary breast lesions.[4-6] The mean age for diagnosis of breast fibromatosis is 35 to 50.3 years. Only approximately 6 case series and 100 case reports of breast fibromatosis are available,[7] and only 5 of them report both breasts involvement. This case report provides an exceedingly rare presentation of breast fibromatosis in a 20-year-old woman who presented to our institution with firm, nontender masses palpable in both breasts.

CASE REPORT

A 20-year-old woman presented to our institution with the chief complaint of a palpable lump in the right breast that had been present for 5 months; this was associated with an occasional mild pricking sensation. She denied nipple discharge or changes in the breast skin. Her medical history was significant for parturition 9 months before her presentation, she was 4 months post lactation and a Mirena (levonorgestrel releasing intrauterine system; Bayer HealthCare Pharmaceuticals, Morristown, New Jersey) device had been placed for 5 months and removed 12 weeks before her visit. There was no history of trauma to the chest wall. Prior surgical history included 2 c-sections but no breast-related procedures. Her family history was negative for familial syndromes and malignancies like familial adenomatous polyposis (FAP), Gardner's syndrome (GS), breast cancer, or ovarian cancer.

On physical examination, a 10 cm × 4-cm mass in the upper inner quadrant of the right breast and a 4 cm × 4-cm mass in the upper outer quadrant of the left breast were present. These were both firm, ill circumscribed, and exhibited mild tenderness to palpation. The overlying skin was not attached to the masses and had no changes; there was a bilateral nipple retraction that had been present since childhood. Neither axillary nor supraclavicular adenopathy were found.

Bilateral mammography revealed a 5.4-cm mass in the upper inner right breast and a 3.6-cm central mass in the left breast, both with surrounding spiculation and no microcalcifications;

Correspondence: Inquiries to Thomas V. Taylor, MD, The University of Texas Medical Branch, 1315 Saint Joseph Parkway, Suite 1800, Houston, TX 77002; fax: (713) 652-3810; e-mail: j_sosah1985@hotmail.com

on ultrasound, these were reported as solid masses with irregular margins.

An evaluation of core biopsy tissue of both lesions showed a proliferation of spindle cells lacking significant cytologic atypia or increased mitotic activity; a few mast cells were identified and few ductal epithelial cells were observed. Within spindle cell areas, almost no epithelium was identified. These features indicated a phyllodes tumor (PT), but the presence of mast cells in a bland spindle cell lesion raised the possibility of a neurofibroma.

Initially, an excisional biopsy was performed on the right breast, which contained the bigger lesion. A nonencapsulated irregular mass occupying the breast tissue in the upper and medial part of the breast was found. This displaced most of the normal breast tissue inferolaterally; a wide margin excision of the tumor and surrounding tissue was performed.

Histopathology revealed a mesenchymal lesion with some areas suggestive of fibromatosis. Special stains were applied to determine whether this represented a form of fibromatosis or a smooth muscle lesion. Smooth muscle actin stain was negative, but β-catenin nuclear staining demonstrated an important positivity, confirming an extra-abdominal type of fibromatosis. A surgical excision with wide margins was next performed for the left breast lesion and the same pathology features and diagnosis were obtained. On both surgical procedures, the chest wall was not involved.

The postoperative courses for these procedures were uneventful, and the patient was discharged with a right breast mastectomy scheduled after pathology revealed that the specimen obtained on right breast had focal involvement of the inked margins on evaluation. After this, the patient will continue to be followed with annual magnetic resonance imaging evaluation for 3 years to evaluate for recurrence of fibromatosis in the involved regions.

DISCUSSION

DT is an uncommon, bland-appearing, and nonencapsulated soft tissue tumor comprised of a fibroblastic and myofibroblastic spindle cell proliferation and collagen. These lesions do not metastasize but are characterized by local aggressiveness with infiltrative growth pattern and a high recurrence rate[4,8,9]; lesions are usually solitary but are multiple in a 15% of all cases.[10]

McFarland in 1832 made the first histologic description of DTs. Johannes Müller (1801-1858), considered the first tumor pathologist, named these tumors, developed from the abdominal wall. He introduced the term "*desmoid*" from the Greek "*desmos,*" which means "*tendonlike*" in 1838.[11] Before Müller's assays no one had collected tissue samples from tumors to carry out detailed microscopic descriptions; therefore, they were described according to their macroscopic appearance.[12] Nichols applied first the term desmoid to extra-abdominal tumors with the same histopathologic characteristics as those described by Müller.[13]

DTS are extremely rare, comprising 3% of soft tissue tumors[2] and accounting for less than 0.03% of all neoplasms.[14,15] The incidence is less than 5 individuals per million of population per year.[1] In the United States, only 900 tumors are diagnosed annually with no significant racial or ethnic distribution.[16] The woman-to-man ratio was 1.2-1 in a 60-patient case series.[17] The maximum incidence occurs mainly within 25 to 35 years of age, usually in the abdominal wall of parous women.[3] The most common locations for nonabdominal lesions are the shoulder girdle, pelvis, and thigh.[18]

The histologic features are characterized by abundant collagen surrounding irregular bundles of spindle cells with regular nuclei; a low mitotic activity rate; and cellularity that varies from significantly collagenized low cellular lesions to those that are relatively cellular. Macrophages, lymphocytes, and giant cells are observed mostly peripherally.[19]

DTS are classified into 2 groups: The first group is intra-abdominal tumors, which are related with FAP, and 68% of DTs developed in these patients appeared in an abdominal surgery scar.[20] This group is also related with GS; both are autosomal dominant diseases with multiple colonic polyps, the latter with extra colonic features, including extra-abdominal DTs, osteomas, and epidermoid cysts. Approximately 2% of patients with a DT have polyposis coli.[21] FAP and GS are caused by mutations at chromosome 5q21-22, locus of the adenomatous polyposis coli (APC) gene. The second group are extra-abdominal tumors, which are mainly sporadic and likewise may exhibit APC gene mutation.[16,22] DTs can also be divided into superficial tumors, usually not achieving sizes bigger than 5 cm and not involving deep structures, and deep tumors, with a higher recurrence, more aggressiveness, a rapid growth pattern, often reaching very large sizes.[17] Tumors bigger than 54 cm have been reported.[23] In despite of rapidly increasing size, no hemorrhage or necrosis is found.[3]

Currently, the etiology of these tumors remains unknown,[7] as aforementioned germ line mutations seem to be involved and the foremost predisposing factor is the diagnosis of FAP or GS.[24] FAP patients have a 1000-fold increased risk of developing a DT, which occur in a 10% to 15% of them.[2] According to the CHAMP study group reported data, the most common cytogenetic abnormalities are chromosomal aberrations, present in 48% of all DTs, although this is less frequent in the superficial tumors. In deep extra-abdominal DTs, the 3 commonest features found are trisomies of chromosomes 8 and 20 as well as loss of 5q material like monosomy or interstitial deletion.[25] A mutation of a β-catenin interacting gene has been considered.[22] β-Catenin is a protein with an important role in the E-cadherin and Wnt pathways, which are both related with tumorigenesis.[26] A dysregulation of these pathways would allow β-catenin to accumulate and translocate to the nuclei interacting with several transcription factors, stimulating target oncogenes as cyclin-D1,[27] and constituting a risk for inherited or sporadic DTs. High levels of nuclear β-catenin staining are found in 71% of DTs, 40% of solitary fibrous tumors, 40% of endometrial stromal sarcomas, and 28% of synovial sarco-