Bilateral breast fibromatosis : a rare case report

Abstract

Breast fibromatosis is a rare benign tumor of soft tissues with local spread, often frequently recurrent but never results to metastasis. We present a case diagnosed in a 41 years old patient who was referred to a Douala private medical center in February 2012 presenting bilateral breast tumors. Histological analysis of the excised samples showed fibroblastic proliferation without atypia nor metastasis which reappeared 15 months in the left breast after the initial intervention. A mastectomy and lymp nodes curettage has shown resolution of the symptoms .*Key- words*: fibromatosis - histopathology – breast -Cameroon

Introduction

Fibromatosis is a rare tumor of the breast in females between puberty and menopause . That tumor can arise from the fascia musculo-aponeurotic of the chest wall or from fibroblasts and myofibroblasts of the mammary parenchyma [1, 2] This neoplasm is characterized by being locally aggressive with a frequent recurrence without metastasis [3,4]. The mammary fibromatosis represents only a particular aspect of these tumors, its frequency is less than 0.2% of the primitive tumors of the breast and 4% of the extra- abdominal fibromatosis [3]. Bilateral desmoid tumors have been reported in up to 4% of patients [3]. The clinical presentation is a palpable mass that is sometimes associated or not with skin modification [5]. The diagnosis is based on histological examination of the biopsy or surgical specimen [1, 2, 6]. We are reporting a case of bilateral desmoid tumor of the breast. The informed consent from the patient for this study was obtained.

Case presentation

A 41 years old female patient was referred to a private medical center in Douala town because of bilateral breast masses, a mass in the inferior external quadrant on the right and another mass in the upper external left breast. In the two cases, the masses were mobile, painless and they appeared polylobular with no apparent changes in skin pigmentation and without any suspecting lesion.

Mammography showed in the both cases large opaque masses with regular contours. The right breast measurement was 10x6x4 cm and the left was 11x7x5 cm. The cytological examination of the tissue aspirate taken under ultra-sound guidance showed abnormal cells.

The excised samples measured a 9x7x5 cm and weighted 58 g in the right breast In the left, we obtained the sample measurement of 7x4x3 cm and 46 g in weight (Figure 1). They were of firm consistency, well circumscribed with smooth and whitish surface homogenous on different sections. The specimens were treated according routine technic with fixation in 10% formalin, staining in eosin-haematein and interpretation under optical microscope.

Figure 1. Breast fibromatosis resected specimen

In histological examination, the lesion was characterized by well and differentiated fibroblastic proliferation, which was rich in collagen without atypia, no images of mitosis .In some areas, simple mesenchymatous tissue and few blood vessels were observed (figure2)

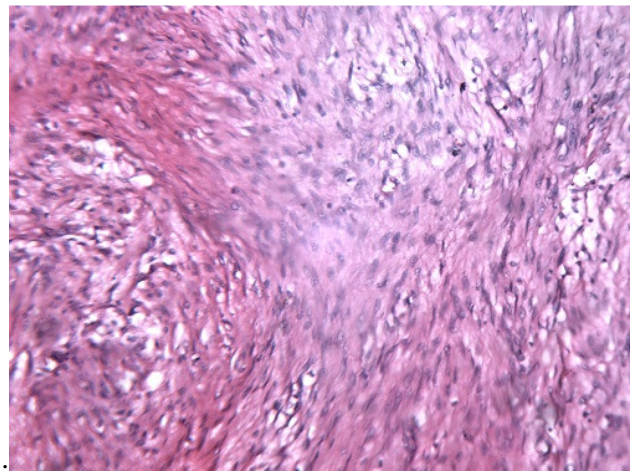

Figure 2. Fibroblastic proliferation, without atypia.

The patient was carefully followed up. 15 months after the initial operation, the patient was seen with a recurrence on the left previous tumor site. A left mastectomy was with axillary curettage (lymph nodes clearance) was done. The histological aspect was similar to the previously described. The right breast and the left axillary nodules were normal. A careful follow-up over the past 26 months did not show any recurrence.

Discussion

Desmoid tumors (DT) also known as aggressive fibromatosis (AF) constitute a rare fibroblastic proliferative disease. They may occur in any musculoaponeurotic or fascial tissue [7]. These tumors generally are divided by anatomic designation as extra-abdominal, abdominal, or intra-abdominal and the most common locations are around the limb girdles or the proximal extremities, the abdominal wall, and intra-abdominal or mesenteric [8, 9]. The mammary fibromatosis is a rare lesion; bilateral type have been reported in up to 4% of

patients and mostly it occurs in the breasts of women between 13 to 80 years with an average of 43 years, regardless of their ethnicity [2,3,5]. Our presented case, with her breast location, the bilateral position and the age confirms the data of the literature make it a rare and interesting case.

Clinically, our patient presented bilateral breast masses, a mass in the inferior external quadrant on the right and another mass in the upper external left breast, associated with painless and no apparent changes in skin pigmentation with no suspecting lesion . Wongmaneerung et al described two cases of bilateral breast fibromatosis with pain (of the patients) and nipple retraction, the presence of skin involvement and invasion by the tumor (in another patient) [5]. Mehdi et al reported a case with ulceration of all the nipple-areolar plate [1]. That means the clinical manifestations of breast fibromatosis are variable, making each case is exceptional.

Although his etiopathology is not fully understood, some theories have however been issued. Most desmoids arise sporadically, slightly more in women than in men, with some DTs related to pregnancy and trauma, and others associated with hereditary cancer syndromes [10]. Desmoid tumors are results of deregulation of connective tissue growth. Increased nuclear expression of β-catenin, a protein responsible for regulation of gene expression, proliferation and survival, is the characteristic feature in sporadic DT [10]. Antecedent trauma, often surgical, has been noted at the site of the DT in approximately 25% of cases [11]. Estrogen receptors (ER) were observed in 33% of all DT examined, with an equal incidence in males and females and with antiestrogen binding sites found in 79% of samples, including some which were ER negative [11]. Implant-associated breast desmoid tumors may also occur [12].

In this case, mammography showed in the both breast large opaque masses with regular contours. In the literature, Mammography typically show a high density, spiculated, stellate tumour without microcalcifications often indistinguishable from carcinoma [4, 13]. The lesion sometimes appears as a mass with a lobulated or no outlines, but still well de-fined. In some rare cases, the lesion has no mammographic presence [14, 15]. Although Ultrasound appearances are various, they are not specific. Demoid tumours could be poorly marginated; it can be an irregularly shaped hypoechoic mass of desmoid tumours showed or even a small-sized lesions that are homogeneous lesions with very regular outlines [15, 16]. By some authors, the magnetic resonance imaging (MRI) and the scanner are important for the estimation of a possible parietal invasion; MRI can also play an invaluable role in preoperative diagnosis and planning. Desmoid tumors are typically isointense on T1-weighted imaging and demonstrate low to high signal intensity on T2-weighted imaging. After the injection of chelated forms of gadolinium, in T1-weighting, and after a fat signal saturation, we notice a heterogeneous raising in the ill-defined outlines [12, 15].

Based on avaible reports in the literature , fine needle aspiration cytology although nor entire specific may reveal some important information in patients with breast fibromatosis [17].The cytoponction could confirm the diagnosis by isolating fibroblasts with minimal

atypia; however, it is mostly little contribution [2]. In our case, the cytological examination only showed abnormal cells.

The histological examination remains the basis for the diagnosis of this pathology. For our patient, histologically they found well and differentiated fibroblastic proliferation, which was rich in collagen without atypia, with no images of mitosis. In some areas, simple mesenchymatous tissue and few blood vessels were observed. According to the literature, the tumor usually has a poorly circumscribed pattern and is composed of proliferating stellate to spindle cells arranged in long fascicles or whorling patterns with bland nuclear features and dense keloid-like collagen in areas [10]. Ultrastructural studies indicate that the spindle cells have features of both fibroblasts and myofibroblasts. Architecturally, the tumor cells are typically arranged in long, sweeping fascicles, and vague whorls in a background of eosinophilic, collagenized stroma with prominent thin-walled vessels. The stroma can also show myxoid features, reportedly more common in the breast and the mesentery, but can be seen in other more common sites [11].

Immunohistochemistry, specifically β-catenin, and more recently, molecular diagnostics can play an important role in its diagnosis. The positive presence of actin and vimentin is useful for the diagnosis of desmoid tumor. Desmin is rarely positive, whereas S100 and CD34 are usually negative; β-catenin nuclear staining is an option for diagnosis, which may be only focally positive. Sporadic DTs are commonly associated with somatic mutations of the codons 41, 45 of exon 3 of the beta-catenin gene (CTNNB1). As such, antibody to β-catenin is useful in distinguishing desmoids from its histologic mimics, which generally lack this feature [5, 10, 11, 18].

The differential diagnosis of desmoid tumor vary from benign reactive lesions such as a hyperproliferative scar, to a more sinister fibrosarcoma [11]

The treatment in this patient was surgical. Radiotherapy was not directed demanded in this case. The management of breast fibromatosis includes surgical excision with clear margins, Systemic therapy and radiation therapy [6, 11]. Surgical therapy remains the cornerstone of desmoid tumor management. The mastectomy can be indicated for vast fibromatosis or for too large recurrences. In case of parietal invasion, the surgery can be extremely decaying going as far as taking away the breast, the pectoral muscles, the thoracic wall, and the parietal pleura [15]. Obtaining microscopically negative margins (R0 resection) is the preferred objective in oncologic surgery, including resection of desmoid tumors. Not all positive margins were recurrent; the true impact of surgical margin negativity as well as the role of adjuvant therapies (i.e., radiation, antiinflammatory drugs, antiestrogen agents, and cytotoxic chemotherapy) in preventing tumor recurrence remains uncertain [11]. Recurrent disease has been observed in negative margins, some authors proposed the algorithm of a more conservative wait-and-see approach in a less critical site with an asymptomatic lesion [19]. If the lesion progresses by RECIST (response evaluation criteria in solid tumor) criteria, then treatment is needed. Desmoid tumors respond to systemic medical therapies, from anti-inflammatory drugs to standard chemotherapy agents. Liposomal doxorubicin, doxorubicin-based regimens, methotrexate in combination with vincristine, and vinorelbine have all

demonstrated notable responses in this disease. Molecular-targeted agents, antiinflammatory drugs, hormonal agents, and interferons have all shown various degrees of activity in DT. However, the mechanisms through which these compounds conduct their antitumor activity are not completely understood [20].

Radiotherapy can be used in patients who have recurred after initial surgery and as primary treatment in those patients who are medically inoperable or those to whom resection presents unacceptable morbidity [21]. Some studies have shown a beneficial effect of the radiotherapy with 60% of decreasing cases non-accessible to the surgery. Therefore, the radiotherapy could be curative in recurrence cases when the surgery is impracticable [22, 23]. The use of radiotherapy must be considered carefully in light of the potential long-term side effects of radiotherapy including the risk of secondary malignancy, particularly in younger patients. Treatment decisions are individualized to each patient depending on a range of patient and tumor characteristics [24].

Conclusion

Breast fibromatosis is a begnin affection which can clinically, radiologically , cytologically and in the curse of surgical intervention , be mistaken as a malignant lesion . The bilateral form is extremely rare. A well-coordinated, multidisciplinary approach involving the input of surgical and nonsurgical specialists is needed to develop an individualized treatment strategy appropriate for each specific patient.

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