Journal of Pathology & Microbiology

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Austin Publishing Group

Case Report

Extra Abdominal Desmoid (Fibromatosis) of the Breast-Report of a Case Highlighting Diagnostic Difficulties and Review of Literature

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Received: December 01, 2016; **Accepted:** December 18, 2016; **Published:** December 21, 2016

Abstract

Fibromatosis of breast, also known as Extra Abdominal Desmoid tumour is extremely rare and poses a diagnostic challenge as it may mimic carcinoma both clinically and radio graphically. We report a case of thirty year old female, who presented with a firm mass in the right breast. The clinical and ultrasound diagnosis was suspicious of carcinoma and since the fine needle aspiration cytology was non diagnostic, an excision biopsy was performed. Routine histopathological examination showed proliferating spindle cells in long fascicles with infiltrative margins. Mitotic figures were less than two per ten high power fields. The differential diagnosis considered were collagenized or fibrous variant of myofibroblastoma, nodular fasciitis, low-grade fibrosarcoma, spindle cell metaplastic carcinoma, fibrous histiocytoma and scar from healed fat necrosis. Immunohistochemical examination revealed the spindle cells to be positive for vimentin, smooth muscle actin, beta catenin and negative for cytokeratin, S100 and desmin. Hence the diagnosis of fibromatosis was rendered, following which a completion wide excision was performed. The patient is asymptomatic without recurrence on eight months follow up. To conclude, we present this rare case and discuss its diagnostic difficulties, differential diagnosis and the role of Immunohistochemistry in evaluation.

Keywords: Extra abdominal desmoids; Fibromatosis; Breast; Immunohistochemistry

Abbreviations

CD: Cluster of Differentiation

Case Presentation

A thirty year old female presented to the surgical outpatient department with complaints of swelling in the right breast of 6 months duration. On clinical examination a 3.5X3 cm firm mass was palpable in the upper outer quadrant. Ultrasound examination showed a hypo echoic infiltrative mass and mammographic examination revealed a stellate tumour suspicious of malignancy. A Fine needle aspiration cytology performed elsewhere, was non diagnostic and hence an excision biopsy was done.

The mass was received in the histopathology laboratory in ten percent neutral buffered formalin. Grossly the mass measured 3X2.5 cm. The cut surface of the mass was gray white, homogenous and firm (Figure 1). Routine processing of the tissue was done, paraffin embedded blocks obtained and 4 micrometer thick sections were cut and hematoxylin and eosin staining was done. Microscopy showed a tumour with infiltrative borders. The spindle cells were arranged in interlacing fascicles, bundles and in focal storiform pattern admixed with abundant blood vessels and collagen bundles (Figure 2A). The cells had uniform oval to spindle nuclei with no pleomorphism. The tumour was more cellular at the periphery with few compressed ducts and had tendency to collagenization in the centre. Focal areas of hyalinization, odema, stromal lymphocytic and eosinophilic infiltrates were noted (Figure 2B). The mitotic figures were less than 1 per 10 high power fields. There were no areas of necrosis or pleomorphism. Immunohistochemistry revealed positivity for vimentin (Figure 3A) smooth muscle actin (Figure 3B) and beta catenin (Figure 5). The tumour cells were negative for S100, desmin and CD34.

After the diagnosis of fibromatosis was rendered, a completion wide excision was performed and the microscopic sections from this specimen showed features of post surgical changes including mixed inflammatory infiltrates, focal areas of fat necrosis, exuberant fibroblastic proliferation, numerous proliferating capillaries and collections of foamy histiocytes (Figure 4). There was no evidence of residual tumour. On follow up, the patient is asymptomatic without

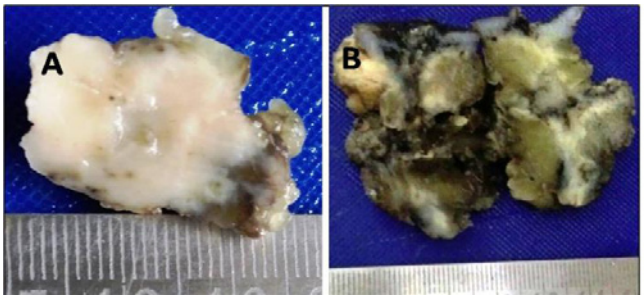

Figure 1: Gross appearance. **A)** Cut surface of the tumors appearing gray white, homogenous and firm. **B)** Cut surface of the completion excision specimen yellow, lobulated, soft with focal whitish areas.

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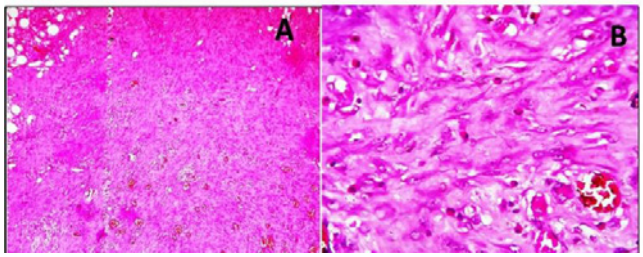

Figure 2: Microscopic appearance of initial excision biopsy specimen hematoxylin & eosin. **A)** Bland spindle cells arranged in long interlacing fascicles and bundles with abundant blood vessels in between (40X). **B)** Spindle cells showing classic myfibroblastic features, including indistinct eosinophilic cytoplasm, uniform vesicular nuclei with occasional nucleoli and interstitial collagen with scattered eosinophils (100X).

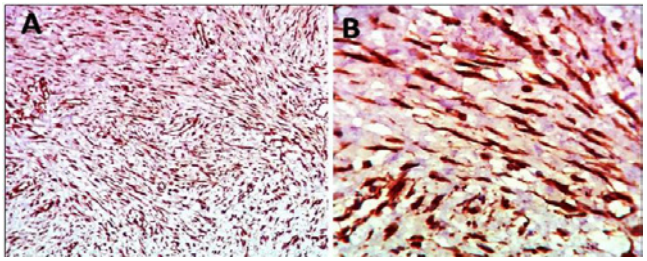

Figure 3: Immunohistochemistry. **A)** Spindle cells showing positivity for smooth muscle actin (100X). **B)** Spindle cells showing positivity for vimentin (400X).

recurrence, eight months after surgery.

Discussion

Fibromatosis or extra abdominal desmoid tumour of the breast is a locally infiltrating, histologically low-grade spindle cell proliferation composed of fibroblastic cells with variable amounts of collagen [1]. Desmoid tumours comprise 0.2% of all breast tumours 2 and 0.3% of all solid tumours [1]. It occurs in a wide age range of 15 to 80 years with a median age of presentation between 25 and 45 years. It usually presents as an ill defined firm to hard breast mass mimicking carcinoma on clinical examination. Skin retraction or dimpling and nipple retraction when present, reinforces the diagnosis of carcinoma. Mammography usually reveals a stellate tumour that may be impossible to differentiate from carcinoma [3]. Majority of cases is sporadic but occasional cases are associated with Gardner's syndrome [4] and familial adenomatous polyposis syndrome [5], suggesting a relationship with beta catenin gene on chromosome 22. High oestrogen states like pregnancy poses a significant predisposing factor for development of desmoid tumour. 6 Association of fibromatosis with silicone breast implants and trauma is also reported [6].

Fibromatosis of the breast usually presents clinically as a painless, mobile, hard lump mimicking a carcinoma. Some cases may have retraction of nipple or tethering of skin further supporting the clinical diagnosis of carcinoma [7]. Our case also presented as hard firm lump with clinical suspicion of carcinoma.

Most desmoid tumours are high density, irregular masses with speculated margins and no calcification, mimicking carcinoma of the breast [3]. Mammographic findings of this case were suspicious of

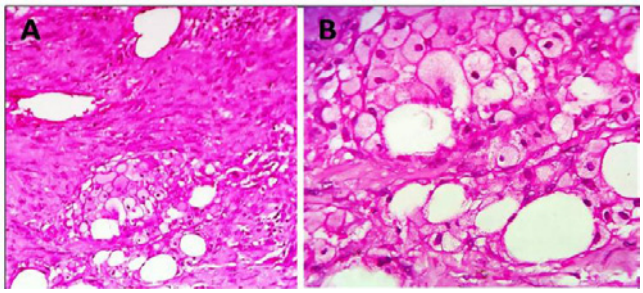

Figure 4: Microscope appearance of completion excision specimen showing postsurgical features, hematoxylin & eosin. **A)** Fibroblastic tissues with mixed inflammatory infiltrates, focal areas of fat necrosis, proliferating capillaries and collections of foamy histiocytes (100X). **B)** Focal areas of fat and collections of foamy histiocytes (400X).

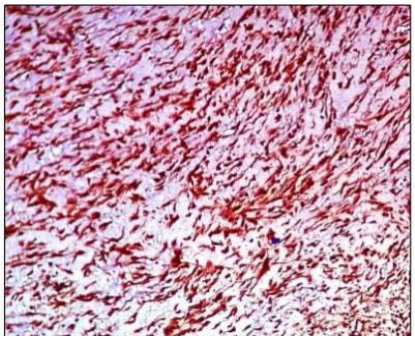

Figure 5: Immunohistochemistry showing positivity for beta catenin (100X).

malignancy.

Fine needle aspiration cytology is usually non diagnostic and most cases only a diagnosis of benign spindle cell lesion can be offered. Some cases may be false positively diagnosed as malignancy in cytology smears [8].

Grossly, the desmoids tumours may be of sizes from one to ten centimetres with an average of 3cms. It is usually ill circumscribed with poorly defined margins or occasional cases have a stellate appearance. The cut surface is gray white or tan and firm or rarely can have a whorled appearance [9].

Microscopically it is composed of spindle cells arranged in fascicles, interlacing bundles, broad sheets or in storiform pattern along with variable amounts of collagen in between. Usually the collagen is relatively uniform, but in occasional cases, collagen may be abundant giving a keloid like appearance. Pleomorphism and mitosis are very rare. In very few cases, round cytoplasmic eosinophilic inclusion bodies, measuring 3 to 10 mm were noted in juxta nuclear position in the tumor cells. Special stains showed that these inclusions are positive for masson trichrome. Immunohistochemistry revealed positivity for muscle specific actin in the inclusions [10]. Immunohistochemically, these tumours show positivity for Beta cartenin, alpha-smooth muscle actin, oestrogen receptor, factor XIIIa, CD34, vimentin and cathepsin D [11]. Thus the spindle cell is shown to be of myofibroblastic origin by Immunohistochemistry [11]. Ultra structural studies also confirm that, these spindle cells are of myofibroblastic origin [10]. The differential diagnosis include:

collagenized or fibrous variant of myofibroblastoma, spindle cell metaplastic carcinoma, nodular fascitis, fibrosarcoma, fibrous histiocytoma and scar from trauma, previous surgery or healed fat necrosis.

Collagenized or fibrous variant of myofibroblastoma is composed of benign spindle cells admixed with variable amounts of collagenous stroma. But in myofibroblastoma, there will be formation of irregular slit like spaces between tumour cells, thus differentiating from fibromatosis. They are positive for vimentin, calponin, CD10 and CD34 by immunohistochemistry [12].

The spindle cell metaplastic carcinoma can mimic fibromatosis, if the epithelial component is absent. But metaplastic carcinoma is a highly cellular neoplasm, exhibiting significant nuclear pleomorphism and many mitotic figures, whereas presence of desmoid like foci and collections of lymphocytes point towards the diagnosis of fibromatois. Immunostains such as cytokeratins and CD10 characterize metaplastic carcinoma, while recent literature suggests that nuclear positivity for β-catenin favours the diagnosis but is not specific for fibromatosis [13].

Another differential diagnosis to be considered is the nodular fascitis. Nodular fasciitis is more circumscribed with a high mitotic count and abundant inflammatory infiltrates scattered throughout the lesion. Nevertheless, fibromatosis has very less mitotic figures and more collagenized stroma, in contrast to the granulation tissue like or myxoid or tissue culture like stroma seen in cases of neurofibromatosis [1].

One more neoplasm included in the differential diagnosis is fibrous histiocytoma. Even though fibromatosis of breast may show focal storiform areas, this is rarely a prominent pattern. In addition the histiocytic, multinucleated and epithelioid cells frequently found in fibrous histiocytoma are not seen in cases of fibromatosis [14].

Scars from remote trauma, previous surgery and healed fat necrosis should be differentiated from fibromatosis. Fat necrosis frequently shows calcifications which are very rare in fibromatosis. Post surgical scars may show remnants of suture material and foreign body type of granulomatous reaction [14].

Finally, fibrosarcoma should also be considered in differential diagnosis, but it is a highly cellular tumor, with cells arranged in long fascicles and distinct hering bone pattern. The cells exhibit significant cytological and nuclear atypia. And in contrast to fibromatosis, there are numerous mitotic figures [14].

The treatment of choice includes a complete wide local excision and this surgical clearance is almost curative in most patients of Fibromatosis [15]. However fibromatosis has a tendency to recur locally and literature review reveals a few cases with local recurrence [15].

Conclusion

To conclude, fibromatosis of the breast may pose diagnostic difficulty due to its clinical and mammographic features mimicking malignancy; nevertheless careful histopathological examination will reveal the true nature and Immunohistochemistry will aid in difficult situations.

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J Pathol & Microbiol - Volume 1 Issue 2 - 2016
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