

**DESMOID MESENCHYMAL FIBROMATOSIS: ABOUT A CASE AND REVIEW OF THE LITERATURE****Khadija Tamim\*, Karima El Matar, Merouan Serroukh, Samir Bargach and Sabah Elamrani.**

CHU Avicenne, Hôpital Maternité Souissi, Service de Gynecologie-Obstétrique-Cancérologie-Grossesse à Haut Risque, Rabat, Maroc.

**\*Corresponding Author: Khadija Tamim**

CHU Avicenne, Hôpital Maternité Souissi, Service de Gynecologie-Obstétrique-Cancérologie-Grossesse à Haut Risque, Rabat, Maroc.

Article Received on 11/04/2021

Article Revised on 01/05/2021

Article Accepted on 21/05/2021

**ABSTRACT**

Mammary desmoid fibromatosis is a rare pathology that clinically and radiologically mimics a malignant tumor. It is characterized by a strictly local evolution but with a tendency to infiltration and recurrence. Its diagnosis is based on histological examination and its treatment is mainly surgical. The differential diagnosis arises mainly with metaplastic spindle cell carcinoma. Complete surgical excision with healthy margins (up to 3cm) is the treatment of choice, radiotherapy remains a therapeutic option in addition to surgery in incomplete resections and in the event of multiple recurrences. Through our case and a review of the literature, we will try to focus on the diagnosis of this rare entity and its management since it will condition the prognosis.

**KEYWORDS:** desmoïde tumor, brest, mammary fibromatosis, histology, surgery.**INTRODUCTION**

Desmoid fibromatosis is a mesenchymal tumor, first described in 1832. Breast localization is very rare, Unlike cancers, they have no risk of metastasis but they are locally invasive, infiltrating, causing pain and / or a functional gene and present a high risk of local relapse after surgery. It develops from the supporting tissues and the aponeuroses. It is made up of elongated cells that resemble normal cells in fibrous tissue (fibroblasts).

It is a benign tumor which does not give rise to metastasis, which can be invasive locally, causing pain or deformity, and for which surgical removal is difficult. Its development is unpredictable; some forms relapse repeatedly and rapidly, others evolve in spurts, interspersed with a phase of prolonged stability. In contrast, there are forms which regress spontaneously. The exact causes are still unknown,

**Patient and Observation**

54-year-old patient, admitted for a nodule in the left breast, with a history of dyslipidemia under diet alone, type 2 diabetes under oral antidiabetics, cholecystectomy 08 years ago, G4P3 with 3 living children in good health, with the notion of taking contraception for several years, who presents a nodule of the left breast discovered on autopalpation with a mastodynia without other associated sign, in whom the examination objectified a straddling nodule of the infero-internal quadrant and the sub-mammary groove, of about 3cm of firm consistency,

Irregular contours fixed with respect to the superficial plane with sign of the dimpling, mobile with respect to the deep plane painful on palpation without inflammatory sign opposite, and without axillary lymphadenopathy.

An echo-mammography: was carried out, objectifying a nodule of the QII on the oval submammary fold, poorly limited with indistinct outlines, heterogeneous isoechoic with posterior attenuation, measuring 14 \* 19 \* 9mm; left breast: ACR4, right breast: ACR1

A trucut biopsy: mammary parenchyma devoid of specific lesion or tumor.

A large extemporaneous lumpectomy was performed in which anapath was in favor of a desmoid mesenchymal tumor without signs of malignancy with excision margins of more than 3cm, and the beta catenin marker positive. The evolution has been unremarkable.



Figure 1: clinical appearance of the nodule.

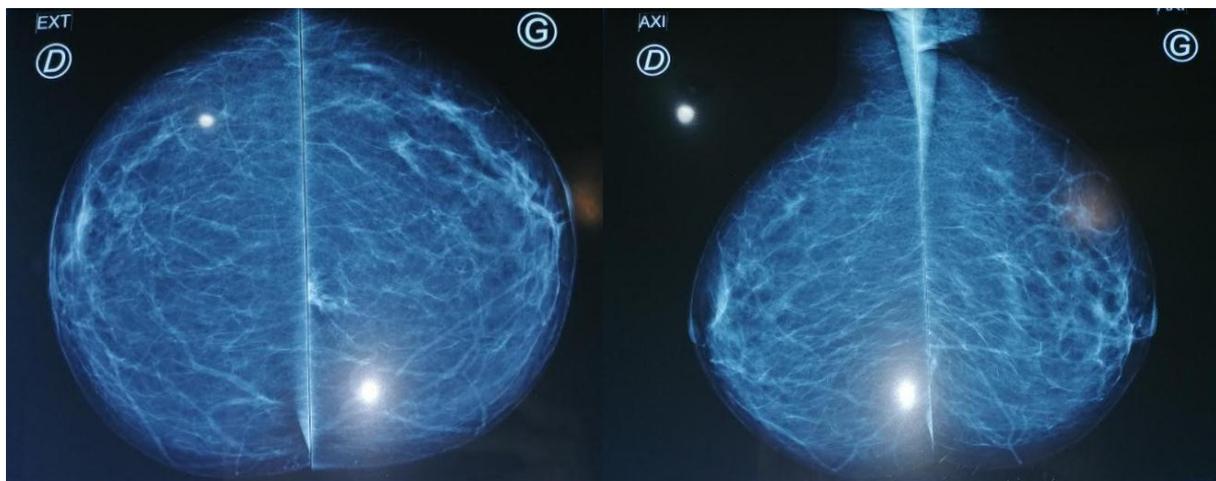


Figure 2 : mammographic aspect of the lesion.

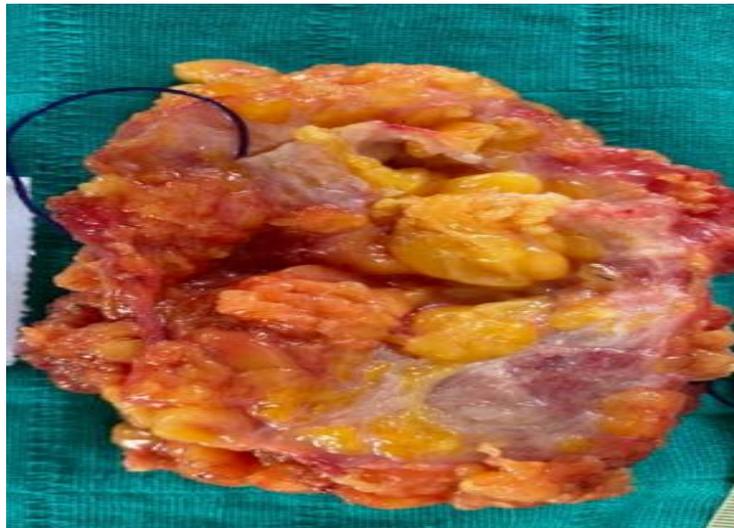


Figure 3: piece of lumpectomy.

## DISCUSSION

Desmoid mammary tumors are very rare, around 100 cases have been reported in the medical literature to date. The frequency reported by the various authors of desmoid mammary tumors, whatever the sex, does not

exceed 3.5% of fibrous tumors, and 0.2% of all breast tumors.<sup>[1-2]</sup>

Breast localization is encountered at any age. This mammary desmoid fibromatosis can also be seen in men.

The pathophysiology is the dark side of this pathology, it could result from the summation of these 3 factors:

- initiation of cell proliferation following trauma, or after breast surgery,<sup>[1,3,4]</sup> or after breast prosthesis;<sup>[5,6]</sup>
- a promoting effect of sex hormones: several arguments plead in favor of this effect in addition to the hormone dependence of desmoid tumors, since fibromatosis occurs preferentially in multiparas, during pregnancy, in a context of hyperestrogenia or during taking oral contraception;<sup>[7,8]</sup>
- a genetic ground by disturbance of the regulation of the growth of fibroblasts.

The occurrence of fibromatosis could be the witness of a family genetic abnormality. In fact, cases of isolated multicentric familial fibromatosis have been described; or part of a GARDNER syndrome associating colonic polyposis, bone malformations, epidermoid cysts and soft tissue tumors including desmoid tumors. In 1986, REITAMO defined desmoid syndrome as an entity associating fibromatosis with skeletal anomalies (exostoses, geodes; on densities in the femurs and mandibles;

sacralization of the 5th lumbar vertebra). This syndrome is said to be autosomal dominant with variable penetrance.<sup>[9]</sup>

Clinically, this tumor manifests as a mass, firm, of variable size, painless most often peripheral, which can be associated with skin thickening or nipple retraction. Radiologically, we can have a non-calcified spiculated mass on mammography, micro or macrocalcifications are rare, on the other hand in 1/3 of cases no lesion is objectified on mammography. The ultrasound appearance corresponds to a solid mass spiculated or microlobulated irregular, hypoechoic, one can even objectify an attack of the pectoral or intercostal muscles, therefore this lesion mimics on the clinical and radiological level a malignant lesion. Breast MRI is of no interest for diagnostic purposes, on the other hand it can assess the tumor extension of large lesions by objectifying a mass with poorly limited outlines, spiculated iso intense to the muscles, in iso / T2 hypersignal of variable and heterogeneous intensity.<sup>[10]</sup>

Histologically, it is a poorly defined lesion, star shaped, firm and whitish macroscopically mimicking a neo. The microscopic examination reveals a proliferation of spindle-shaped cells (fibro and myofibroblastic without nuclear atypia), arranged in bundles, mixed with bands of collagen, without associated epithelial component. In immunohistochemistry, cells express intensely and diffuse smooth muscle actin, so desmin is only expressed by rare cells (myofibroblastic profile). Very frequently there is positivity for beta-catenin which is characteristically localized to nuclei. Estrogen and progesterone receptors are not detectable by immunohistochemistry in mammary fibromatosis, like its

extramammary counterpart, although for the latter, therapeutic responses to hormone therapy have been reported.

The differential diagnosis mainly with metaplastic spindle cell carcinoma, It also arises in front of a phyllodes tumor of intermediate or malignant aggressiveness, in front of fibrosarcoma or myoepithelial carcinoma or even in front of a fibrous scar because they are the same spindle cells but c ' is the inflammatory contingent present in the event of a fibrous scar that will make the difference. The particularity of this fibromatosis whatever the site is the infiltration of the adjacent tissue.

Treatment is essentially based on complete surgical excision with healthy margins (up to 3cm).<sup>[11,12]</sup> Recurrences are frequent from 18 to 29% (3 to 6 years), the thoracic involvement of muscles and ribs is possible. In some women, a mastectomy is recommended in the event of multiple recurrence, in the event of a tumor of large volume or in the event of difficulty of the histological diagnosis. The place of radiotherapy is very controversial in the literature, its efficacy is dose dependent, and tumor control is 60 to 80% for a total administered dose of 50 to 60 GY. Other adjuvant treatments have also been tried: anti-inflammatory, anti-estrogen and low-dose chemotherapy.

## CONCLUSION

Breast fibromatosis is an invasive benign tumor. Its radiological appearance is suggestive of a malignant tumor. Its diagnosis is pathological and its surgical excision must be large and carried out in healthy areas so as to reduce the risk of recurrence. Radiotherapy could be reserved for incomplete resections or in certain recurrent forms.

**Conflicts of interest: none.**

## BIBLIOGRAPHY

1. Erguvan-Dogan B, Dempsey PJ, Ayyar G, Gilcrease MZ. Primary desmoid tumor (extra-abdominal fibromatosis) of the breast. *AJR Am J Roentgenol*, 2005; 185(2): 488-9.
2. Schwarz GS, Drotman M, Rosenblatt R, Milner L, Shamonki J, Osborne MP. Fibromatosis of the breast: case report and current concepts in the management of an uncommon lesion. *Breast J*, 2006; 12(1): 66-71.
3. Neuman HB, Brogi E, Ebrahim A, Brennan MF, Van Zee KJ. Desmoid tumors (fibromatoses) of the breast: a 25-year experience. *Ann Surg Oncol*, 2008; 15(1): 274-80.
4. Harvey SC, Denison CM, Lester SC, DiPiro PJ, Smith DN, Meyer JE. Fibrous nodules found at large-core needle biopsy of the breast: imaging features. *Radiology*, 1999; 211(2): 535-40.
5. Goel NB, Knight TE, Pandey S, Riddick-Young M, De Paredes ES, Trivedi A. Fibrous lesions of the

- breast: imaging- pathologiccorrelation. *Radiographics*, 2005; 25(6): 1547-59.
6. Mesurolle B, Leconte I, Fellah L, Feger C, Nakazono T, Kudo S. Dynamic breast MRI in recurrent fibromatosis. *AJR AmJRoentgenol*, 2005; 184(2): 696-7. (author reply697).
  7. Reitamo JJ, Scheinin TM, Häyry PH. The desmoid syndrome: new aspects in the cause pathogenesis and treatment of the desmoid tumor. *Am J Surg*, 1986; 151: 230- 7.
  8. Mungas JE, Platz CE, Block GE. Desmoid tumors of the abdominal wall. *Surg Clin N America*, 1976; 56: 207-18.
  9. Corbisier C, Garbin O, Jacob D, Weber P, Muller C, Cartier J, Muller J, Plumere C, Dellenbach P. Tumeur rare du sein: la fibromatose mammaire. *Journal de Gynécologie Obstétrique et Biologie de la Reproduction*. Juin, 1997; Vol 26(N° 3): p 315.
  10. Glazebrook KN, Reynolds CA. Mammary fibromatosis. *Am JRoentgenol*, 2009; 193(3): 856-60.
  11. Khayat Raphael, Martins Rogeria, Becette Véronique, Engerand Sandrine, Berment Hélène, Barukh Yonathan, Mohallem Marina, Langer Adriana, Guinebretiere Jean-Marc, Cherel Pascal. Fibromatose mammaire: corrélations anatomo-radio-cliniques et revue de la littérature. *Imagerie de la femme*, 2012; 22(2): 100-109.
  12. Lee A H S. Recent developments in the histological diagnosis of spindle cell carcinoma, fibromatosis, and phyllodes tumor of the breast. *Histopathology*, 2008; 52(1): 45-47.