

# Desmoid Tumor of the Breast as a Manifestation of Gardner's Syndrome in an Elderly Woman: A Case Report and Review of Literature

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## Abstract

Desmoid tumor of the breast is an extremely rare entity. Histologically, it corresponds to a benign fibroblastic proliferation of a local evolution, with a high recurrence potential. We report the first case of elderly woman presented with desmoid-tumor of the breast as a manifestation of Gardner's syndrome. A brief literature review was provided. We report the case of a 72-year-old woman who presented with five months history of painful and an exophytic mass of her left breast. She is known to have familial polyposis and had a total colectomy with Gardner's syndrome. She had a history of osteomas of the maxilla. On clinical examination, there was an exophytic painless mass, on the upper medial quadrant of the breast. She had also lipoma in the left leg and pigmented skin lesions in legs. Her mammograms showed a suspicious stellar image. A computed tomography scan showed an ovoid lesion. Excision of the lesion was performed. Histopathology confirms a desmoid-tumor of the breast. Evolution was marked by lesion recurrence. In conclusion, desmoid tumor of the breast is rare, non-metastatic but locally aggressive. Clinical expression is often nonspecific. Treatment remains controversial; surgical excision is the treatment of choice.

## Keywords

Desmoid-Tumor, Gardner's Syndrome, Elderly, Breast, Familial Polyposis

## 1. Introduction

Desmoid tumors (DT) are rare, characterized by local malignancy with a high

tendency to recur. It is a monoclonal myofibroblastic proliferation producing abundant hyalinized collagen tissue desmoid-type. It is often poorly limited and invades surrounding tissues, making it difficult to remove. The risk of recurrence after excision surgery is very variable according to the studies, which makes the choice of their optimal treatment difficult. The desmoid-tumor has no metastatic potential.

It develops preferentially in the rectus abdominis muscle after an operation or a pregnancy, but can also develop on the limbs, in the neck or at the mesentery. These tumors are usually located in the abdomen or in the abdominal wall, sometimes they are extra-abdominal [1]. DT of the breast is extremely rare, the incidence is <0.2% of primary breast neoplasm. Its occurrence is more frequent and common in young women. Genetic factors in addition to trauma and hormonal factors are the primary known etiologies [2]. In Gardner's syndrome, the incidence ranges from 4% - 17% [3].

We presented a rare case in the elderly woman, to discuss the diagnosis and management of a patient with DT as a manifestation of Gardner's syndrome.

## 2. Case Presentation

A 72-year-old woman presented with a five-month history of breast pain with an exophytic mass of her left breast. She is a smoker and non-obese woman. She is known to have familial polyposis. At the age of 30, she had a subtotal colectomy. At the age of 48, she had rectal resection with ileoanal anastomosis for polyps' recurrence on that remaining digestive tube. Besides, she had a history of dental abnormalities with supernumerary and impacted teeth. At the age of 35, she had an osteoma of the left maxilla removed at the Department of oral and maxillofacial surgery.

Her physical examination revealed a small breast. On the upper medial quadrant of the left breast, there was an exophytic painless mass, resulting in a visible arch in profile with no skin adhesion, no nipple discharge, and no lymphadenopathy. The right breast was normal. The diagnosis of lipoma was initially suspected. She had also lipoma in the left leg and pigmented skin lesions in legs.

The performed mammogram (**Figure 1**) showed a suspicious stellar image and the ultrasound showed a hypoechoic lesion with a dense background. A computed tomography (CT) scan (**Figure 2**) with contrast injection showed an ovoid lesion measuring 30 \* 25 mm, located in the retro-glandular fat, in front of the pectoral muscle.

A core biopsy was then performed and has shown a benign lesion consisting in conjunctive hyperplasia with no apparent evidence of malignancy. But because of the FP history, diagnosis of breast desmoid tumor was suspected, and multidisciplinary meeting's decision was tumor resection.

Excision of the lesion was performed by direct incision next to the tumor, allowing perfect exposal of the lesion. The resection was performed at 2 cm of macroscopic distance from the palpable area of the tumor, including fascia and



**Figure 1.** Mammogram of the left breast showing a stellar image in the exophytic mass.



**Figure 2.** CT scan showing in left breast a retro-glandular homogeneous lesion with irregular limits.

pectoralis muscle next to the tumor. The lumpectomy piece of the left breast measured  $80 \times 60 \times 25$  mm. At the cut, a whitish, firm, poorly limited tumor measuring  $35 \times 28$  mm was located at 5 mm from one of the surgical excision margins. The histological study revealed a proliferation of spindle cells with moderate collagen giant cells without mitotic activity consistent with a fibromatosis. The diagnosis of Gardner's syndrome with coexisting desmoid tumor of the breast was made. The patient didn't receive any other treatment after the resection.

2 years later, evolution was marked by multiple lesions' recurrence on the lumpectomy's scar and overflowing inferior-medial and upper outer (**Figure 3**). In view of the extent of the recurrence, the patient underwent a second surgery to remove the recurrence of the mammary gland and flap reconstruction of the dorsal muscle.



**Figure 3.** Breast desmoid tumor's recurrence 2 years after surgical treatment.

### 3. Discussion

Our patient presented a Gardner's syndrome (GS). She initially presented with polyposis of the colon and had a subtotal colectomy, this was followed by the recurrence of digestive polyposis followed by rectal resection with ileoanal anastomosis. Also, She had a history of dental abnormalities and osteomas of the maxilla confirming the diagnosis of GS.

Gardner's syndrome is a rare autosomal dominant inherited disorder reported initially by Gardner in 1951 and characterized by the triad of colonic polyposis, multiple osteomas and mesenchymal tumors of the skin and soft tissues (epidermoid cysts, lipomas, ...) [4]. The syndrome may present at any age. Symptoms are most of the time evident by the 20<sup>th</sup> year of age; this is the first case in elderly woman. It represents a multisystemic disease with a variety of symptoms and often diagnosis is delayed. Usually, the cutaneous and bone abnormalities manifest earlier than polyposis [5]. However, in our case, the polyps were diagnosed five years before the extracolonic manifestations. The majority of tumors in patients with GS present intra-abdominally. The incidence of mammary desmoid tumors in GS is extremely rare 4%, the incidence of desmoid tumor of the breast is less than 0.2% of primary breast neoplasms [4]. Our patient with the history of recto-colic polyposis, presented at the age of 72 with an exophytic painless mass in the left breast. Histological evaluation of the surgical specimens leads to the diagnosis of breast fibromatosis.

Breast fibromatosis is a benign breast tumor, but its clinical, mammographic and ultrasound presentation is often very suspicious. The etiology of breast fibromatosis is most of the time unknown, the risk factors reported include silicone implants, surgical trauma as well as in association with GS [1]. In this present case, the tumor is associated with Gardner's syndrome. In this group, an alteration of the  $\beta$ -catenin pathway is considered to be the cause of the disease

[6]. Most of these cases were reported in young and fertile women with rare cases in male patients [7] [8]. Glazebrook KN *et al.* reported an extensive study of 125 cases of mammary fibromatosis (114 women and 11 men). Patient age ranged from 22 to 49 years [1]. To our knowledge, this is the first case report in the literature of Gardner's syndrome associated with a fibromatous tumor of the breast in the elderly woman.

Clinically, mammary fibromatosis is a firm, painless, unilateral nodule, poorly circumscribed, of variable size, more often peripheral than peri-areolar, sometimes associated with a cutaneous retraction [1]. This lesion can be painful especially when it infiltrates the deep planes. Our patient showed no skin retraction. The images observed on the mammogram can mimic a malignant lesion. Echography most often finds an irregular hypoechoic masse. Computed tomography and magnetic resonance imaging have the interest to show chest wall involvement, important for surgical planning [2] [9].

Large core biopsy is not always successful in the differential diagnosis of a tumor of mesenchymal origin [11]. A diagnosis can be made from the microscopic exam after surgery. Macroscopically, fibromatosis may be poorly circumscribed, firm, white-gray lesion of variable size. Histological examination usually reveals the presence of bundles of long sweeping and characteristic spindle cells with regular nuclei surrounded by varying amounts of collagen [7] [12] [13]. The immunohistochemistry exam shows positivity for  $\beta$ -catenin in 70% of cases [7].

Mammary fibromatosis is a locally aggressive tumor with no metastatic potential. The risk of local recurrence explains the wide nature of the surgery, which always consists of wide fibromatosis excision with clear margins, followed by biannual local monitoring because of the high risk of recurrence. If the margins are invaded, surgical revision is indicated. The literature reports recurrence rates of 5% to 25%, most of them within three to six years after excision [1] [10]. In the present case, the follow-up at 2 years showed the recurrence of the lesion. A mastectomy followed by reconstruction can be proposed for forms with multiple recurrences. In the case of parietal invasion, the surgery can be extremely dilapidating up to take the breast, the pectoral muscles, the chest wall and the parietal pleura [14] [15].

Various adjuvant and complementary treatments to surgery (radiotherapy, hormonal therapy, anti-inflammatory, cytotoxic) have been evaluated, allowing in rare cases partial or complete responses [3] [16] [17] [18] [19] [20]. In the case of desmoid tumors of the breast associated with Gardner's syndrome, hormone receptors are in general negative; hence, our patient was not offered hormonal therapy. Ishizuka *et al* reported a positive response with Tamoxifen even in estrogen receptor-negative breast desmoid tumors, this may be explained by apoptosis phenomena induced by transforming growth factor  $\beta$ 1 [19]. Radiation therapy has been shown to improve local control of desmoid tumor when used in combination with surgery [3].

## 4. Conclusion

Desmoid tumors of the breast are rare, infiltrating, and non-metastatic but have a high tendency to recur. The clinical expression is often nonspecific and mimics malignancy. Breast imagines are nonspecific and cannot distinguish between benign lesion and malignancy. Ultrasound scan and MRI are helpful for determining tumor extent and chest wall involvement. The treatment remains controversial because of the low incidence. Due to the high risk of recurrence, a wide local excision with clear margins is recommended. Adjuvant ant hormonal, chemotherapy and radiation therapy are controversial.

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## Consent for Publication

Written informed consent was obtained from the patient. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

## Availability of Data and Materials

The authors can confirm that all relevant data are included in the article.

## Competing Interests

The authors declare that they have no competing interests.

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## Authors' Contributions

MO conceived of the idea for the case report and participated in the writing of the article. AK, SZ, wrote the first draft of the manuscript. All authors read and approved the final manuscript.

## References

- [1] Glazebrook, K.N. and Reynolds, C.A. (2009) Mammary Fibromatosis. *American Journal of Roentgenology*, **193**, 856-860. <https://doi.org/10.2214/AJR.08.1892>
- [2] Raphael, K., Rogeria, M., Véronique, B., Sandrine, E., Hélène, B., Yonathan, B., Marina, M., Adriana, L., Jean-Marc, G. and Pascal, C. (2012) Fibromatose Mammaire: Corrélations Anatomo-Radio-Cliniques et revue de la littérature. *Imagerie de la Femme*, **22**, 100-109. <https://doi.org/10.1016/j.femme.2012.04.006>
- [3] Rammohan, A. and Wood, J.J. (2012) Desmoid Tumour of the Breast as a Manifestation of Gardner's Syndrome. *International Journal of Surgery Case Reports*, **3**, 139-142. <https://doi.org/10.1016/j.ijscr.2012.01.004>

- [4] Fotiadis, C., Tsekouras, D.K., Antonakis, P., Sfiniadakis, J., Genetzakis, M. and Zografos, G.C. (2005) Gardner's Syndrome: A Case Report and Review of the Literature. *World Journal of Gastroenterology*, **11**, 5408-5411. <https://doi.org/10.3748/wjg.v11.i34.5408>
- [5] Koh, K.-J., Park, H.-N. and Kim, K.-A. (2016) Gardner Syndrome Associated with Multiple Osteomas, Intestinal Polyposis, and Epidermoid. *Imaging Science in Dentistry*, **46**, 267-272. <https://doi.org/10.5624/isd.2016.46.4.267>
- [6] Gómez García, E.B. and Knoers, N.V. (2009) Gardner's Syndrome (Familial Adenomatous Polyposis): A Cilia-Related Disorder. *The Lancet Oncology*, **10**, 727-735. [https://doi.org/10.1016/S1470-2045\(09\)70167-6](https://doi.org/10.1016/S1470-2045(09)70167-6)
- [7] Abdelwahab, K., Hamdy, O., Zaky, M., Megahed, N., Elbalka, S., Elmetwally, M. and Denewer, A. (2017) Breast Fibromatosis, an Unusual Breast Disease. *Journal of Surgical Case Reports*, **21**, rjx248. <https://doi.org/10.1093/jscr/rjx248>
- [8] Smarrito, S. and Salmon, R. (2005) Desmoid Tumor in a Male Breast in the Context of Gardner's Syndrome: Case Report. *Annales de Chirurgie*, **130**, 40-43. <https://doi.org/10.1016/j.anchir.2004.10.011>
- [9] Perniciaro, C. (1995) Gardner's Syndrome. *Dermatologic Clinics*, **13**, 51-56. [https://doi.org/10.1016/S0733-8635\(18\)30106-2](https://doi.org/10.1016/S0733-8635(18)30106-2)
- [10] Rakha, E.A., Badve, S., Eusebi, V., Reis-Filho, J.S., Fox, S.B., Dabbs, D.J., Decker, T., Hodi, Z., Ichihara, S., Lee, A.H., Palacios, J., Richardson, A.L., Vincent-Salomon, A., Schmitt, F.C., Tan, P.H., Tse, G.M. and Ellis, I.O. (2016) Breast Lesions of Uncertain Malignant Nature and Limited Metastatic Potential: Proposals to Improve Their Recognition and Clinical Management. *Histopathology*, **68**, 45-56. <https://doi.org/10.1111/his.12861>
- [11] Lee, A.H.S. (2008) Recent Developments in the Histological Diagnosis of Spindle Cell Carcinoma, Fibromatosis, and Phyllodes Tumor of the Breast. *Histopathology*, **52**, 45-47. <https://doi.org/10.1111/j.1365-2559.2007.02893.x>
- [12] Erguvan-Dogan, B., Dempsey, P.J., Ayyar, G. and Gilcrease, M.Z. (2005) Primary Desmoid Tumor (Extra-Abdominal Fibromatosis) of the Breast. *American Journal of Roentgenology*, **185**, 488-489. <https://doi.org/10.2214/ajr.185.2.01850488>
- [13] Benej, R., Mečiarová, I. and Pohlodek, K. (2017) Desmoid-Type Fibromatosis of the Breast: A Report of 2 Cases. *Oncology Letters*, **14**, 1433-1438. <https://doi.org/10.3892/ol.2017.6337>
- [14] Wongmaneerung, P., Somwangprasert, A., Watcharachan, K. and Ditsatham, C. (2016) Bilateral Desmoid Tumor of the Breast: Case Series and Literature Review. *International Medical Case Reports Journal*, **22**, 247-51. <https://doi.org/10.2147/IMCRJ.S106325>
- [15] Shen, C., Zhou, Y. and Che, G. (2013) Management of a Female with Recurrence of Fibromatosis of the Chest Wall Adjacent to the Breast: A Case Report. *Journal of Cardiothoracic Surgery*, **8**, 41. <https://doi.org/10.1186/1749-8090-8-41>
- [16] Mendenhall, W.M., Zlotecki, R.A., Morris, C.G., Hochwald, S.N. and Scarborough, M.T. (2005) Aggressive Fibromatosis. *American Journal of Clinical Oncology*, **28**, 211-215. <https://doi.org/10.1097/01.coc.0000144817.78549.53>
- [17] Privette, A., Fenton, S.J., Mone, M.C., Kennedy, A.M. and Nelson, E.W. (2005) Desmoid Tumor: A Case of Mistaken Identity. *The Breast Journal*, **11**, 60-64. <https://doi.org/10.1111/j.1075-122X.2005.21679.x>
- [18] Ballo, M.T., Zagars, G.K., Pollack, A., Pisters, P.W. and Pollack, R.A. (1999) Desmoid Tumor: Prognostic Factors and Outcome after Surgery, Radiation Therapy, or Combined Surgery and Radiation Therapy. *Journal of Clinical Oncology*, **17**,

158-167. <https://doi.org/10.1200/JCO.1999.17.1.158>

- [19] Bhat, D., Wear, V., Weisenberg, E. and Alvarado, R. (2016) Desmoid-Type Fibromatosis of the Breast: A Case Report. *Breast Disease*, **36**, 149-152. <https://doi.org/10.3233/BD-160227>
- [20] Ishizuka, M., Hatori, M., Dohi, O., Suzuki, T. and Miki, Y. (2006) Expression Profiles of Sex Steroid Receptors in Desmoid Tumours. *The Tohoku Journal of Experimental Medicine*, **210**, 189-198. <https://doi.org/10.1620/tjem.210.189>

### List of Abbreviations

GS: Gardner's syndrome

DT: Desmoid tumor