



Case report

Breast neurofibroma: A case report

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ABSTRACT

Introduction: Neurofibromatosis type 1 or Von Recklinghausen disease is a rare autosomal dominant hereditary disease with total penetrance. It is characterized by an extreme clinical variability that is also found within the same family.

Presentation of case: We report a case of neurofibroma exceptional by its location in the breast in a 40-year-old woman with Von Recklinghausen disease, discovered by a breast nodule on self-examination. A biopsy-exeresis had concluded on the anatomopathological examination to the diagnosis of neurofibroma.

Discussion: Breast involvement in neurofibromatosis is extremely rare and possible. Rapid and adequate management of patients with Von Recklinghausen disease is essential in order to make the diagnosis early and to institute appropriate treatment as soon as possible, given the risk of possible malignant transformation.

Conclusion: Diagnosis is essentially based on anatomopathological study. This confirms the diagnosis and rules out possible associated malignancy.

1. Introduction

Neurofibromatosis type 1 or von Recklinghausen disease belongs to the group of phacomatoses. It is an autosomal dominant hereditary disease of variable penetrance, with neurocutaneous manifestation but also multi-visceral and predisposing to benign and malignant tumors [1]. Breast localization of neurofibromatosis is exceptional [2].

We report here a new case of a breast neurofibroma in a 40-year-old woman. It occurred in the context of neurofibromatosis 1 (NF1) or von Recklinghausen disease. Following this observation and through a review of the literature, we report the epidemiological, diagnostic and therapeutic particularities of this rare entity. All our work has been reported in line with the SCARE criteria and guidelines [12].

2. Presentation of case

A 40 year old patient, married, without any particular pathological antecedent. She consulted for a breast nodule which was discovered by self-examination Palpation of a nodule. The clinical examination revealed diffuse café au lait spots all over the body. Breast examination showed café au lait spots (Fig. 1), with palpation of a nodule located in the superior-internal quadrant of the left breast, measuring 20 × 10 mm, well bounded, firm, painless, mobile in relation to the superficial and

deep planes, without inflammatory skin signs. The axillary areas were free.

A mammogram showed very dense breasts. The skin covering was thin and regular. There was no architectural deformation of the mammary gland (Fig. 2). Breast ultrasound showed two anechoic, oval, well-limited formations, one located in the left superomedial quadrant measuring 20 × 10 × 6 mm, the second measuring 6 mm and located in the left inferolateral quadrant (Fig. 3). Excision of the nodule with extemporaneous examination was performed by our team. Macroscopic examination was performed on a breast lumpectomy specimen weighing 5 g and measuring 20 × 10 × 15 mm. The section showed a well limited nodule, firm in consistency, myxoid in appearance, measuring 20 × 10 × 5 mm. Examination of frozen, paraffin-embedded sections showed diffuse fusocellular proliferation within a loose fibrillar myxoid stroma interspersed with a few mononuclear inflammatory elements. The tumor cells were elongated, with a nonatypical, nonmitotic wavy nucleus. The excisional borders were healthy. An immunohistochemical study showed diffuse and intense expression of PS100 by the tumor cells.

The postoperative course was simple and the patient was compliant with the treatment. The diagnosis of diffuse neurofibroma was retained. After 12 months, no recurrence was observed.

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Fig. 1. Physical examination of mammary glands showing café-au-lait spots.

3. Discussion

Neurofibromatosis type 1 (NF1) or Von Recklinghausen disease, first described in 1882, is the most common of all neurofibromatoses [3,4]. It is inherited in an autosomal dominant fashion [8]. The NF1 gene is located on chromosome 17 in the 17q11.2 region and encodes neurofibromin, a protein involved in the control of cell proliferation and differentiation that inhibits the p21 ras activation pathway [1]. Its incidence is approximately one in 3000 to 4000 births without gender predominance [6]. NF1 is a progressive multisystem disease that mainly affects the skin, eyes, central and peripheral nervous system, skeleton and vessels. The lesions observed may be tumors, hamartomas or dysplasias [7]. Affected patients have a higher lifetime risk of developing a malignant tumor compared to the general population [6].

The clinical presentation is highly variable. The diagnosis of NF1 is clinico-radiological [7]. It is based on the criteria of the National Institute of Health (1987).

Neurofibroma in the breast is extremely rare, especially outside the context of neurofibromatosis type 1 [8]. When neurofibromatosis occurs in the trunk, breast examination becomes difficult and is often ignored [9]. Clinically, it is asymptomatic. It is most often a painless nodule, gradually increasing in size. Mammography is a very important examination in these patients. It usually shows the characteristics of a benign

lesion by showing a homogeneous circumscribed mass with a regular border, without calcifications. The adjacent mammary parenchyma does not show any architectural distortion, as is the case in our patient. Ultrasound showed a hypoechoic and heterogeneous mass [10]. Mammary neurofibroma is a benign tumor arising along the peripheral nerve sheaths. It is variable in shape and size. It is heterogeneous, composed mainly of schwann cells (60–80 %) associated with neurons, fibroblasts, mast cells and other cells [11]. Macroscopically, the lesion is usually well limited, rarely encapsulated, and firm in consistency. It may be polypoid or fusiform. In most cases, the size varies between one and two centimeters. However, it may manifest as a large volume. The color is typically grayish white, sometimes brown [10]. Histologically, the tumor has the same characteristics as a tumor located in another site. The tumor proliferation is composed of intersecting bundles of Schwannian differentiated cells, spindle-shaped with dark wavy nuclei, mixed with bands of collagen and patches of mucoid material [3]. The cells are free of nuclear atypia or mitosis. Any presence of mitotic activity should raise suspicion of malignancy, especially in the context of neurofibromatosis. The stroma may contain mast cells, lymphocytes, or xanthelast cells [3–8]. The immunohistochemical profile is quite specific. It allows to confirm the histological diagnosis by showing an expression of PS100 by the tumor cells, as in the case of our observation. While epithelial markers are usually negative [4–11]. Neurofibroma can

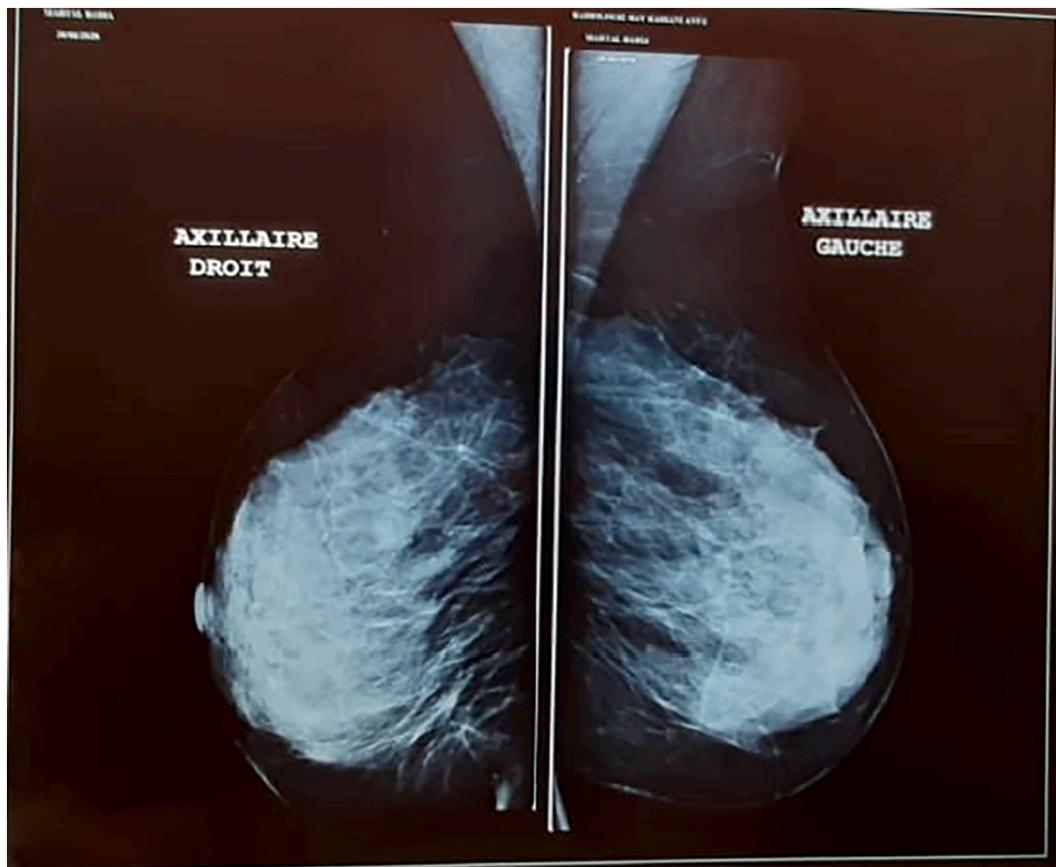


Fig. 2. Mammography view showing dense breasts. The skin is thin and even.

be confused with other tumors. First, schwannoma, an encapsulated tumor composed almost exclusively of Schwann cells. The distinction can be difficult. The S100 protein does not allow to make the distinction with certainty. The expression of the latter is lower in neurofibroma. A second differential diagnosis is that of a malignant tumor of the peripheral nerves, except that the cell density is higher with the presence of cellular abnormalities and mitoses. Diagnostic confirmation is required before initiating any therapy [4,5].

The treatment of choice for neurofibromas of the breast is complete surgical removal [6–10]. The risk of recurrence is low, provided that the surgical removal is complete. The literature reports a risk of sarcomatous transformation with an incidence varying from 2 to 6 % [10]. Patients with neurofibromatosis type 1 are considered to be at high risk of developing breast cancer. This risk is estimated at 8.4 % at the age of 50 years [5]. Invasive ductal carcinoma is the most common histological type reported in several series [4,5]. Only one case of anaplastic carcinoma of the breast has been reported. Also, two cases of Paget's disease have been reported in association with neurofibromatosis type 1 [4]. Physicians must be absolutely aware of this risk of malignancy since the management of patients will be more complex and the prognosis totally different [5,9]. The radiologist must be aware of this risk of association or carcinomatous transformation of the neurofibroma of the breast when interpreting breast imaging. The pathologist should examine the specimens received correctly, taking multiple specimens if necessary to rule out an associated malignancy.

4. Conclusion

Von Recklinghausen's disease is an uncommon genetic disorder, with a rarely reported mammary localization. The clinical examination can be difficult due to the presence of multiple cutaneous neurofibromas.

The diagnosis is histological. Complete excision with negative margins remains the treatment of choice. However, the evolution remains marked by the risk of malignant transformation or of developing breast cancer, which requires special surveillance.

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I declare on my honor that the ethical approval has been exempted by my establishment.

Consent

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Author contribution

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Fig. 3. Ultrasound of left breast demonstrating two oblong, circumscribed masses, the first in the supero-internal quadrant measuring $20 \times 10 \times 6$ mm, the second in the infero-internal quadrant measuring 6 mm.

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Registration of research studies

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Declaration of competing interest

The authors declare having no conflicts of interest for this article.

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