

Neurofibroma of the breast revealing a Von Recklinghausen disease: a case report

Rezgani Mona¹, Adouni Olfa², Hadiji Achref¹, Bettaieb Ilhem², Goucha Ahlem², Ben Abdallah Fakher², Jaidane Olfa¹, Ghaleb Montassar¹, Bouzaiène Hatem¹, Rahal Khaled¹, Ben Hassouna Jamel¹, and Gamoudi Amor²

¹Service de chirurgie carcinologique, Institut Salah Azaiz, Faculté de médecine de Tunis, Université Tunis El Manar, Tunisia

²Service d'anatomie pathologique, Institut Salah Azaiz, Faculté de médecine de Tunis, Université Tunis El Manar, Tunisia

Copyright © 2018 ISSR Journals. This is an open access article distributed under the **Creative Commons Attribution License**, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

ABSTRACT: Neurofibromatosis type 1 is a genetic disease characterized by changes in skin pigmentation and the growth of tumors along nerves in the skin, brain, and other parts of the body. Neurofibromas are the most common benign tumours of Neurofibromatosis type 1 developing at any point along a nerve with a neuro-cutaneous tropism. This genetic affection is an autosomal dominant disorder with complete penetrance and an important phenotypic variability. Disease manifestations occur at the childhood mostly before five years. Nevertheless, many de novo cases remain undiagnosed well into adult life. Neurofibroma that occur in the breast and constitute a revealing signs of the disease at adulthood are extremely rare. We report a case of a 30-year-old woman presenting a neurofibroma of the breast revealing Neurofibromatosis type 1.

KEYWORDS: Neurofibroma, breast, Von Recklinghausen.

1 INTRODUCTION

Neurofibromatosis type 1 (NF1), also known as Von Recklinghausen disease, is one of the most common genetic diseases in humans, affecting one case in 3,000 births. (A) (1). It's a multisystem disease but with a neuro-cutaneous tropism. This affection is characterized by changes in skin pigmentation (café-au-lait spots, skin fold freckling and Lisch nodules) and the growth of tumors along nerves in the skin, brain, and other parts of the body. Neurofibromas, which are the most common benign tumours of NF1, develop at any point along a nerve. Their presence of in the skin is more common than the deeper soft tissues. This autosomal dominant disorder with complete penetrance is characterized by an important phenotypic variability. Therefore we noted many clinical forms of benign to severe. Disease manifestations occur at the childhood mostly before five years. Nevertheless, many de novo cases remain undiagnosed well into adult life. Neurofibroma that occur in the breast and constitute a revealing signs of the disease at adulthood are extremely rare(2).

We report a case of a 30-year-old woman presenting a neurofibroma of the breast revealing NF1.

2 CASE REPORT

This is a case of a 30-year-old woman with no medical history. She presented with a left breast lump evolving for a year.

The clinical examination revealed multiple café-au-lait spots and a left breast tumour measuring 3 cm in the upper-outer quadrant. The mass was well limited and mobile. No other signs were found.

The ultrasound (US) demonstrated a 3 cm well-defined, hypoechoic solid mass suggesting fibroadenoma. The mammography was performed and showed the well defined mass. The patient had a lumpectomy.

Histopathologic examination of the mass showed benign tumour structure nonencapsulated but well circumscribed (Fig. 1), measuring 3cm composed of interlacing bundles of elongated cells with wavy nuclei in the breast tissue areas constituted of lobuli (Fig. 2). Several small nerves fibres were also present. The tumour cells were set in a fibromyxoid background (Fig. 3).

Mast cells were present. Mitotic activity was low. Immunohistochemical examination revealed that tumor cells were S100 protein positive. Actine and CD 34 were negative. The diagnosis of neurofibroma was made on the basis of the histopathological and immunohistochemical findings.

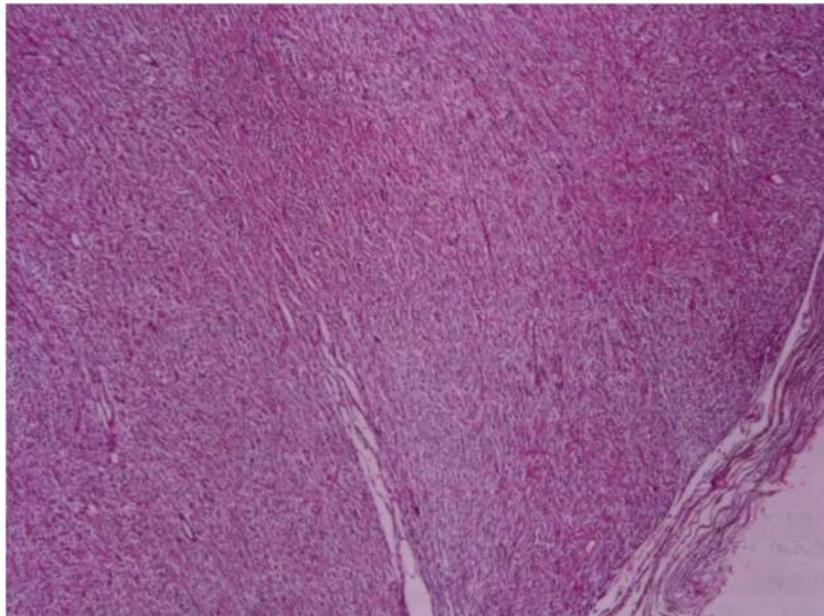


Fig. 1. Benign tumor non-encapsulated but well circumscribed

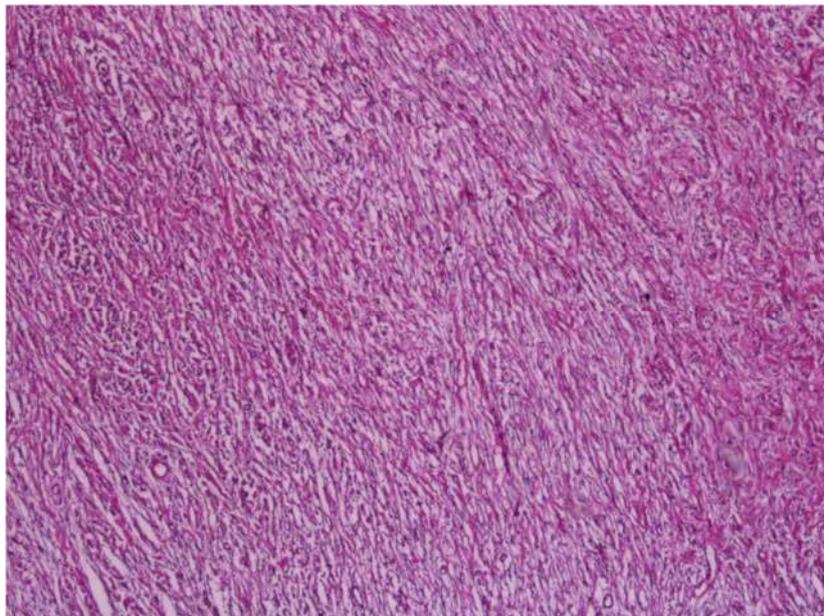


Fig. 2. Tumour composed of interlacing bundles of elongated cells

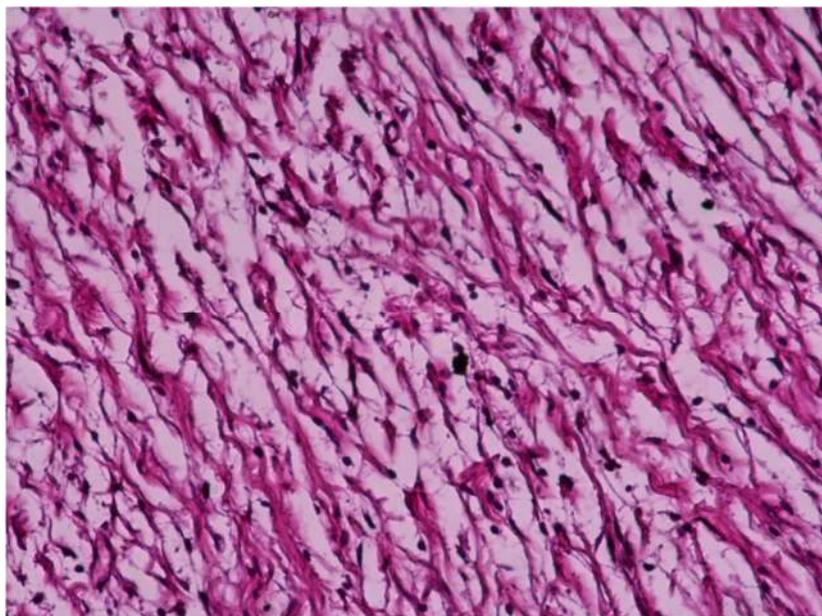


Fig. 3. *Tumour cells, with wavy nuclei, were set in a fibromyxoid background*

3 DISCUSSION

Neurofibromas are common benign tumours of the skin. They are nonencapsulated tumours of the nerve sheath of peripheral nerves and usually occur in groups.(3)

They were first described by Smith in 1849 and later by Von Recklinghausen in 1882. Because of the latter's major contribution, the syndrome of multiple neurofibromatosis bears his name.

The NF1 gene is located on chromosome 17 in the 17q11.2 region. Penetrance close to 100% at the age of five and de novo mutations account for about half of cases.(1)

Diagnostic criteria for NF1 are: six or more café-au-lait spots, two or more neurofibromas or one or more plexiform neurofibromas, freckling in the axilla or groins, optic glioma, two or more Lisch nodules, sphenoid wing dysplasia or thinning of a long bone cortex with or without pseudoarthrosis, and a first-degree relative who meets the above criteria for NF1. Two or more of the criteria are required for diagnosis. (4) In our case, the presence of multiples café-au-lait and a breast neurofibroma made the diagnosis of NF1.

Neurofibromas are the most common benign tumours of NF1. They are characteristically distributed over the trunk, head and neck area. Occurring in the breast is very rare, and in such cases they are most common in the areolar area(5). In our case it was the palpable mass that motivated the consultation and permitted to reveal the disease. In male patient, gynecomastia may be observed.(2)

Radiologically, neurofibromas usually appear as oval or round lesions with circumscribed margins on both mammography and US. On US, they appear as hypoechoic lesions with posterior acoustic enhancement, resembling a cyst, which may cause misdiagnosis.(6)

In our case, the mass was thought to be a fibroadenoma, that's way our patient didn't have further examination beside US.

On magnetic resonance imaging (MRI), they demonstrate high signal intensity on T2-weighted images, especially if the tumour has a myxoid matrix. They may demonstrate either non- enhancement or gradual enhancement after contrast material injection.(6)

Macroscopically, neurofibromas are typically white-grey, well circumscribed and not encapsulated tumours. They vary in size and shape but most range between 1 and 2 cm. However, a huge 20cm neurofibroma of the breast has also been reported. Their diagnosis ultimately depends on histological standard examination.(2)

They are distinct from schwannomas or neurilemmomas, which are also tumours of the peripheral nerve sheaths. They contain interlacing bundles of elongated cells with wavy, dark staining nuclei and slender cytoplasmic processes. These cells are arranged closely and are separated by small amounts of mucous material.

The primary differential diagnosis for this tumour is a neurilemmoma. However, fibroadenoma, phyllodes tumour, malignant peripheral nerve sheath tumour and myofibroblastoma should all be considered.

Some cancers are regularly observed during NF1 such as pheochromocytoma, neurofibrosarcoma, myelogenous leukaemia, optic glioma, malignant Schwannoma and rhabdomyosarcoma(7). Few cases of breast cancer with neurofibromatosis have been reported. The first report of an association between NF1 and breast cancer was published in 1972.(8)(9)

Neurofibromas rarely grow rapidly; such growth can suggest malignant transformation. The incidence of sarcomatous changes of neurofibromas varies from 2% to 16% (10).

Treatment of breast neurofibromas is carried out through surgical excision. However, despite a complete resection, recurrences are frequent. The risk of malignant transformation of these neurofibromas is estimated between 2 and 29% according to Enzinger(5). That's why a complement of excision (in a case of incomplete surgery) and long-term surveillance are imperatives.

4 CONCLUSION

Neurofibromas occurring in the breast are very rare. However, these must be taken into account since they can be revealing signs of the disease. Imagery certainly has an important diagnosis role. MRI remains the favourite diagnosis means to highlight neurofibromas in the breast and to detect a possible malignant transformation. Diagnosis is based on the standard histological and immunohistochemical study especially in atypical forms.

REFERENCES

- [1] Pinson S, Wolkenstein P. La neurofibromatose 1 (NF1) ou maladie de Von Recklinghausen. *Rev Med Interne*. 2005;26(3):196–215.
- [2] El Ounani F, Alami D, Dafiri R. Gynaecomastia revealing neurofibromatosis type 1 in a young child. *Eur J Radiol Extra*. 2009;69(3):2008–10.
- [3] Sherman JE, Smith JW. Neurofibromas of the breast and nipple-areolar area. *Ann Plast Surg*. 1981 Oct;7(4):302–7.
- [4] Murat A, Kansiz F, Kabakus N, Kazez A, Ozercan R. Neurofibroma of the breast in a boy with neurofibromatosis type 1. *Clin Imaging*. 2004;28(6):415–7.
- [5] Lamalmi N, Benkiran L, Gamra L, Regragui A, Amrani M, Belabbas M. Neurofibrome géant du sein révélateur d'une maladie de Von Recklinghausen : À propos d'un cas. *Rev Française des Lab*. 2002;2002(347):71–3.
- [6] Lee H, Kim HJ, Yang B, Lee SM, Jeong JY. Neurofibroma of the Breast Detected on Positron Emission Tomography-CT in a Patient with Neurofibromatosis Type 1 : A Case Report 제1형 신경섬유종증 환자에서 양전자방출단층촬영으로 발견된 유방의 신경섬유종 : 증례 보고. 2016;74(2):128–31.
- [7] Seminog OO, Goldacre MJ. Risk of benign tumours of nervous system, and of malignant neoplasms, in people with neurofibromatosis: population-based record-linkage study. *Br J Cancer*. Nature Publishing Group; 2013;108(1):193–8.
- [8] Brasfield RD, Das Gupta TK. Von Recklinghausen's disease: a clinicopathological study. *Ann Surg*. 1972 Jan;175(1):86–104.
- [9] Da Silva AV, Rodrigues FR, Pureza M, Lopes VGS, Cunha KS. Breast cancer and neurofibromatosis type 1: a diagnostic challenge in patients with a high number of neurofibromas. *BMC Cancer*. BioMed Central; 2015 Mar 26;15:183.
- [10] Millman SL, Mercado CL. An unusual presentation of neurofibromatosis of the breast. *Breast J*. 10(1):45–7.