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Case Report

Multiple Breast Neurofibromas in Neurofibromatosis Type 1: Case Report

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Abstract

Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder associated with a wide spectrum of benign and malignant tumors, while breast involvement remains rare and poorly described. We report the case of a 66-year-old woman with long-standing NF1 who underwent routine breast cancer screening and was found to have multiple intramammary and cutaneous neurofibromas. Ultrasound revealed well-circumscribed, homogeneous oval masses with posterior acoustic enhancement, and MRI confirmed numerous cutaneous and intramammary lesions that were T1 hypointense, heterogeneously T2 hyperintense, and showed variable gadolinium enhancement without features of malignant transformation. This case highlights the uncommon manifestation of breast neurofibromas in NF1 and underscores the importance of recognizing their characteristic imaging features, given the increased risk of breast carcinoma and malignant peripheral nerve sheath tumors in this population, warranting careful and ongoing clinical and radiologic surveillance.

Keywords: *Neurofibromatosis Type 1 (NF1); T1 Hypointense; T2 Hyperintense*

Introduction

Neurofibromatosis type 1 (NF1), or von Recklinghausen disease, is an autosomal dominant disorder with a prevalence of approximately 1 in 3,000 live births worldwide. It is characterized by cutaneous, neurologic, and skeletal manifestations and carries an increased lifetime risk of both benign and malignant tumors. Although cutaneous neurofibromas are a hallmark of the disease, breast involvement is rare and poorly documented. Recognition of breast neurofibromas is clinically important because patients with NF1 are predisposed to malignant peripheral nerve sheath tumors and early-onset breast carcinoma. We report a case of multiple breast neurofibromas detected during routine breast cancer screening in a patient with NF1, highlighting the imaging features and the need for vigilant surveillance.

Case Report

We report the case of a 66-year-old female patient with a long-standing history of Neurofibromatosis Type 1 (NF1), known for several years. Clinical examination revealed multiple cutaneous neurofibromas (Figure 1B) as well as numerous café-au-lait macules distributed across the entire body (Figure 1C). As part of a routine breast cancer screening program, breast ultrasound was performed. It demonstrated several intra-mammary masses with well-circumscribed oval contours, homogeneous echotexture, and posterior acoustic

enhancement. In addition, superficial nodules were identified within the subcutaneous tissue, suggestive of cutaneous neurofibromas involving the breast. A complementary breast MRI confirmed the presence of multiple cutaneous and intra-mammary neurofibromas. These lesions appeared hypointense on T1-weighted sequences, heterogeneously hyperintense on T2-weighted sequences, and showed variable enhancement after gadolinium administration, with no suspicious features of malignant transformation. Taken together, these findings support the diagnosis of multiple breast neurofibromas, a rare but documented manifestation in extensive forms of NF1. Given the potential risk of malignant transformation in NF1 patients, the case underscores the need for regular clinical and radiological follow-up.

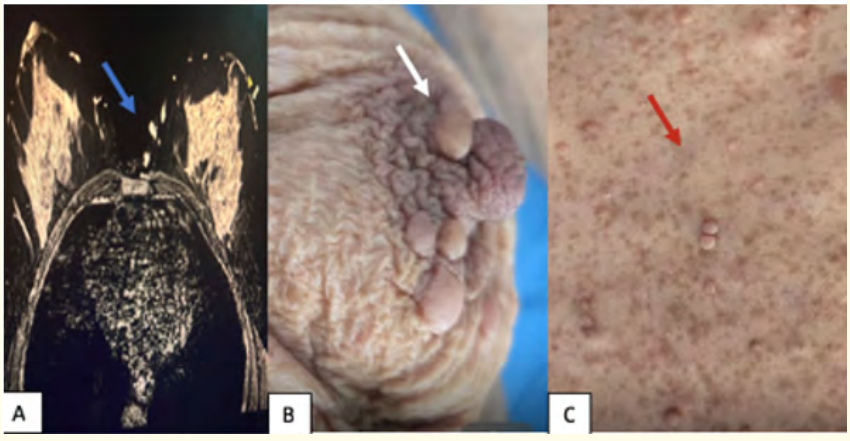

Figure 1: *Breast MRI, axial T2-weighted sequence, showing a left breast neurofibroma with high T2 signal intensity (blue arrow) as well as multiple subcutaneous nodules also displaying high T2 signal (A). Clinical aspect of cutaneous neurofibromas visible on the chest (white arrow) in a patient with neurofibromatosis type 1 (B). Presence of multiple café-au-lait macules characteristic of neurofibromatosis type 1 (red arrow) (C).*

Discussion

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is the most frequent of the phakomatoses, with an estimated incidence of 1 in 3,000 births worldwide [1]. It is an autosomal dominant disorder with complete penetrance but variable expressivity, and approximately 50% of cases result from *de novo* mutations [1]. The disorder is characterized by multisystem involvement, with manifestations affecting the skin, nervous system, eyes, skeleton, and, less commonly, visceral organs. The diagnosis of NF1 remains clinical, based on the criteria originally defined in 1988 and recently revised in 2021 [2]. The presence of multiple café-au-lait macules remains one of the most evocative findings. Depending on family history, the diagnosis requires either at least two criteria (Category A, no family history) or one criterion in the presence of an affected first-degree relative (Category B) [2]. From a molecular perspective, NF1 results from mutations in the NF1 gene located on chromosome 17q11.2. This gene encodes neurofibromin, a tumor suppressor protein that negatively regulates the RAS/MAPK signaling pathway, thereby controlling cell proliferation and survival [1,5]. Loss or reduction of neurofibromin function leads to constitutive RAS pathway activation, resulting in excessive proliferation of Schwann cells, fibroblasts, and melanocytes, which accounts for the development of neurofibromas, café-au-lait spots, and the increased predisposition to tumors [5]. Patients with NF1 have an increased risk of malignancies and disease-related complications, which significantly reduce life expectancy compared to the general population [3]. Therefore, regular clinical and radiological surveillance is recommended, and imaging plays a critical role not only in diagnosis but also in follow-up [4]. Cutaneous neurofibromas and café-au-lait macules are the most common manifestations, but involvement of the breast is uncommon and only sporadically reported in the literature. When present, breast neurofibromas are most often located in the periareolar region and usually manifest as multiple nodular lesions. From an imaging standpoint, breast neurofibromas show relatively characteristic features:

- On mammography, they appear as well-circumscribed, oval masses, sometimes with partially obscured margins due to their superficial location [7].
- On ultrasound, they present as oval, circumscribed, homogeneous hypoechoic masses located in the subcutaneous tissue, typically with posterior acoustic enhancement [7].
- On MRI, neurofibromas usually appear hypointense on T1-weighted sequences, heterogeneous on T2-weighted sequences, and show low or variable contrast enhancement after gadolinium injection [8].

The main differential diagnosis is fibroadenoma, particularly in young women, given the similar sonographic appearance [7]. However, the clinical background of NF1, multiplicity of lesions, and superficial location strongly support the diagnosis of neurofibroma. In male patients with NF1, breast involvement may present differently, most commonly as gynecomastia, multiple neurofibromas, or less frequently as pseudoangiomatous stromal hyperplasia [6,9]. Although benign in nature, neurofibromas in NF1 require careful follow-up because of the increased lifetime risk of malignant peripheral nerve sheath tumors (MPNSTs), as well as the higher incidence of breast carcinoma in NF1 patients, particularly in women under 50 years of age. This highlights the importance of early screening and close surveillance in this population.

Conclusion

Breast neurofibromas are an uncommon manifestation of NF1 and can mimic other benign breast lesions. Awareness of this entity is crucial for accurate diagnosis and appropriate management, particularly given the elevated risk of breast malignancy in women with NF1. This case highlights the importance of regular clinical and radiologic follow-up to enable early detection of malignant transformation.

Bibliography

1. Friedman JM. "Neurofibromatosis 1". In: GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle (1998).
2. Legius E., *et al*. "Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation". *Genetics in Medicine* 23.8 (2021): 1506-1513.
3. Evans DG., *et al*. "Mortality in neurofibromatosis 1: an assessment of the utility of hospital admissions data". *American Journal of Medical Genetics Part A* 155A.3 (2011): 572-577.
4. Stamatopoulos A., *et al*. "Imaging features of neurofibromatosis type 1: from head to toe". *Diagnostic and Interventional Imaging* 97.1 (2016): 59-72.
5. Cichowski K and Jacks T. "NF1 tumor suppressor gene function: narrowing the GAP". *Cell* 104.4 (2001): 593-604.
6. Jett K and Friedman JM. "Clinical and genetic aspects of neurofibromatosis 1". *Genetics in Medicine* 12.1 (2010): 1-11.
7. Soo MS., *et al*. "Neurofibromatosis involving the breast: mammographic and sonographic findings with pathologic correlation". *American Journal of Roentgenology* 169.2 (1997): 417-420.
8. Kransdorf MJ. "Benign soft-tissue tumors in a large referral population: distribution of specific diagnoses by age, sex, and location". *American Journal of Roentgenology* 164.2 (1995): 395-402.
9. Tan PH., *et al*. "Stromal pseudoangiomatous hyperplasia of the breast: a benign lesion mimicking angiosarcoma". *Archives of Pathology and Laboratory Medicine* 129.3 (2005): 348-352.

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