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Diffuse Neurofibroma of the Breast in an Adolescent Female: A Rare Case Report

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ABSTRACT

Neurofibroma of the breast is an extremely rare benign peripheral nerve sheath tumour, often associated with neurofibromatosis type 1 (NF1). It is even more uncommon in adolescents and when occurring as an isolated breast mass in the absence of systemic signs of NF1. This case report highlights an unusual presentation of a large solitary breast neurofibroma in an 18-year-old Nigerian female, emphasising diagnostic challenges and the importance of histopathologic confirmation. An 18-year-old female presented with a painless right breast lump that had rapidly enlarged over six months. There was no family history or other systemic signs of NF1. Examination revealed a large, multilobulated, mobile breast mass with visible surface veins. Breast ultrasonography showed a multilobulated mass with posterior acoustic enhancement and increased Doppler flow. A core needle biopsy revealed a diagnosis of diffuse neurofibroma. She underwent wide local excision with uneventful postoperative recovery. Isolated breast neurofibroma, particularly in adolescents without NF1, is rare and may mimic more common fibroepithelial tumours such as phyllodes tumours. Histopathologic examination with immunohistochemistry remains the diagnostic cornerstone. This report underscores the need for high clinical suspicion and contributes to the limited African literature on this rare entity.

Keywords: *Adolescent; Case Report; Breast Tumour; Breast Neurofibroma; Diffuse Neurofibroma; Nigeria; Phyllodes Mimic.*

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Introduction

Neurofibromas are benign peripheral nerve sheath tumours composed of Schwann cells, fibroblasts, perineural cells, and mast cells. They are commonly associated with neurofibromatosis type 1 (NF1) but may also occur as solitary lesions in individuals without clinical features of the syndrome. Breast involvement is exceptionally rare, particularly in adolescents, and breast neurofibromas may clinically and radiologically mimic more common lesions such as fibroadenomas and phyllodes tumours.

Solitary neurofibromas of the breast, particularly the diffuse variant, are rarely reported in the literature. Reports from sub-Saharan Africa are particularly limited, with very few documented cases from Nigeria. The diagnosis may be challenging, especially in younger females, because these lesions can present as rapidly enlarging masses and may reach considerable dimensions, raising clinical suspicion of malignancy or other aggressive benign breast tumours, including phyllodes tumours and juvenile fibroadenomas {1-3}.

This case report describes an 18-year-old Nigerian female who presented with a large, rapidly enlarging breast mass that was initially considered clinically and radiologically to be a phyllodes tumour but was subsequently confirmed histologically as a diffuse neurofibroma.

The case contributes to the limited African literature on isolated breast neurofibromas and highlights the important clinical, radiological, histopathological, and diagnostic considerations associated with this rare entity.

Case Presentation

An 18-year-old premenopausal Nigerian female presented to the surgical outpatient department with a painless, progressively enlarging lump in her right breast, first noticed six months prior. The swelling started as a small, non-tender nodule but rapidly increased in size, causing significant breast asymmetry. She denied nipple discharge, skin changes, trauma, or systemic symptoms such as fever, weight loss, or fatigue. There was no history of similar masses elsewhere on her body. She had no chest, spinal, brain or abdominal symptoms, and no family history of breast disease or neurocutaneous syndromes.

On physical examination, she was a healthy-appearing young woman with normal vital signs. The right breast was significantly enlarged compared to the left, with visible superficial venous distention but intact overlying skin. Palpation revealed a multilobulated, non-tender, firm mass measuring approximately 18 × 14 cm. The mass was freely mobile within the breast tissue. There was no nipple retraction or discharge, and no skin ulceration. The left breast was normal. No ipsilateral or contralateral axillary lymphadenopathy was noted.

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Based on clinical features—a large, fast-growing, lobulated mass in a young female—a diagnosis of phyllodes tumour was considered. Breast ultrasonography revealed a multilobulated, solid mass with posterior acoustic enhancement and increased internal vascularity on Doppler interrogation, further supporting the provisional diagnosis.

Laboratory workup, including complete blood count, renal and liver function tests, and coagulation profile, was within normal limits. The patient subsequently underwent a core needle biopsy of the breast mass. Histologic analysis revealed a tumour composed of elongated spindle cells with wavy nuclei embedded in a collagenous and myxoid stroma. There was no atypia or mitotic activity. Immunohistochemistry showed strong positivity for S-100 protein, confirming the diagnosis of diffuse neurofibroma.

She was counselled extensively on the diagnosis and treatment options. Given the size and potential for local complications, wide local excision was planned and performed under general anaesthesia. The mass was completely excised, and postoperative recovery was uneventful. Final histopathology confirmed the core biopsy findings with clear surgical margins. The patient has remained well on follow-up up to 12 months and showed no evidence of recurrence.

Discussion

Neurofibromas are well-characterised peripheral nerve sheath tumours commonly associated with neurofibromatosis type 1 (NF1), an autosomal dominant disorder with multisystem involvement {4,5}. Solitary neurofibromas unrelated to NF1 are rare and typically occur in adults between 30 and 60 years of age. Breast involvement is exceedingly uncommon, while the diffuse type is even more rarely reported {6}.

The global literature contains only a limited number of reports of breast neurofibromas, particularly among adolescents. Solitary breast neurofibroma has been documented in a 14-year-old female, representing one of the youngest reported cases {7}. Another case involved a 27-year-old female in France, further highlighting the rarity of this lesion and the importance of histological confirmation {8}.

Neurofibroma of the breast is extremely rare. A review of soft tissue breast tumours from Ilorin, Nigeria, included neurofibroma among the reported soft tissue lesions {18}, further emphasising its exceptional rarity in both local and international literature.

In young females, benign breast lesions, particularly fibroadenomas, are common. However, rapidly enlarging breast masses warrant consideration of phyllodes tumours and, less commonly, sarcomas {9}. In the present case, the large size (18 × 14 cm), mobility, and lobulated configuration of the mass clinically favoured a diagnosis of phyllodes tumour.

Ultrasonography demonstrating a hypoechoic, lobulated, and vascular mass with posterior acoustic enhancement is non-specific but may suggest a fibroepithelial tumour {10}. Because of the considerable overlap in the clinical and radiological features of these lesions, tissue diagnosis is essential. Histopathological examination and immunohistochemical analysis are therefore critical for definitive diagnosis. Neurofibromas are typically composed of spindle-shaped Schwann cells arranged in interlacing bundles or whorls within a myxoid matrix. The nuclei are characteristically serpentine, while mitotic figures are uncommon. In the diffuse variant, the tumour cells infiltrate the surrounding tissue in a poorly circumscribed pattern {11}.

Immunohistochemically, strong S-100 protein expression supports Schwann cell differentiation and is an important diagnostic feature. CD34 and EMA may also be expressed in some lesions {12}. In contrast, phyllodes tumours demonstrate both epithelial and stromal components with characteristic leaf-like architecture, while fibroadenomas generally show a more uniform stromal proliferation.

The differential diagnoses in this case included phyllodes tumour, which is typically characterised by rapid growth, lobulated margins, and increased vascularity, but does not characteristically demonstrate S-100 protein expression. Juvenile Fibroadenoma, although common in adolescents, is generally smaller and rarely exceeds 5–7 cm in diameter. Low-grade peripheral nerve sheath tumours may share some histological features with neurofibromas but are more likely to demonstrate nuclear atypia. Myofibroblastoma and other spindle-cell tumours were also considered but can be distinguished through their characteristic morphological and immunohistochemical profiles.

Wide local excision with clear margins remains the treatment of choice for a localised breast neurofibroma. In the present case, complete excision resulted in satisfactory resolution with a good cosmetic outcome and no evidence of recurrence during follow-up. Malignant transformation is exceedingly uncommon in isolated neurofibromas but may occur in plexiform lesions, particularly in patients with NF1 {13}.

Postoperative surveillance remains important. Although there was no family history or clinical evidence of NF1 in this patient, the possibility of segmental or late-onset manifestations cannot be completely excluded. Annual clinical assessment, including appropriate dermatological surveillance, is therefore recommended {14}.

Published literature on breast neurofibromas and peripheral nerve sheath tumours from sub-Saharan Africa remains limited. A South African series of peripheral neurofibromas involving 30 patients did not report breast involvement {15}. Similarly, reports of NF1 with cutaneous manifestations from Calabar, Nigeria, did not identify breast involvement {16}. A histopathological review of paediatric soft tissue tumours from Jos, Nigeria, included neurofibromas, but no breast lesions were reported {17}. The scarcity of reported Nigerian cases may reflect the genuine rarity of breast neurofibromas or possible underdiagnosis related to limited access to immunohistochemistry and specialised expertise in peripheral nerve sheath tumours.

This case therefore represents a rare and well-documented occurrence of a histologically confirmed breast neurofibroma in an adolescent from Nigeria. It adds to the limited regional literature and highlights the diagnostic challenges posed by the clinical and radiological similarity between neurofibromas and more common breast tumours, particularly phyllodes tumours and fibroadenomas. The case also underscores the importance of histopathological examination and immunohistochemistry in establishing the diagnosis and guiding appropriate management.

Conclusion

Breast neurofibromas are uncommon tumours, especially in adolescents without neurofibromatosis type 1 (NF1). This report of an 18-year-old Nigerian female presenting with a large solitary diffuse neurofibroma underscores the diagnostic challenges involved and the critical role of histopathological and immunohistochemical evaluation.

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Although clinical and radiological findings may resemble more prevalent tumours such as phyllodes or fibroadenomas, biopsy remains the gold standard for definitive diagnosis. Wide local excision is the treatment of choice and all that is needed. This case contributes valuable data to the scarce African literature and emphasises the need for increased awareness and documentation of such rare breast tumours in young patients.

Patient Consent: Consent was obtained from the patient for publication of this case report and accompanying images.

Conflict of Interest: The authors declare no conflict of interest.

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