An Unusual Presentation of Neurofibromatosis with Breast Carcinoma


Abstract
We came across a rare case of a 58 yr old female who presented with a growing left breast mass since 2 and 1/2 months. On clinical examination a mass of 3 cm diameter was palpated close to the nipple, along with multiple neurofibromas on both breasts and trunk, and café au lait spots were seen. Patient underwent mastectomy and removal of three neurofibromas. Histologically breast mass was infiltrating duct carcinoma (T1N1M0). Considering that neurofibromatosis is genetic disorder, and its known association with sarcomas and leukaemias, its relation to carcinomas is rarely documented and we think intentional general exploration is required in all cases of neurofibromatosis.

Introduction
The protein product of NF-1 gene neurofibromin is a GTPase activating protein that facilitates conversion of active ‘ras’ to inactive ‘ras’. With loss of NF-1 gene ‘ras’ is trapped in an active continuous signal emitting state thereby causing dysregulation of signal transduction.¹

Individuals who inherit one mutant allele develop numerous benign neurofibromas. They are acknowledged to be at increased risk for malignancy. The overall risk of cancer was 2.7 times higher in NF-1 patients than the general population. The most frequent types of cancer were connective tissue and brain tumours.²

Case Report
58 yr female presented with chief complaints of lump in breast rapidly growing since 2 and ½ months. On clinical examination she had multiple cutaneous neurofibromas on breast, café au lait spots and a deep firm mass 3 cm in diameter on upper outer quadrant. Multiple neurofibromas were also seen on the other breast and trunk.

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A clinical diagnosis of multiple neurofibromas with suspected carcinoma breast was made and after FNAC report of breast carcinoma, patient was posted for modified radical mastectomy (MRM).

We received an MRM specimen with skin flap (nipple and areola) and axillary tail measuring 8 x 5 x 2 cm (Fig.1). Cut section showed a tumour (Fig. 2). Grossly all surgical margins were free of tumour. 12 lymph nodes were milked from axillary tail.

Histopathology sections from the tumour mass showed features of infiltrative ductal carcinoma (Fig. 3) with Bloom Richardson score- 8. 7 out of the 12 lymph nodes were involved by the tumour. The pigmented papules showed typical features of neurofibroma (Fig. 4).

Discussion
In patients with NF-1 there is a high incidence of neoplasms especially tumours of central and peripheral nervous system including optic gliomas, astrocytomas, meningiomas and schwannomas. Other less frequent malignant tumours such as neurofibrosarcomas, phaeochromocytomas and leukaemias may also occur.¹ Very few cases of NF-1 with epithelial malignancies especially breast have been reported.³ Studies suggest that loss of heterozygosity in the tumour supports the role of NF-1 gene in the aetiology in some cases of breast cancer.⁴
Multiple neurofibromas may obscure breast mass at palpation leading to delayed detection of cancer.  

Systemic and careful exploration is essential for patients with von Recklinghausen’s neurofibromatosis to detect breast cancer at an early stage.  

References  