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## Case Report

# Male breast cancer with neurofibromatosis- a rare presentation

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## ABSTRACT

Neurofibromatosis (NF-1) was formerly known as Von Recklinghausen's disease after the researcher (Friedrich Daniel Von Recklinghausen) who first documented the disorder. Patient with Neurofibromatosis type 1 have a higher risk of developing various type of cancers, especially tumors derived from embryo genic neural crest.

**Keywords:** Neurofibromatosis, CA breast, Epithelial malignancy, Mastectomy

## INTRODUCTION

Male breast cancer is a rare disease and accounts for less than 1% of all breast cancers.[1] Neurofibromatosis type 1 (NF1) or Von Recklinghausen disease is an autosomal dominant condition that affects 1in 3000 individuals.[2] NF1 characterized by café –au-lait spots and multiple neurofibromas, associated with various type of cancers, especially tumors derived from the embryogenic neural crest, including phemochromocytoma, leukaemia, gliomas, rhabdomyosarcomas, astrocytoma and neuro-fibrosarcoma.[3,4] Male Breast Cancer is a rare disease and accounts for less than 1% of all breast cancers.[5] Here we are reporting a case of male cancer with neuro-fibromatosis.

## CASE REPORT

A 38 year male with a lump in left breast since 6 months with multiple skin nodules all over body since childhood

### *HOPI*

Patient presented with a complaint of lump in left breast since 6 months with no history of any pain, no history of any discharge, any change color of areola, retraction of nipple. Multiple skin nodules all over body .No, history of weight loss, breast, ovary, prostate cancer in the family, any medical allergies or any other medical problems.

### *General examination*

Vitals were stable

### *Local examination*

Lump of size 2x3 cm in the upper inner quadrant of left breast, firm in consistency, non-tender and mobile, axillary lymph nodes were not palpable.

### *Systemic examination*

Systemic examination was within normal limits.

### *Laboratory examination*

Haematological, liver, renal function tests were normal. Ophthalmic examination was normal. FNAC s/o: epithelial malignancy.

Genetic analysis could not be conducted because of financial limitations. Patient underwent modified radical mastectomy and axillary lymph node dissection.

***Histopathological examination***

Infiltrating ductal carcinoma

***Immunohistochemistry***

Estrogen and progesterone receptors (ER, PR) were both positive and HER2 was positive, post-operative recovery was uneventful.

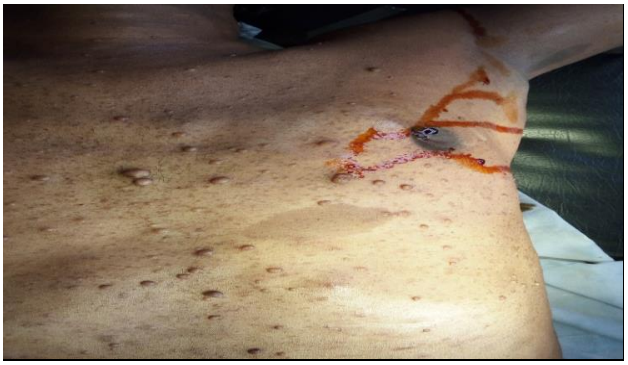

**Figure 1: External appearance of male cancer breast with multiple neurofibromatosis.**

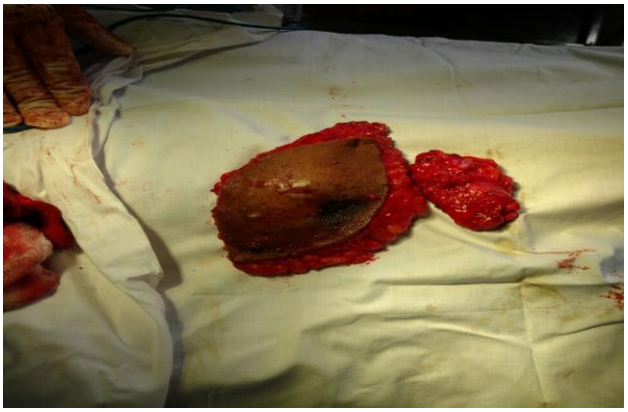

**Figure 2: Specimen of cancer breast with axillary LN dissection upto level 3.**

## DISCUSSION

NF1 is an autosomal dominant condition and has penetrance but wide variability in expression.[6] NF 1 gene is located in the peri-centomeric region of the long arm of chromosome 17 (which also houses the BRCA 1 gene). It regulates the conversion of the active Ras-GTP to inactive Ras-GDP. Ras is known as an essential component of signal transduction pathways that regulate growth, proliferation, differentiation, and apoptosis.

The conversion from the GTP- to the GDP- bound form is mediated by the intrinsic GTPase activity of Ras. The impairement of this hydrolytic reaction is associated with an increased risk of cancer.[4] Hence, it has potential role as a tumor suppressor gene.[7]

In, neurofibromatosis type 1, Skin manifestations can appear in early life with the development of more than five smooth surfaced café-au-lait spots over 5mm in diameter in prepubertal individuals and over 15 mm in diameter in post pubertal individuals, subcutaneous Neurofibroma, armpit or groin freckling and Lisch nodules, optic gliomas, scoliosis of spine, learning disabilities vision disorder, mental disabilities. It represents a risk factor for development of various malignancies, including female breast cancer.[8]

Association between NF1 and malignant tumors has been widely described, most common reported are with gliomas, malignant peripheral nerve sheath tumors, leukaemia, and rhabdomyosarcoma. Concerning the association between NF1 and breast cancer, only a few cases have been reported.[9,10]

About 28% of sporadic breast cancers are missing atleast one copy of NF 1 gene, either due to deletion or mutation.[11] The first cases describing the association of NF 1 with breast cancer were reported in the 1970's by Brasfield and Das Gupta. They described their experience with five patients, including one who had bilateral breast cancer.[12] Since, then many case reports have been published. Murayama et al reported 37 cases of breast cancer associated with NF1; most OF cases were diagnosed at an advanced stage and had invasive ductal carcinoma.[13]

In an earlier report by Nakamura et al, the authors noted that breast cancer5 affected young woman (<35 years old), in 18.5% of the cases which is relatively high when compared.[9] To the findings of other series of breast cancer not associated with NF 1, which reported a percentage of 6.7%.

In a study of Sharif et al, the main objective was to evaluate the risk of developing breast cancer among patients with NF 1.[14] A cohort of 304 women aged 20 years and above who were diagnosed with NF 1 was studied over a period of 30 years. The authors reached the conclusion that women with NF 1 had five times more chances of

Developing breast cancer when compared to the general population. Despite these clinical Findings, patients with NF 1 are still not stratified as high risk patients and current guidelines. Do not give specific considerations regarding any screening program for this category of patients.[15,16] Case report by Yamamoto in which a 33 year old woman had undergone Reconstructive surgery using the abdominal rectus muscle.[17]

## CONCLUSION

Few cases of male breast cancer associated with Neurofibromatosis type 1, so far woman are at higher risk of developing breast cancer. Genetic evaluation should

be done with patient of neurofibroatosis and proper approach should be kept in such cases.

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