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 embryonic 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 1 in 3000 individuals.² NF1 characterized by café au-lait spots and multiple neurofibromas, associated with various type of cancers, especially tumors derived from the embryonic neural crest, including phemochromocytoma, leukaemia, gliomas, rhabdomyosarcomas, astrocytoma and neurofibrosarcoma.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 neurofibromatosis.

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.

Associated with NF1 are still not stratified as high risk population. Despite these clinical associations 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 at least one copy of NF1 gene, either due to deletion or mutation.11 The first cases describing the association of NF1 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 NF1, 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 NF1.14 A cohort of 304 women aged 20 years and above who were diagnosed with NF1 was studied over a period of 30 years. The authors reached the conclusion that women with NF1 had five times more chances of developing breast cancer when compared to the general population. Despite these clinical findings, patients with NF1 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 women 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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REFERENCES
