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Bleeding Breast Tumor: Case of Neurofibromatosis Breast, Dilemma in Diagnosis

F. Shahizzat[1*], D. H. Ismail[1] and B. Anita[1]

[1]Breast and Endocrine Unit, Department of Surgery, Hospital Putrajaya, Malaysia.

Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Neurofibromatosis in breast swelling is a rare presentation. We encountered a female presented with odd presentations of bleeding breast swelling. Initial imaging shows appearances of haemangioma with a lesion based on Breast Imaging Reporting and Data System (BIRADS) BIRADS 4a. Initial biopsy shows a characteristic of Phyllodes tumor, however post-operative histopathological shows peripheral neural sheath tumor in favor neurofibroma.

Keywords: Neurofibromatosis; NF1; breast; breast cancer; bleeding breast tumor.

1. INTRODUCTION

Neurofibromatosis (NF) is a benign tumor that affects the neural sheath. There are two types of neurofibromatosis NF1 and NF2. The most common is Neurofibromatosis Type 1 (NF1). Its prevalence is 1 in 3000 birth. The disease is caused by a spontaneous mutation and can be due to an inherited mutation. Neurofibromas are very rarely found in the breast. The common type found in the breast is neurofibromatosis type 1. Malignant transformations in NF1 patient is up to 18.5% for woman who less than 35 years old.

2. CASE REPORT

A 55 old married multiparous Chinese female, presented with a right breast lump for more than

**Corresponding author: E-mail: shahizzatfahmi@gmail.com;*

30 years. She noted the right breast lump slowly increasing in size with thick tissue and skin darkening. The right breast larger compare to the left side with a history of spontaneous bleeding from the right breast multiple times since 2017. On further history no family history of similar illness, breast cancer, no history taking contraceptive pills or antithrombotic drugs.

On examinations show hyperpigmentation of the whole right breast and chest wall. The right breast appears larger and the nipple deviates to the left side. Soft firm mass with bluish discoloration felt over the right lower quadrant of the right breast. The nipple deviated to the right. No axillary lymph nodes were palpable. Left breast and axilla examination appear normal. Other parts of the skin show multiple Café au lait spots and cutaneous neurofibroma.

Serial mammogram and ultrasound breast 2017-2018 shows thicken skin at right periareolar with increase vascularity which may represent haemangioma. Lesions were categorized according to the Breast Imaging Reporting and Data System (BIRADS) BIRADS 4a. An ultrasound-guided biopsy was taken, however histopathological shows appearances of Phyllodes tumor.

Fig. 1. Image shows huge café-au-lait macules occupied right breast. There are also multiple neurofibromas seen at the right arm

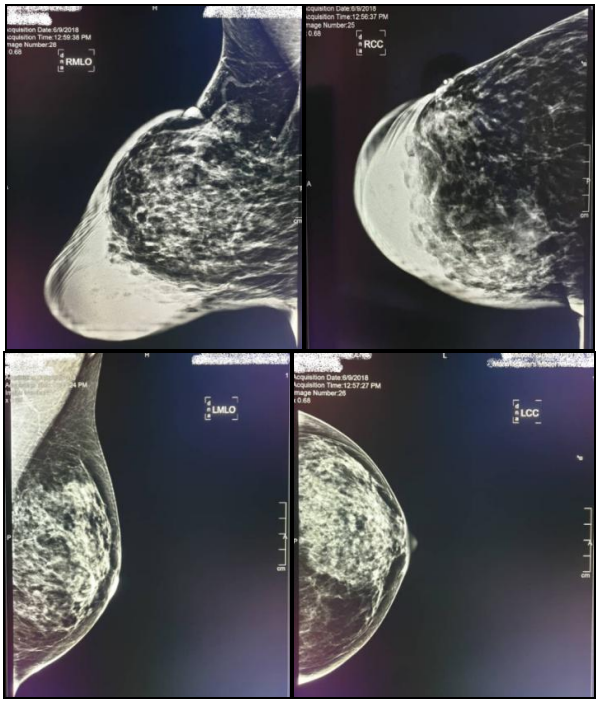

Fig. 2. Shows thicken skin at peri areolar region of the right breast at about 2-7 o'clock. Breast Imaging Reporting and Data System (BIRADS) BIRADS 4a

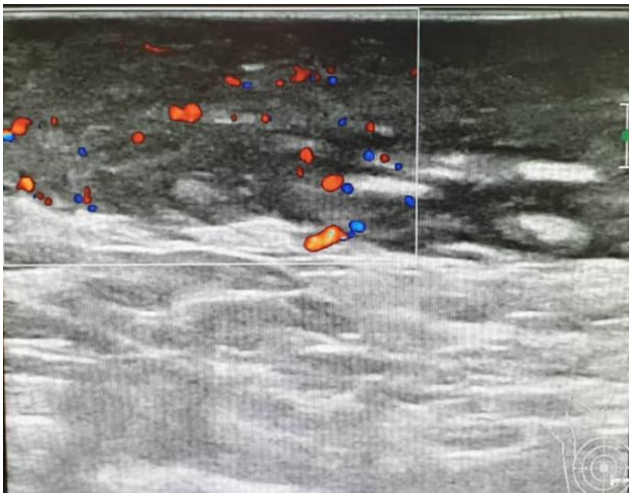

Fig. 3. Shows ultrasound peri areolar region with increase color doppler

A discussion was made as appearances of presentations, mammogram suggestive haemangioma of breast however histopathological shows Phyllodes tumor. Discussion for a less invasive technique such as embolization and wide local excision of the tumor. However, imaging shows huge haemangioma which appears not feasible as unsure placement of embolization. The decision for a simple mastectomy was made after a discussion with the patient. Post-operative histopathology shows a peripheral neural sheath tumor in favor of neurofibroma with no evidence of malignancy or Paget disease. Postoperatively the patient well. Further plan for the patient as no evidence of malignancy is for mammogram surveillance.

3. DISCUSSION

Neurofibromatosis in the breast is a rare presentation. There were some various reports of these rare presentations of breast.

In 2019, a case was reported by Yoji Yamagishi [1]. A 67-year-old female was diagnosed with NF1 at the age of 25 years with a mastectomy done for left-sided breast cancer (T2N0M0 stage IIA) at the age of 33 years. Presented with a lump in the right breast in December 2016. Physical examination, a 2 cm tumor was palpated in the upper-outer quadrant of the right breast. The patient had neurofibromas over the trunk. A high-density microlobulated mass with micro calcifications was observed in the right breast on a mammogram. Core needle biopsy specimens from the mass revealed invasive ductal carcinoma.

In 2020, *Dawood Findakly* [2] *reported a* 39-year-old G4P3 Hispanic woman with a past medical history of NF1 was diagnosed in July 2003. Noted left breast lump since 2019. On examination, there were multiple cutaneous neurofibromas over the bilateral breast and trunk. The ultrasound showed an irregular mass with speculated margins and a surrounding hyperechoic halo. Mammography was performed and it demonstrated a solitary, speculated mass within the central inferior left breast with a subtle area of architectural distortion. Subsequently, an ultrasound-guided core biopsy was done, and histological diagnosis of a poorly differentiated, grade III left breast invasive ductal carcinoma.

In 2015, Khalil, J. [3] reported a 39-year-old white Arabic woman was diagnosed with NF1 by her neurologist when she was eight years old; she was noted to have several cafe-au-lait spots. On palpation, this lump was noted to be hard and irregular. Mammography was suggested malignancy (BI-RADS 4). Her biopsy was positive for invasive ductal carcinoma. Subsequently proceeded with mastectomy and axillary clarence.

In 2019, *Martin Ignacio Zapata Laguadoa* [4] reported a 55-year-old female patient was diagnosed with NF1. Presented with 1-month of a right breast lump, associated with multiple axillary lumps (lymphadenopathy). Biopsied

finding an infiltrating ductal carcinoma, poorly differentiated.

NF1 is an autosomal dominant genetic disorder. Varies gene abnormalities such as chromosomal deletions, insertions, rearrangements, and duplications are seen in the NF1 gene [5]. A cohort study shows in NF1 patients had a risk of more than five-fold developing breast cancer [6]. The molecular study concludes that the pathogenesis of breast cancer with NF1 correlates with the proximity location of Breast cancer 1 (BRCA1) and NF1 genes on chromosome 17 [7]. Patients with NF1 who are younger than 50 years are at increased risk of developing breast cancer [8]. Diagnosed a breast lump or cancer in an NF1 patient is difficult. Cutaneous neurofibroma is hardly differentiated with pure breast lump via examinations or mammogram which results in a false negative. Most of the patient also often ignored the mass and treat as simple skin manifestation which results in advance of disease or late diagnosis. A study suggested an early mammogram at age > 40 years old [9]. However, recommendation modalities such as MRI is the best imaging modality to evaluate morphology and enhancement of lesion or ultrasound when MRI cannot be done in certain situations [10].

4. CONCLUSION

Early screening with the aid of a mammogram and ultrasound breast in a patient with NF1 helps to differentiate suspicious breast mass with a cutaneous lesion. Understanding the association of NF1 with breast cancer can help the prevalence of detection cases related. Careful clinical breast examination, screening mammography, and further diagnostic evaluation is a must.

CONSENT

As per international standard or university standard, patients' written consent has been collected and preserved by the authors.

ETHICAL APPROVAL

It is not applicable.

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COMPETING INTERESTS

Authors have declared that no competing interests exist.

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