Case Report

Journal Homepage: http://crcp.tums.ac.ir

Breast Cancer in Teenager with Neurofibromatosis 1

Nariman Khan*, **Isadora Linford**, **Sara Ortiz Romero**

Department of Diagnostic Radiology University of Texas Health Science Center San Antonio San Antonio, Texas, USA

Use your device to scan and read the article online

Citation Khan N, Linford I, Ortiz Romero S. Breast Cancer in Teenager with Neurofibromatosis 1. Case Reports in Clinical Practice. 2024; 9(1): 39-41.

Running Title Breast Cancer in NF1

Article info:
Received: January 25, 2024
Revised: February 17, 2024
Accepted: February 26, 2024

Keywords:
Breast cancer; Teenager

ABSTRACT

Breast cancer is highly prevalent amongst older women but much less common in younger women, especially those under the age of 20. Here we present a case of an 18-year-old woman with Neurofibromatosis 1 diagnosed with breast cancer. Review of literature demonstrates a five-fold increased risk of developing breast cancer before age 50 with a mutation in this tumor suppressor gene. Incorporation of earlier screening recommendations for at-risk younger women is warranted in more guidelines.

Introduction

Breast cancer affects 1 in 8 women in the United States, with the median age at diagnosis being 63 years [1]. Approximately 4 percent of breast cancers occur in women under age 40, with a nearly negligible percentage occurring in women under age 20 [1]. Here we present the case of an 18-year-old female with neurofibromatosis 1 (NF1) who was found to have invasive ductal carcinoma.

Case Presentation

An 18-year-old Hispanic female with a past medical history of NF1 complicated by bilateral optic nerve gliomas at age 6 (s/p vincristine and carboplatin) and hydrocephalus (s/p ventriculoperitoneal shunt) at age 14 presented to the clinic with spontaneous bloody right nipple discharge. The patient endorsed associated slight nipple retraction since the onset of her symptoms. Physical exam of the right breast was negative for palpable masses or reproducible discharge. Extensive laboratory testing, including pregnancy test, prolactin, TSH, FSH, LH, cortisol, and AFP, proved unremarkable. Right breast ultrasound was obtained and revealed a simple cyst in the retroareolar region measuring 6 mm. No suspicious masses or architectural distortion were visualized. However, due to the patient's concerning presentation, MRI of the bilateral

*** Corresponding Author:**
Nariman Khan
Address: Department of Diagnostic Radiology University of Texas Health Science Center San Antonio San Antonio, Texas, USA
E-mail: khan_nariman@yahoo.com

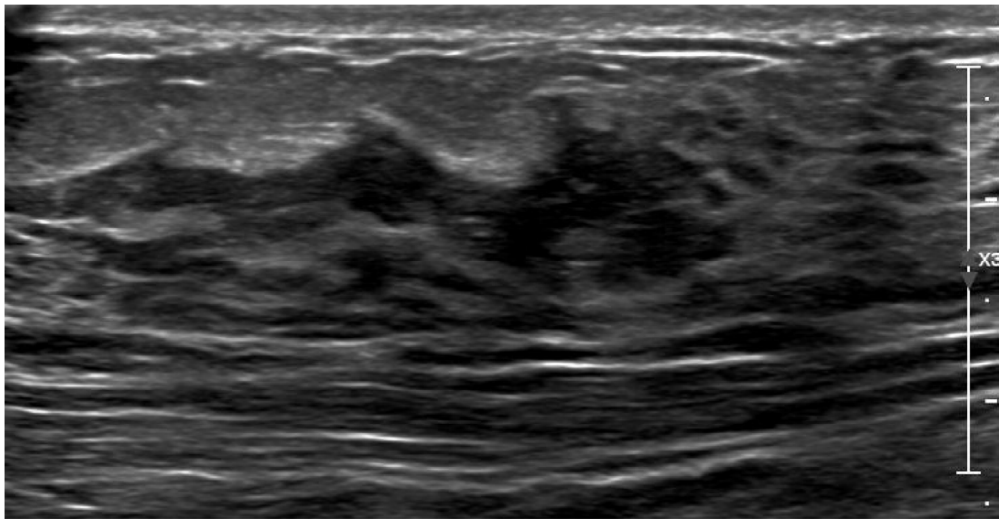

Fig. 1. Ultrasound of Right Breast Upper Inner Quadrant Mass

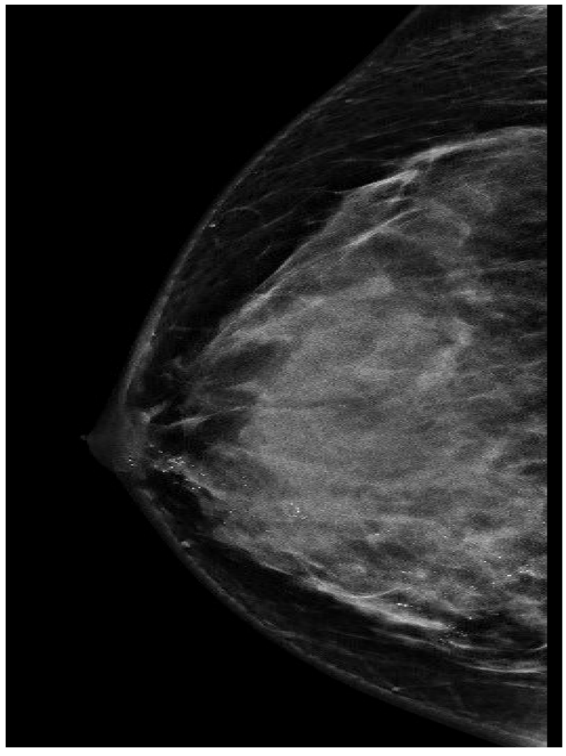

Fig. 2. Diagnostic Mammogram: RCC view with Suspicious Intraductal Calcifications

breasts was recommended for further evaluaton. MRI of the right breast revealed extensive 5.8 cm segmental non-mass enhancement from 2:00 - 4:00 with a 2 cm oval mass at 4:00. The left breast was unremarkable(Figure 1) . The patent underwent a diagnostic right breast mammogram, which revealed highly suspicious intraductal calcifications and ductal irregularity involving the medial right breast, for which biopsy was recommended(Figure 2). Ultrasound and stereotactic biopsies of the right breast were performed and revealed grade 2, ER+ Her2+ invasive ductal carcinoma and extensive ductal carcinoma in situ.

The patient was started on TCHP chemotherapy (Docetaxel, Carboplatn, Trastuz umab, Pertuzumab) with an appropriate response. Genetic testing was performed and revealed known NF1 along with a variant of uncertain signifcance (VUS) in BR CA 1.

Discussion

Breast cancer in teenage women is exceedingly rare, with an incidence of 0.1 per 100,000 or 1 in a million teenagers [2]. The most common causes for

breast cancer in young women include harboring the BRCA 1/2 mutation, prior chest radiation, and having a family history of a first-degree relative with breast cancer [2]. This patient had a VUS in BRCA 1, which may have contributed to her developing cancer; however, there is limited evidence at this time to determine if this was the leading cause. She also had NF1, which is caused by a mutation in the NF1 tumor suppressor gene. This increased her risk of developing breast cancer before age 50 by five-fold [3]. It is currently recommended by the National Comprehensive Cancer Network that women with NF1 undergo an annual screening mammogram beginning at age 30, with consideration for MRI between ages 30 and 50 [3].

Conclusion

Women with cancer-predisposing syndromes such as NF1 should be closely monitored for signs and symptoms of malignancy. Incorporation of earlier screening recommendations for at-risk younger women is warranted in more guidelines.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this article.

Funding

No funding was received to assist with the preparation of this manuscript.

Conflict of Interests

The authors have no conflict of interest to declare.

References

[1] Susan G. Komen®. Breast Cancer Statistics | Susan G. Komen®. Susan G. Komen®. Published November 17, 2023. Available from: https://www.komen.org/breast-cancer/facts-statistics/breast-cancer-statistics/

[2] Miller KD, Fidler-Benaoudia M, Keegan TH, Hipp HS, Jemal A, Siegel RL. Cancer statistics for adolescents and young adults, 2020. CA Cancer J Clin. 2020;70(6):443-459. https://doi.org/10.3322/caac.21637

[3] Maani N, Westergard S, Yang J, Scaranelo AM, Telesca S, Thain E, Schachter NF, McCuaig JM, Kim RH. NF1 Patients Receiving Breast Cancer Screening: Insights from The Ontario High Risk Breast Screening Program. Cancers (Basel). 2019 May 22;11(5):707. https://doi.org/10.3390/cancers11050707