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Case Report

Triple-Negative Breast Cancer in a Young 23-Year-Old Woman with BRCA Mutation A about A Case and Literature Review

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Abstract

Triple-negative breast cancer (estrogen receptor negative, progesterone receptor negative and HER2 negative) is a particularly high-risk breast cancer which does not receive specific therapy targeting these proteins. We report the case of a 23-year-old patient diagnosed with triple-negative breast cancer of the left breast with a family history of breast cancer and carrying a BRCA1 mutation. The classification of breast cancer based on genomic data is required to allow us to optimize therapies and improve the management of breast cancer patients.

Keywords: Breast Cancer, Triple Negative, HER2.

INTRODUCTION

Triple-negative breast cancer (TNBC) is a type of breast cancer characterized by the absence of estrogen and progesterone hormone receptors, as well as the non-expression of the HER2 receptor. This particularity renders hormonal treatments and targeted therapies inapplicable, posing a major challenge in the management of this disease [1].

TNBC accounts for about 16.09% in Morocco of all breast cancers and is more common in younger women, often diagnosed before the age of 40. Patients with TNBC generally have a less favorable prognosis due to the aggressive nature of the disease [2].

CLINICAL CASE

A 23-year-old patient presented with a palpable mass in the left breast. Family history of breast cancer in the mother and serous adenocarcinoma of the ovary in the aunt. Mammography and ultrasound imaging revealed two lesions with spiculated contours, one in the supero-lateral quadrant measuring 2 cm and the other in the infero-internal quadrant, with no axillary lymphadenopathy. A biopsy confirmed the diagnosis of TNBC with a ki67 of 60%.

MRI showed a Upper and outer quadrant lesion. spiculated contours, hyposignal T1, intermediate signal T2, diffusion hypersignal, intensely contrast-enhancing after injection of Gadolinium, a second lobulated oval lesion in the Inferior and Inner Quadrant, with irregular contours and a long axis perpendicular to the skin, with a similar signal.

The genetic test was in favour of a BRCA1 mutation the decision was to undergo neoadjuvant chemotherapy followed by mastectomy with subsequent breast reconstruction using prostheses. Contralateral mastectomy should be discussed from the age of 30.

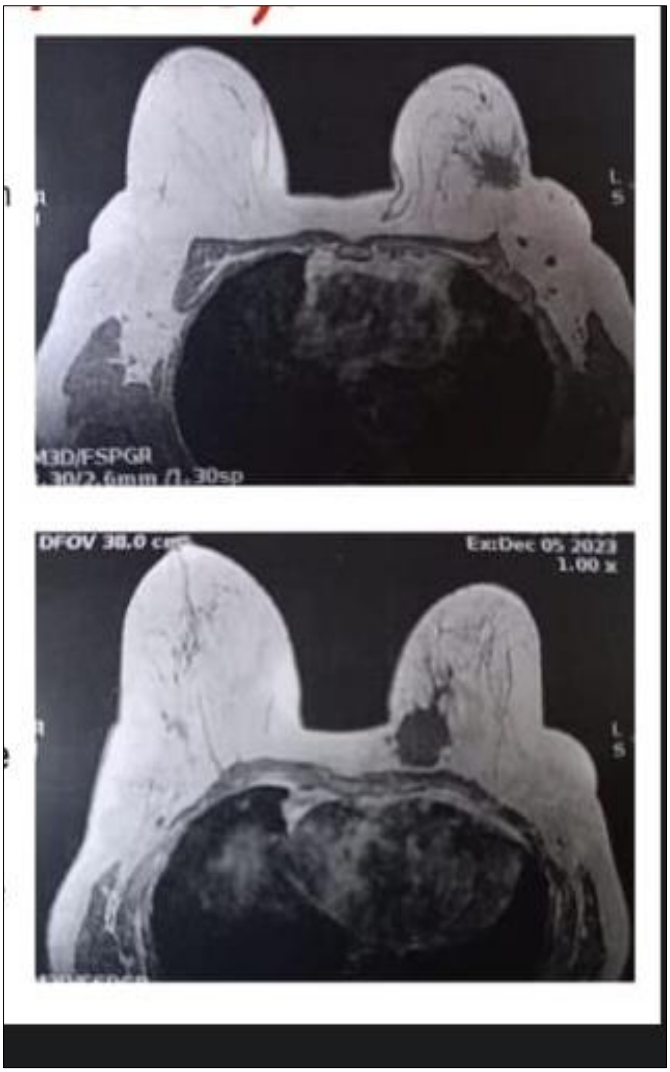

Figure 1: MRI showing two lesions on the left breast

DISCUSSION

Clinicopathological and molecular definitions of the subtypes of invasive breast cancer were adopted by the 13th international conference of the group of experts on breast cancer in Saint Gall (2013). These definitions are based on interesting immunohistochemical criteria, including RO, RP, ERBB2 (HER2), and Ki-67, with in situ hybridization confirmation when necessary [3].

These subtypes are HER2-positive (non-luminal), Luminal A-like, Luminal B-like (HER2-negative), Luminal B-like (HER2-positive) and Triple-negative. Lehmann *et al*., (2011) performed gene expression profiling of tumors from 587 breast cancer patients. Basal-like 1 (BL1), basal-like 2 (BL2), mesenchymal (M), mesenchymal stem-like (MSL), immunomodulatory (IM) and luminal androgen receptor (LAR) are the six CSTN subtypes that this study classified. This new classification has provided a precise cellular model for modifying the therapeutic orientation of CSTN [4].

Umemura *et al*., [5]. Have revealed that 11 out of 58 cases of breast cancer are combined estrogen receptor-negative and HER2-negative tumors, or 19% of cases. When compared to other tumor groups, these tumors had the lowest expression of cyclinD1 and the highest ki-67 Labeling Index. They were also linked to high expression of p53, vimentin, and EGFR.

Studies provide strong evidence for the assessment of basal cytokeratins and androgen receptors, in addition to traditional pathological parameters (tumor size and lymph node status), to provide prognostic guidance in the group of tumors with a triple-negative phenotype. Assessment of p53, P-cadherin and E-cadherin did not offer significant prognostic information in this class of tumors. In the lymph node-less subgroup, the basal phenotype may provide strong prognostic information independently of other well-known markers, and may identify a specific subgroup of patients who may profit from a more aggressive adjuvant therapy approach. We therefore emphasize the importance of systematically staining triple-negative breast cancers for androgen receptors and basal cytokeratins [6].

Hereditary predisposition to this type of cancer is essentially linked to mutation of the BRCA1 gene. A BRCA1 or BRCA2 mutation also increases the risk of developing adnexal cancer, which is even higher and earlier in the case of a BRCA1 mutation. The cumulative risk at age 70 is 22% to 59%, for a median age at diagnosis of 52±10 years; the cumulative risk at age 70 and median age at diagnosis for BRCA2 mutations are 4% to 18% and 60±11 years respectively [7].

In contrast to tumors expressing hormone receptors, hormone receptor-negative breast cancers are a diverse category of breast cancers that are typically considered to be aggressive, have a poor prognosis, and have less cancer preventive and treatment techniques. Furthermore, treatment that specifically targets this protein (such as trastuzumab-based therapy) is not helpful in HER2-negative malignancies [8]. Consequently, there are fewer therapy choices available for these triple-negative cancers.

The management of TNBC relies on a combination of surgery, chemotherapy, and radiotherapy. Surgery may involve a lumpectomy or mastectomy, followed by neoadjuvant or adjuvant chemotherapy to eliminate residual cancer cells. Radiotherapy is also used to treat the affected breast area and regional lymph nodes.

Recent advances have been made in the treatment of TNBC, including the introduction of immunotherapy and new chemotherapeutic agents. Trodelvy (sacituzumab govitecan) has been approved for patients who have failed therapeutic treatment after two or more lines of systemic treatment. Additionally, the addition of capecitabine to neoadjuvant chemotherapy has shown an improvement in prognosis in certain cases

Adjuvant chemotherapy is administered primarily on the basis of certain clinical and pathological factors (tumor size, tumor stage and lymph node status) based on cyclophosphamide, methotrexate and fluorouracil (CMF) and cyclophosphamide, doxorubicin, fluorouracil (CAF). For TNBC tumors, the difference between CAF and CMF was not significant [1].

However, responses to chemotherapy are not always similar even in patients with similar clinical and pathological factors. For patients with positive lymph nodes, relapse is more frequent [1].

CONCLUSION

TNBC in young women requires a multidisciplinary and personalized approach. Active research for new therapeutic targets and the improvement of screening strategies are essential to improve the prognosis of these patients.

This article provides a detailed overview of TNBC in a young patient, focusing on diagnostic and therapeutic challenges as well as recent advances in the treatment of this disease. It is important to note that each case is unique and requires individual evaluation by a specialized care team.

DECLARATIONS

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Consent Informed: written consent obtained from the patient.

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