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CASE REPORT

Triple-Negative Accessory Breast Cancer Associated with a Germline BRCA1 Mutation: A Case Report

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Abstract

Background: Accessory breast carcinoma is a rare malignancy arising from ectopic mammary tissue along the embryologic milk line, most frequently located in the axillary region. Due to its atypical anatomical presentation, diagnosis may be delayed or mistaken for benign axillary lesions or metastatic lymphadenopathy. However, the molecular characteristics and hereditary predisposition patterns of accessory breast cancer remain poorly understood. **Case Presentation:** A 36-year-old postpartum woman presented with a progressively enlarging right axillary mass. Imaging demonstrated a lesion arising from accessory breast tissue without evidence of malignancy in the orthotopic breasts. Core needle biopsy revealed invasive ductal carcinoma of no special type with a triple-negative phenotype (estrogen receptor-negative, progesterone receptor-negative, and HER2-negative) and a Ki-67 index of 70%. The patient received neoadjuvant dose-dense anthracycline-based chemotherapy followed by paclitaxel and carboplatin, achieving a near-pathologic complete response after surgical excision and sentinel lymph node biopsy. Adjuvant radiotherapy was subsequently administered. Germline genetic testing identified a pathogenic BRCA1 mutation. At 24 months of follow-up, the patient remains clinically and radiologically disease-free. **Conclusion:** Triple-negative carcinoma arising from accessory breast tissue is a rare presentation that may be associated with hereditary breast cancer predisposition. This case highlights the potential value of germline evaluation in young patients with triple-negative accessory breast carcinoma and emphasizes the role of molecular characterization in individualized clinical management.

Keywords: Accessory breast carcinoma; Ectopic breast tissue; Triple-negative breast cancer; BRCA1 mutation

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Introduction

Accessory breast tissue, also known as ectopic or supernumerary breast tissue, results from incomplete regression of the embryologic mammary ridge along the milk line. It is most commonly located in the axillary region and can undergo the same physiological and pathological changes as normal breast tissue, including malignant transformation [1]. Although accessory breast tissue is relatively common, primary carcinoma arising within this tissue is rare and is mainly described through case reports and small case series [2–4].

The diagnosis of accessory breast carcinoma can be challenging because axillary masses are frequently attributed to benign lesions or metastatic lymphadenopathy. Distinguishing primary carcinoma arising from accessory breast tissue from metastatic involvement of axillary lymph nodes is essential because it influences surgical planning and treatment decisions [5–7]. Management generally follows principles established for conventional breast cancer; however, limited evidence exists regarding the molecular characteristics and optimal treatment strategies for these rare tumors.

Invasive ductal carcinoma is the most frequently reported histological subtype of accessory breast cancer, whereas information regarding molecular subtypes remains limited. In particular, the prevalence and clinical implications of triple-negative breast cancer (TNBC) arising from accessory breast tissue are poorly characterized. TNBC, defined by the absence of estrogen receptor (ER), progesterone receptor (PR), and HER2 expression, represents an aggressive breast

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cancer subtype with distinct therapeutic considerations. BRCA1-associated breast cancers frequently exhibit a triple-negative phenotype and may demonstrate increased sensitivity to platinum-based chemotherapy due to impaired DNA repair mechanisms [8-10].

Pregnancy-associated and postpartum breast cancers represent another biologically distinct group, with unique hormonal and microenvironmental characteristics that may influence tumor behavior [11, 12]. However, the coexistence of postpartum presentation, accessory breast carcinoma, triple-negative phenotype, and germline BRCA1 mutation has rarely been described.

Here, we report a rare case of triple-negative invasive ductal carcinoma arising from accessory axillary breast tissue in a young postpartum woman with a pathogenic germline BRCA1 mutation. This case highlights the diagnostic challenges of ectopic breast malignancy and emphasizes the potential importance of molecular evaluation in guiding personalized treatment strategies.

Case Presentation

A 36-year-old postpartum woman presented in July 2022 with a progressively enlarging right axillary mass arising from accessory breast tissue. She had first noticed the lesion in November 2021, shortly after childbirth, as a small palpable lump that gradually increased in size over several months. She had no previous history of breast disease, significant comorbidities, or known family history of breast or ovarian cancer at initial presentation.

Diagnostic mammography performed on July 16, 2022, showed no abnormalities in either orthotopic breast; however, an irregular dense mass was identified within the right axillary accessory breast tissue. Targeted ultrasound of the right axilla (July 14, 2022) demonstrated a solid-cystic mass measuring 38.7 × 22.4 mm. Subsequent breast magnetic resonance imaging confirmed a 46 × 26 mm mass confined to the right accessory breast tissue without suspicious lesions in either native breast. Abdominal and pelvic ultrasound revealed no evidence of distant metastasis.

Core needle biopsy of the axillary lesion performed on July 14, 2022, revealed invasive ductal carcinoma of no special type. Immunohistochemical analysis demonstrated negativity for estrogen receptor (ER), progesterone receptor (PR), and HER2 expression, confirming a triple-negative breast cancer phenotype. The Ki-67 proliferation index was 70%.

The patient received neoadjuvant chemotherapy beginning in August 2022, consisting of dose-dense doxorubicin and cyclophosphamide administered every two weeks for four cycles, followed by four cycles of paclitaxel combined with carboplatin every three weeks. Treatment was generally well tolerated, although she experienced mild gastrointestinal symptoms, mucositis, transient bone pain, and chemotherapy-induced peripheral neuropathy. One chemotherapy cycle was delayed due to neutropenia (absolute neutrophil count 1000/μL), and granulocyte colony-stimulating factor support was subsequently administered.

Following completion of neoadjuvant therapy, surgical excision of the accessory breast tumor with sentinel lymph node biopsy was performed on January 1, 2023. Final histopathological examination revealed only a microscopic residual focus of invasive ductal carcinoma, with clear surgical margins and no metastatic involvement of the four examined sentinel lymph nodes. The findings indicated an excellent treatment response with minimal residual disease, although a complete pathological response was not achieved.

Adjuvant radiotherapy targeting the right axillary region and accessory breast bed was completed in March 2023. The treatment course was uncomplicated except for mild localized radiation dermatitis.

Because of her young age and triple-negative tumor phenotype, germline genetic testing was subsequently performed. In August 2024, testing identified a pathogenic BRCA1 mutation. Genetic counseling was provided, and risk-reducing bilateral mastectomy and salpingo-oophorectomy were discussed. However, the patient wished to preserve the possibility of future pregnancy and elected to defer prophylactic surgery. At the last follow-up in February 2025, she remained clinically and radiologically free of disease.

Discussion

Primary carcinoma arising in accessory breast tissue is a rare clinical entity, with available evidence largely limited to case reports and small case series. These tumors most commonly present as axillary masses and may be difficult to distinguish from benign lesions or metastatic lymph node involvement. Previous reports have emphasized the importance of accurate diagnosis and appropriate staging because management generally follows established breast cancer principles.

The present case is distinctive because of the coexistence of an uncommon anatomical presentation with a biologically defined triple-negative phenotype and a pathogenic germline BRCA1 mutation in a young patient. Although molecular data on accessory breast carcinoma remain limited, the clinical characteristics of this tumor appear consistent with those of BRCA1-associated triple-negative breast cancer (TNBC).

BRCA1-associated breast cancers frequently exhibit triple-negative characteristics due to defects in homologous recombination-mediated DNA repair. This biological feature provides a rationale for increased sensitivity to DNA-damaging agents, including platinum compounds. In the present case, the excellent response following neoadjuvant chemotherapy containing carboplatin is consistent with the potential platinum sensitivity observed in BRCA1-deficient tumors. However, this response should be interpreted in the context of the combined multi-agent chemotherapy regimen rather than attributed exclusively to platinum exposure.

An important aspect of this case is that the diagnosis of accessory breast carcinoma requires careful differentiation from metastatic axillary disease. Imaging demonstrating a lesion confined to accessory breast tissue and negative sentinel lymph nodes supported the diagnosis of a primary

ectopic breast malignancy and allowed treatment planning according to breast oncology principles.

Beyond treatment considerations, identification of a germline BRCA1 mutation has important implications for hereditary cancer counseling and long-term risk management. Young patients with triple-negative breast cancer, even in unusual anatomical locations, may benefit from consideration of genetic evaluation, particularly when clinical features suggest hereditary predisposition.

The postpartum presentation of this patient also warrants consideration. Pregnancy-associated and postpartum breast cancers may exhibit distinct biological characteristics; however, available evidence does not establish a causal relationship between postpartum status and tumor development in this case. The molecular profile, particularly BRCA1-associated triple-negative biology, provides a more plausible explanation for the tumor phenotype and treatment response.

Overall, this case expands the limited literature on accessory breast carcinoma by demonstrating that ectopic breast malignancy may share important molecular characteristics with conventional breast cancer. The presence of a triple-negative phenotype in a young patient with accessory breast carcinoma should prompt consideration of germline testing and individualized therapeutic planning.

In conclusion, accessory breast carcinoma is a rare malignancy that may present diagnostic challenges because of its atypical axillary location. This case highlights the occurrence of triple-negative carcinoma arising from ectopic breast tissue in a young patient with a germline BRCA1 mutation. The favorable response to platinum-containing neoadjuvant therapy is consistent with the biological characteristics of BRCA1-associated breast cancer. Recognition of hereditary features in young patients with triple-negative accessory breast carcinoma may support genetic counseling and personalized management strategies.

Ethics Statement

This case report was prepared in accordance with institutional ethical standards and the principles of the Declaration of Helsinki. Ethical approval was not required according to institutional policies for single-patient case reports. Written informed consent was obtained from the patient for publication of clinical information and any accompanying materials. All identifying information has been removed to protect patient confidentiality.

Conflict of Interest Statement

The authors declare that they have no conflicts of interest related to this work.

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