

Bilateral Risk-Reducing Mastectomy with Immediate Prosthetic Reconstruction in a Brca2 Mutation Carrier with Previous Breast Cancer: a Case Report and Review of the Literature

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Abstract

Women carrying a pathogenic variant in the BRCA2 gene have a substantially elevated lifetime risk of breast and ovarian cancer [1]. Risk-reducing bilateral mastectomy (RRBM) is an established option that can reduce breast-cancer incidence by more than 90% [2-4]. We report the case of a 34-year-old nulligravid woman with a documented BRCA2 mutation who had previously undergone breast-conserving surgery, chemotherapy, radiotherapy and hormonal therapy for a left-sided luminal-B invasive carcinoma in 2021. During subsequent high-risk surveillance a stable, small retro-areolar nodule of benign appearance was identified in the contralateral breast. After thorough multidisciplinary counseling the patient elected bilateral mastectomy with immediate implant-based reconstruction. This case illustrates the decision-making process surrounding secondary risk-reducing surgery in a young BRCA2 carrier with a personal history of breast cancer and underscores the importance of individualized counseling, imaging surveillance and shared decision-making.

Keywords: BRCA2 mutation, Risk-reducing mastectomy, Bilateral mastectomy, Hereditary breast cancer, Case report

Abbreviations: MRI: Magnetic resonance imaging; RRBM: Risk reducing bilateral mastectomy

Case Report

A 34-year-old married, nulligravid woman was referred to our department for evaluation of surgical risk-reduction options. She had a documented pathogenic BRCA2 variant and a personal history of left breast cancer diagnosed in 2021.

In 2021 she underwent left tumorectomy. Histopathology revealed an invasive carcinoma of no special type (NOS), Scarf-Bloom-Richardson grade III, luminal-B phenotype, HER2 scored ++, with two metastatic axillary lymph nodes out of eleven examined (2N+/11N). She subsequently completed adjuvant chemotherapy, radiotherapy and hormonal therapy. Her mother was under follow-up for breast neoplasia, further sup-

porting the hereditary context.

On clinical examination the patient was in good general condition (blood pressure 120/70 mmHg, heart rate 82 bpm, respiratory rate 16 cycles/min). The left breast showed a well-healed tumorectomy scar without palpable nodule or mass; the right breast was free of any palpable abnormality. Axillary and supraclavicular lymph-node basins were clear.

Breast imaging performed as part of high-risk surveillance demonstrated a small, smooth-contoured retro-areolar enhancement measuring 5 mm in the right breast, classified ACR 3, while the left breast was ACR 2 (post-therapeutic changes). Magnetic resonance imaging (MRI) in June 2024 confirmed a

stable, oval, regular-contoured retro-areolar nodule of 6×2.5 mm on the right side, judged of benign appearance, together with discrete thickening of the left nipple-areola complex related to prior treatment. Ultrasound in December 2024 corroborated these findings.

The use of MRI in high-risk breast surveillance is consistent with published EUSOMA recommendations [5]. The imaging findings themselves are patient-specific observations.



Figure 1: Breast magnetic resonance imaging (MRI) demonstrating the right retro-areolar lesion.

After extensive genetic counseling, psychological support and discussion within a multidisciplinary tumor board, the patient, motivated by her personal and family history and by the residual lifetime risk of contralateral and ipsilateral recurrence, elected to undergo bilateral mastectomy with immediate prosthetic reconstruction. Surgery was performed without major complications. Final histopathology of the resected tissue showed no invasive or in-situ carcinoma.

Discussion

Pathogenic variants in BRCA2 confer a lifetime breast-cancer risk of approximately 45–69 % and an ovarian-cancer risk of 11–17 % [1]. After a first breast-cancer diagnosis the cumulative risk of contralateral breast cancer reaches 20–30 % at 10–15 years, substantially higher than in non-carriers [1]. Consequently, international guidelines (NCCN, ESMO) recommend discussing risk-reducing bilateral mastectomy (or contralateral risk-reducing mastectomy after unilateral disease) as part of comprehensive counseling [6,7].

In women with a prior personal history of breast cancer who carry a BRCA2 mutation, secondary risk-reducing surgery aims to eliminate residual breast tissue that remains at high risk of a new primary. Several series and systematic reviews have documented the risk-reducing effect of prophylactic or contralateral mastectomy in high-risk women [2-4,8,9]. The procedure reduces the incidence of subsequent breast cancer by more than 90 % [2], although the magnitude of an overall-survival benefit has varied across observational studies

[10-13].

Our patient presented the classic profile of a young BRCA2 carrier: early-onset luminal-B disease, positive family history, and ongoing high-risk imaging surveillance that identified only a stable, radiologically benign contralateral lesion. The decision for bilateral mastectomy rather than continued surveillance was driven by patient preference after balanced information on residual risk, surgical morbidity, impact on body image and sexuality, and the availability of immediate implant-based reconstruction.

Immediate reconstruction with implants is a widely accepted option after risk-reducing mastectomy and is associated with high patient satisfaction when performed by experienced teams. Nipple-sparing or skin-sparing techniques may further improve aesthetic outcomes, provided oncologic safety is preserved [14]. In the present case, prosthetic reconstruction was chosen and yielded a satisfactory result.

Surveillance alternatives (annual MRI $\pm$ mammography) remain valid for women who decline surgery [5-7]. Chemoprevention with tamoxifen or aromatase inhibitors may also be considered, particularly for hormone-receptor-positive disease, although data specific to BRCA2 carriers are still limited [6,7]. Risk-reducing salpingo-oophorectomy is generally recommended between ages 40 and 45 for BRCA2 carriers once childbearing is complete [6,7], both for ovarian-cancer risk reduction and for an additional modest reduction in breast-cancer risk [15].

This case adds to the growing literature illustrating that, even after successful treatment of a first breast cancer, a young BRCA2 carrier may reasonably elect bilateral mastectomy with reconstruction. Shared decision-making, multidisciplinary input and careful preoperative imaging remain essential [5-7].

Conclusion

Risk-reducing bilateral mastectomy with immediate prosthetic reconstruction is a valid and effective option for BRCA2 mutation carriers who have already been treated for breast cancer and who wish to minimize residual risk [2,4,9,10]. Individualized counseling that integrates personal history, family history, imaging findings, reproductive plans and patient values is mandatory [6,7]. Continued long-term follow-up and consideration of risk-reducing salpingo-oophorectomy complete the preventive strategy [7,15].

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