Prophylactic Gender-Affirming Mastectomy: Unique Considerations in BRCA

Bailey Johnson¹, Ivan Hadad¹, Aladdin H. Hassanein¹, and Carla Fisher²

November 14, 2024

Introduction

The transgender and gender diverse population continues to rise as society shifts towards increased awareness and acceptance, with an estimated 2.6 million adults identifying as transgender in the United States [1]. Population growth and improved financial accessibility to healthcare has led to an increase in chest masculinization surgery, or gender-affirming mastectomy (GAM) [2]. GAM differs from risk-reduction, or prophylactic, mastectomy in that more breast tissue is left in-situ to create a natural-appearing chest contour. This is compared to prophylactic mastectomy which removes nearly all breast tissue for maximum oncologic risk reduction. This difference complicates breast cancer risk assessment in the post-GAM population [2]. Breast cancer risk in cisgender women is well-defined at 1 in 8 or 12.5% risk over a lifetime, however this is less clear in transgender patients due to confounding factors such as testosterone therapy or previous opphorectomy [3]. It has been theorized that after GAM transmale patients are at lower risk of breast cancer than cisgender women, yet higher risk than cisgender men [2-4]. No standardized preoperative screening guidelines prior to GAM currently exist, however strong family history and need for breast imaging should be assessed in this setting. Risk factors such as inherited genetic mutations have specific implications for this population as they are less likely to undergo cancer screening or participate in long-term follow up [4]. Approximately 5-10% of breast cancers are thought to be associated with inherited genetic mutations, BRCA being most common [5]. Risk reducing mastectomy is often offered to these patients, decreasing breast cancer risk by 90-95%[4,5]. Here we present a transgender male patient with the BRCA1 mutation. Rather than standard GAM, he underwent prophylactic bilateral mastectomy followed by chest masculinization with liposuction and free nipple grafts, as well as concurrent oophorectomy for gynecologic risk reduction. Here we highlight the nuanced problem that post-GAM patients have a higher, more variable risk of breast cancer compared to cisgender men and likely postmastectomy cisgender women. We demonstrate that appropriate risk reduction and desired cosmetic outcome can be achieved concurrently. The following case in presented accordance with the CARE reporting checklist.

Case Presentation The patient is a 32-year-old transgender male who presented for surgical consultation regarding prophylactic mastectomy in the setting of a BRCA1 mutation. At the time of consultation, he had been on testosterone therapy for 7 years. Genetic testing was ordered by his endocrinologist due to a strong family history of breast, ovarian, and prostate cancers. He was referred to our high-risk screening program, breast surgical oncology, and gynecology to discuss risk reduction surgery. He had no previous breast imaging and was recommended to undergo breast MRI. This showed a 5.4 cm area of nonmass enhancement in the right breast for which biopsy was recommended, however at the time of biopsy the abnormality was no longer visualized and no tissue sample was obtained. After multidisciplinary discussion, he underwent laparoscopic hysterectomy and bilateral salpingo-oophorectomy followed by bilateral risk-reduction mastectomies. The plastic surgery team worked in conjunction with the oncological breast surgery team for markings. The

¹Indiana University School of Medicine

²Division of Breast Surgery, Indiana University School of Medicine, Indianapolis, IN

double incision mastectomy approach was designed to place the scar slightly below the pectoralis major muscle. The nipples were thinned out as an oval full thickness skin graft placed on the closed chest. The nipple grafts were positioned along the lateral border of the pectoralis major muscle between the 4th and 5th ribs [6]. Suction assisted lipectomy was performed laterally and medially to optimize contour. Final pathology was without abnormality. Postoperatively he was satisfied with the aesthetic outcome (Figure 1) and plans to undergo annual chest wall exams by his primary care physician.

Discussion Here we describe a transgender male patient found to have the BRCA1 mutation on preoperative evaluation for GAM. He was found to have a strong family history including breast, ovarian, and prostate cancers which prompted genetic testing. He was evaluated in a multidisciplinary setting, and ultimately underwent risk-reduction mastectomy and oophorectomy at the time of chest masculinization surgery. There has yet to be established guidelines for breast cancer screening in transgender men undergoing GAM. In general, transgender patients face many barriers in healthcare including stigmatization, psychosocial trauma associated with undergoing procedures that do not align with their gender identity, and lack of insurance coverage of procedures [7]. Disparity in healthcare for this population is especially prevalent in preventative care. When compared to cisgender individuals, transgender patients are less likely to be offered recommended cancer screening, which is reflected by suboptimal cancer outcomes in this population [1,7]. A recent single arm pilot trial published by C.S Cortina et. al highlights implications of patient education regarding individualized breast cancer risk in the preoperative setting can impact one's decision to undergo risk reduction mastectomy as a part of gender-affirming top surgery [1]. Though rare, development of breast cancer after GAM has been described in multiple case series [7]. It is important to emphasize that GAM should not be considered equivalent to oncologic or prophylactic mastectomy due to the amount of breast tissue left behind, as the residual breast tissue imparts a theoretical future risk of breast cancer. In terms of long-term breast cancer screening, similar to postmastectomy cisgender females, physical exam is the recommended screening modality for post-GAM patients.

Conclusion As the transgender population continues to grow, the role of breast cancer risk assessment prior to GAM is of particular importance. This case demonstrates the clinical utility for formalized, individualized preoperative screening to better identify patients who would benefit from oncologic risk-reduction at the time of gender-affirming chest masculinization. As exemplified here, formal guidelines for breast cancer risk assessment in patients pursuing GAM could mitigate lifetime risk of breast cancer development in the transgender and gender diverse population. A limitation of this case report includes discussion about a relatively small patient population, however with profound population growth the question of breast cancer risk in this group has become relevant in genetics, surgical oncology, and plastics. More work is needed to develop and implement formal guidelines to assess long term outcomes.

Bailey N. Johnson: Conceptualization; writing – original draft.

Ivan Hadad: Writing – review and editing.

Aladdin H. Hassanein: Conceptualization; writing – review and editing.

Carla S. Fisher: Conceptualization; writing – review and editing.

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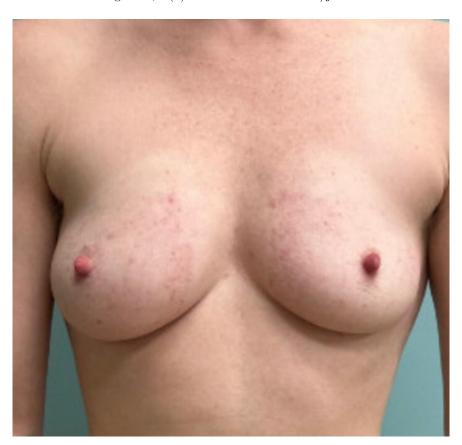




Figure Legend:

Figure 1: Transgender male with BRCA1 mutation. Prophylactic, risk-reducing mastectomy was indicated and the patient requested masculinization. (Left) Preoperative image. (Right) 3 months postoperative.

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