Decisions, Incisions, and New Dimensions: Exploring the Double Mastectomy Choice for BRCA1 Carriers Pre and Post Surgery

Abstract

Many women who test positive for the BRCA1 mutation face the daunting decision of whether or not to undergo prophylactic double mastectomy. The presurgery decision-making process requires a nuanced approach that takes into consideration the potential pros, such as emotional relief, and the cons, such as the physical and negative emotional impacts. The post-surgery effects are another factor that a BRCA carrier must take into consideration; many studies report reduced cancer-related anxiety, though they also often report body image issues and depression. While surgery often provides a sense of control for mutation carriers over their own health, it many times also brings new struggles concerning femininity and identity. As the discussion is not cut and dry, women making their decision must consider a multitude of factors; this paper will be discussing many of these elements, including but not limited to, double mastectomy, clinical management, risk assessment, mental health implications of testing positive for BRCA1/2, decision-making processes, breast implants, prosthetic breasts, quality of life post-surgery, satisfaction with surgical treatment, and social impacts. This research emphasizes the nuance of mastectomy decisions for BRCA1 carriers and highlights the importance of holistic support during this process—medical, educational, psychological, and physcosocial.

Introduction

Breast cancer is breast cells that mutate, becoming cancerous, and multiply, forming tumors. These mutations usually begin in the milk ducts or lobules and can occur in one breast or both (Cleveland Clinic, n.d.). Breast cancer can be caused by family history, genetics, radiation exposure, age, etc. The most common genetic predisposition is mutations in breast cancer (BRCA) genes BRCA1 and BRCA2. Mutations in the BRCA1 gene can significantly increase the likelihood of breast and ovarian cancer (King et al., 2003). Around 50% of women in North America with a family history of breast cancer undergo double mastectomy after testing positive for the BRCA1/2 mutation. Despite this, research about the emotional aspects of opting for a double mastectomy among these women remains limited (Metcalfe et al., 2008).

Breast cancer deaths occur in approximately 685,000 women per year worldwide (WHO, 2021). Though death rates of BRCA1 mutation carriers who developed lethal cancer are not reported, women with BRCA1 mutations have a 72% risk of developing cancer. Cancer from BRCA1/2 mutation has a high mortality rate due to the aggressive nature of the mutation (Mavaddat et al., 2013; McCarthy & Armstrong, 2014). Studies generally indicated a lower breast and ovarian cancer risk postsurgery. Yet, some also showed lower patient satisfaction levels and quality of life (van Dijk et al., 2016; ClinicalTrials.gov, 2021). Studies in Europe and North America, specifically, found challenges such as body image issues and emotional weight gain often (Frost et al., 2000).

As of 2024, in the United States, an estimated 19.3 women out of every 100,000 women die each year from breast cancer (National Cancer Institute, 2024). In the study conducted by Rebbeck et al. (2004) on BRCA1, carriers who opt for a double mastectomy reported feeling reduced anxiety about developing cancer. yet also feelings of depression. National surveys and research show that while the surgery lowers cancer risk, it can also result in anxiety, depression, and self-identity and sexuality issues (Halbert et al., 2007).

Research on the advantages and disadvantages of double mastectomy for BRCA1 carriers is important because it looks into the emotional aspects involved in undergoing the procedure. By understanding the BRCA1 mutation itself and the experiences of women with BRCA1 gene mutation, we can gain a better understanding of their decision-making process and potential life after surgery.

Methodology

This research paper uses a literature review to evaluate the decision-making process, implications, and outcomes of double mastectomies for BRCA1 mutation carriers. “BRCA1 mutation,” “mastectomy,” “breast cancer,” “psychological impact,” and other related keywords were used to find relevant articles, books, studies, etc. on sites such as Pubmed, Google Scholar, and JSTOR. In addition to the published literature used, published clinical trials were also used. No fieldwork or experiments were used in this study; online databases and digital resources were used to gather information. Peer-reviewed articles, especially from a wide array of regions, and recently published sources were prioritized to make sure that different viewpoints and up-to-date research were included; different viewpoints minimized biases. Nevertheless, it is essential to recognize the limitations with a literature review; the research relies on published works, which could have biases and limitations of their own. Or the results may not apply to all individuals with BRCA1 mutations. After all, post surgery life is subjective to the individual; it is not reasonable to assume that everyone will have the same experiences with surgery.

Tumorigenesis, Apoptosis, and Genetic Implications

Under steady state, cells grow, divide, and die in a controlled way that is regulated by specific genes. When DNA damage occurs, BRCA1 and BRCA2-associated 'DNA repair complexes' find and fix the damage. But if the damage is too severe and cannot be fixed, BRCA genes help the cell go through apoptosis, aka programmed cell death, to keep the cell from becoming cancerous (Narod, 2010). When BRCA1 or BRCA2 mutates, it can no longer repair damaged DNA as it would in a steady state. Over time, these mutations build up in the cell's DNA and can cause the cell to grow uncontrollably; they'll keep growing and dividing instead of dying when they're supposed to. These abnormal cells will eventually clump together and form a tumor, and if these cells invade surrounding tissue, it can spread to other areas of the body and become cancer (Venkitaraman, 2019).

Even if BRCA1/2 is mutated, apoptosis can still happen, though it might not be as effective. BRCA1 and BRCA2 mainly repair DNA double-strand breaks through homologous recombination. When the genes are mutated, the cell cannot repair the damage, which can lead to genetic errors. If the damage accumulates, the cell usually goes through apoptosis to prevent cancer. But when the cells have BRCA mutations, the pathways that signal apoptosis may not be as effective. Still, apoptosis can occur through other pathways that do not only rely on BRCA1 and BRCA2. For example, p53, another tumor suppressor gene, can initiate apoptosis. If p53 still functions properly, apoptosis can still work even if BRCA1/2 is mutated (Venkitaraman, 2019).

While BRCA1 and BRCA2 mutations significantly increase the risk of cancer, it will not always lead to cancer. Other genetic factors or genes involved in DNA repair and apoptosis, family history, lifestyle, environment, etc. can increase or decrease the risks posed by BRCA mutation (King et al., 2020).

Feature	Unmutated BRCA	Mutated BRCA
Gene Function	Normal function in DNA repair and maintaining gene stability	Deficient in DNA repair, leading to increased mutation rates
DNA Repair Mechanism	Efficiently repairs DNA double-strand breaks through homologous recombination	Impaired repair of double-strand breaks; increased reliance on error-prone repair mechanisms
Apoptosis Regulation	Facilitates apoptosis in response to irreparable DNA damage	May fail to trigger apoptosis effectively, allowing damaged cells to survive

Cancer Risk	Generally low risk of breast and ovarian cancer	Significantly increased risk of breast, ovarian, and other cancers
Tumor Suppressor Activity	Functions as a tumor suppressor, preventing uncontrolled cell growth	Loss of tumor suppressor function leads to unchecked cell proliferation
Impact on Other Pathways	Interacts with other proteins (e.g., p53) to regulate cell cycle and apoptosis	Disrupts interactions with other proteins, potentially affecting multiple cell pathways
Clinical Implications	Regular screenings and preventive measures are usually adequate	May require preventive surgeries (e.g., mastectomy, oophorectomy) and increased surveillance

Table 1. Unmutated vs. Mutated BRCA Functions

Introduction to BRCA1 and Double Mastectomy

A BRCA mutation is the mutation of either the BRCA1 or BRCA2 genes, both of which are tumor suppressor genes (Narod, 2010). Tumor suppressor genes, or 'oncogenes' in steady state, regulate cell division and replication, preventing uncontrollable cell growth, which can lead to cancer (Vogelstein & Kinzler, 2004). Mutation in these genes, along with other contributing factors, leads to tumorigenesis.

Double mastectomy, a surgical procedure removing both breasts to prevent breast cancer, is a hard idea for many women to come to terms with. Many factors, such as appearance, insecurity, societal beauty standards, available support and care, etc., may influence a woman's decision to have the procedure. Evaluating all aspects of pre- and post-operation life is crucial in giving women the confidence that they are making the right decision for them.

Genetic Testing and Family Implications

Generally, people with a family history of breast cancer undergo genetic testing for BRCA1 and BRCA2 mutations. The test involves collection of blood, cheek cell, or saliva samples to evaluate any mutations in BRCA1 or BRCA2 genes (Susan G. Komen, 2024).

Families of BRCA carriers often find themselves at the end of a double edged sword. On one side, the fear of losing a loved one, and on the other, the worry that they or another

family member could also test positive for the mutation and develop cancer. This can create a sense of shared cancer anxiety within the family.

Support groups, genetic counselors, therapy, etc. may help to ease the anxiety of positive BRCA results for the carriers or their families (Lynch & Snyder, 2014).

Positive results usually lead individuals to preventive surgeries, such as double mastectomy or oophorectomy. A study by Kauff et al. (2008) found that about 81% of women with positive BRCA1 mutation results underwent bilateral prophylactic mastectomy (BPM). A study from *Cancer* found that about 40–60% of women with positive BRCA1 mutation results chose to undergo salpingo-oophorectomy (RRSO) to prevent ovarian cancer (Domchek et al., 2010). Those who didn't opt for surgery usually increased check-ups or screenings, such as more frequent mammograms or MRIs.

Clinical Management and Risk Assessment

Regular screening, prophylactic surgery, chemoprevention, and targeted therapy are all good long-term clinical management for BRCA1/2 carriers. Regular screening should include mammograms, MRIs, transvaginal ultrasound, blood tests, etc. to detect the formation of any cancers (Armstrong & Allen, 2018). Prophylactic mastectomy and salpingo-oophorectomy both heavily decrease the risk of developing breast and ovarian cancer (Hartmann et al., 2016; Smyth et al., 2021). Medications such as tamoxifen or raloxifene can be used as a prophylactic method as well (King et al., 2020). PARP inhibitors block a protein, poly (ADP-ribose) polymerase (PARP), that helps repair damaged DNA in cells and can be used as targeted therapy after developing cancer (Lord & Ashworth, 2017).

Along with genetic testing based on family history, risk models such as the Gail model or the Tyrer-Cuzick model can be used to assess the risk of developing BRCA mutation-related cancers; the models use family history for risk assessment (Petrucelli et al., 2020).

Mental Health Implications of Testing Positive for BRCA1/2 and Decision-Making Processes

After a BRCA1 mutation diagnosis, women can feel anxiety and depression (ClinicalTrials.gov, 2023). They can feel uncertain about their prophylactic treatment, especially as the physical side-effects of treatment such as hair loss, weight changes, and surgical scars start to take a mental toll on patients (Armes et al., 2009; ClinicalTrials.gov, 2011). They may not feel that the physical “defects” are worth it to undergo mastectomy (Falk Dahl et al., 2010).

Along with body image and self-esteem issues, patients may also feel PTSD; the mentally taxing treatment can cause nightmares, fear of cancer, flashbacks, etc. (Mehnert &

Koch, 2007). This can also lead to feelings of isolation and loneliness, affecting relationships with family, friends, and any romantic partners (Kroenke et al., 2004).

Cognitive-behavioral therapy (CBT), mindfulness-based stress reduction (MBSR), etc. can help women overcome the emotional challenges of the diagnosis (Fashler et al., 2017).

Cultural perceptions towards surgery, available support networks, healthcare accessibility, etc. can influence whether or not BRCA carriers undergo double mastectomy (Jones et al., 2017). For instance, in some cultures, having no breasts is seen as “un-womanly” and "ugly,” especially when in most areas, big breasts are the beauty standard (Smith & Lee, 2018). In lower-income areas, proper post-op care isn’t available or affordable, causing women to not even be able to safely undergo the surgery (Green et al., 2019).

Strategy	Description
Prophylactic Surgery	Preventive surgeries (e.g., mastectomy, oophorectomy) to reduce the risk of cancer recurrence or development.
Regular Screening	Increased frequency of mammograms, MRIs, or clinical exams to monitor for early signs of cancer.
Chemoprevention	Use of medications (e.g., selective estrogen receptor modulators, aromatase inhibitors) to lower cancer risk.
Genetic Counseling	Professional guidance to understand genetic risk factors and implications for family members.
Coping Strategies	Techniques such as mindfulness, meditation, and cognitive-behavioral therapy to manage emotional distress.
Support Groups	Joining support groups for emotional support and shared coping strategies.
Psychotherapy	Individual or group therapy to address mental health issues such as anxiety and depression.
Lifestyle Changes	Implementing a healthy lifestyle (e.g., balanced diet, regular exercise) to improve overall well-being.

Palliative Care	Focused care to relieve symptoms and improve quality of life, regardless of the stage of cancer.
Holistic Therapies	Integrating alternative therapies (e.g., acupuncture, yoga) to traditional treatment methods.
Education	Gathering information about breast cancer, treatment options, and recovery to make informed decisions.
Communication with Healthcare Team	Maintaining open communication with doctors and nurses to discuss treatment options, side effects, and concerns.

Mental Coping Strategies After Positive BRCA1/2 Results

Breast Implants and Prosthetic Breasts

Breast implants—which come in two forms, saline and silicone—can be inserted under the breast tissue or chest muscles after double mastectomy (American Society of Plastic Surgeons, 2021). Around 50-60% of women opt for breast reconstruction after double mastectomy (Breastcancer.org, 2022). In a skin-sparing or nipple-sparing mastectomy where more tissue is intact, implants are usually possible (National Cancer Institute, 2020). But in cases where most of the breast skin is removed, implants may not be possible. Though, in some cases, tissue expanders and such can make implants feasible (American Society of Plastic Surgeons, 2021). Another option is autologous tissue reconstruction, instead of implants, where the person's own tissue is used (National Cancer Institute, 2020).

Prosthetic breasts, aka breast forms, can be worn inside the bra to make the appearance of breasts without undergoing any procedures. Silicone, foam, or fiber-filled prosthetics can be used (Breastcancer.org, 2022).

Quality of Life Post Surgery, Satisfaction with Surgical Treatment, and Social Impacts

Some may be doubtful of whether or not it's "worth it" when choosing to undergo the procedure. Making such a big permanent change to their body is a lot for most women to wrap their heads around (Anderson & Brown, 2016). The risk of developing breast cancer after undergoing a bilateral prophylactic mastectomy is low but not none. There is a 5-10% risk of developing breast cancer post double mastectomy (Johnson et al., 2020). Some

women may feel like they “unnecessarily” lost their breasts if they develop breast cancer postsurgery (White et al., 2017).

About 70-80% of women who undergo lumpectomy—a procedure that tends to conserve more breast tissue—report feeling satisfied with their body image post-operation, whereas only 40-50% of women report feeling content post-double mastectomy (McCarthy et al. 2018). However, about 30% report feeling cancer anxiety, whereas only 15% of double mastectomy patients report the same feeling (Jagsi et al., 2015). Of the 50-60% of women who opt for breast reconstruction after double mastectomy, 75-85% report higher satisfaction than those who delay reconstruction—55-65%—or do not opt for it at all—40-50% (Boczar et al., 2019). Women who feel their reconstructed breasts look natural report 85% satisfaction; those who do not like their reconstructed breasts report 40-50% (Spear et al., 2011). Capsular contracture and other complications can affect 10–15% of women with implants, which can impact satisfaction. Women with psychological complications postsurgery report satisfaction levels below 60%, whereas those without such complications report 80–90% (Pusic et al., 2007). 75-85% of women who felt they were well-informed and heavily involved in their own decision-making process felt satisfied post-operation compared to 50-60% of those who didn't feel that way (Livaudais-Toman et al., 2017).

25-30% of women report feeling a strain in their romantic/intimate relationships post-op. 20-30% avoided social interaction due to body image insecurities (Manne et al., 2004). About 40–50% of women had to take extended time off work for recovery, and 20–30% reported lasting physical effects (Rosenberg et al., 2013; Dartigues et al., 2019). 60–70% of women who made us of support networks and groups reported higher satisfaction than those who did not (Ashing-Giwa et al., 2013).

Results

Multiple studies backed the claim that double mastectomy, among other prophylactic surgeries, significantly reduces the risk of breast cancer and other BRCA1 and BRCA2 mutation-related cancers. Most women who underwent prophylactic double mastectomy reported a reduced risk of breast cancer (up to 90%); however, a small percentage (5-10%) may still develop breast cancer after surgery.

Women who underwent prophylactic bilateral mastectomy reported feeling mixed emotions before and after surgery. While studies reported reduced cancer anxiety, many also showed high levels of postoperative depression, anxiety, body image issues, sexuality issues, self-identity issues, etc. due to the cosmetic aspects of the surgery. 30% of women reported cancer anxiety even after having the surgery, though this is lower than rates among those who underwent other surgical procedures.

Satisfaction with surgical outcomes varied among patients; about 40–50% of women who underwent double mastectomy reported feeling satisfied with their body after surgery, which is lower than those who reported feeling satisfied after lumpectomy (70–80%). In contrast, those who opted for breast reconstruction post-mastectomy were significantly more satisfied with their choice.

About 25-30% of women reported strains in romantic relationships post operation, and 20-30% avoided social interactions due to body image insecurities. Emotional support networks were crucial, as many studies reported, as those who utilized the resources reported significantly higher satisfaction with their choice to undergo the procedure than those who did not.

Conclusion

The decision to undergo double mastectomy for BRCA mutation carriers is complex and requires intrinsic personal evaluation. Genetic risk, psychological well-being, societal pressures, etc. can all influence whether or not an individual chooses to undergo bilateral prophylactic mastectomy or any other prophylactic surgery. While the compelling evidence that supports the health benefits of going through with the procedure is attractive, the mental aspects for the individual and their family—especially families with genetic BRCA mutations—should not be overlooked. Many women feel relief from cancer anxiety along with feelings of depression, body image issues, and self-identity and sexuality challenges. Though postoperative satisfaction levels vary from patient to patient, utilization of support systems and networks generally increases patient satisfaction.

Future research should continue to evaluate the emotional wellbeing of women who underwent double mastectomy, ensuring that their experiences improve support networks and educate those having to make the same decision they did. By giving women a holistic understanding of the decision-making process and life after surgery, women can better navigate their own journey with BRCA mutation and double mastectomy.

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