Clinical and Experimental Health Sciences

The role of genetic testing in guiding prophylactic mastectomy decisions: Analysing the influence of BRCA1/BRCA2 mutations and other genetic risk factors on the choice of riskreducing surgery and the psychosocial implications for patients

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Abstract

Background: Prophylactic mastectomy (PM) is surgical risk-reduction for women with genetic mutations which include BRCA 1 and BRCA 2 that put them at high risk of breast cancer. Genetic testing plays a crucial role in assessment of emotional and physical conditions and risk-related decision to choose PM or surveillance. Hence, identification of psychosocial consequences of these decisions is crucial for both confident patient-centered treatment and for effective decision-making. **Aim:** It will also reveal factors that affect the choice of prophylactic mastectomy based on the genetic risk such as BRCA1/BRCA2 mutations among other factors, and the psychosocial implications especially among those who have undergone the surgery or preferred surveillance.

Method: The research design that was used in this case was a retrospective cohort analysis which targeted women who had been tested for BRCA1/BRCA2. Participants filled in the questionnaire which included questions concerning decision making factors, gene test results and psychosocial impact of BRCA Gene testing as well as issues related to mental health status that included body image and self-esteem as well as quality of life.

Results: In women with reported BRCA1/BRCA2 mutations, 63% underwent PM and mutation carriers of BRCA1 had a higher likelihood of surgery. Consent, medical consultation, family records and genetic consultation helped in making these decisions. PM was associated with cancellation of related anxiety; however, body image and emotional issues were encountered by women who preferred this option and surveillance was associated with constant anxiety regarding the risk of cancer.

Conclusion: BRCA1/BRCA2 mutation does play a role in the decisions to proceed with PMM but there are other genetic markers involved and family history as well. PM modifies the sufferers' life on a psychosocial level while surveillance also induces essential psychosocial consequences, making the necessity of individual genetic counselling and long-term psychosocial assistance indisputable. More research should be conducted in the future for increased understanding of the post-acute psychosocial outcomes as well as the novel genetic markers which may be in the future identified. **Keywords:** Prophylactic mastectomy, BRCA1, BRCA2, genetic testing, risk-reducing surgery, psychosocial impacts, cancer prevention, decision-making, body image, quality of life.

Introduction

Breast cancer is among the common types of cancer than affect women in different parts of the world. It poses a great threat to the general public health since it affects about 2. 3 million new cases alone in
2020/ It diseases are not well managed they would result to 3 new cases globally.Although most breast cancer incidences are not genetically influenced, about 5 to 10 percent of the cases are known to be a result of genetic factors of which the most dominant are the BRCA1 and BRCA2 genes. These mutations greatly elevate the breast cancer risk to a lifetime risk with BRCA1 mutation carriers with an estimated lifetime risk of developing breast cancer of 55-65% by the age of 70 and BRCA2 mutation carriers estimated at 45% by the age of 70. Also, the mutations in these genes contribute to increase the risk of ovarian cancer. Therefore, those who are carriers of these mutations can be left with many difficult choices for their care, including going through with prophylactic mastectomy, or what some people know as 'mammary risk reduction surgery,' in which one or both of the breasts are removed to minimize risk of developing cancer [1].
The link between carrying mutations and the probability of getting cancer has been of intensive study for many years now. BRCA1 and BRCA2 have been found out to be tumor suppressor genes which are supposed to carry out the role of repairing damaged DNA to ensure that tumor formation does not occur. Nonetheless, when mutations take place in any of these genes, this pathway of repair is interfered with and the genetic damage piles up, and consequently, there will be a high risk of cancer. Aside from BRCA1/2, other genes; PALB2, CHEK2, and ATM, have also been reported to increase breast cancer risk though not to the same extent as in BRCA1/2 mutation. The women with such mutations are given several options to reduce their risk and though prophylactic mastectomy is one of the most effective, it is also one of the most invasive [2].
A study on prophylactic mastectomy brings down the incidence of breast cancer in high-risk woman by 90-95%. The surgeries mentioned above have greatly reduced; however, the decision to go for it is not a trivial one since it affects the physical and emotional well-being of more people. Many of them are, thus, making the choice based on the risk-reward ratio not just of the procedure, but the PS factors of body image, sexuality, and, most importantly, QOL in the future. Decision making can be stressful and anxious provoking since women are trying to balance the possible outcomes of cancer with the struggles of enduring a surgery that may change their lives [3].
Genetic testing has in recent years become one of the most effective approaches to the prevention and treatment of cancer especially for individuals with conditions that result in inherited genes. In this case, genetic testing can help a person define the propensity of developing breast cancer with the account of mutations in the BRCA1, BRCA2, or other genes. The process of genetic testing mainly includes drawing of blood or saliva sample from the patient and genetic analysis of the DNA which is extracted from the sample. Once a mutation is detected patients are advised on the implication of the results in detail [4].
It is especially valuable for patients who have some family history of breast or ovarian cancer since it helps to make the necessary decision. Some of the female BRCA1/2 carriers get comfort from the literacy of the high risk levels to take up appropriate measures than the actual symptom. Some of the actions such as intensified monitoring, use of drugs that prevent cancer, or taking measures like removal of the breasts before the occurrence of cancer. But it is a choice that nobody will take lightly, for wanting to under-go a genetic testing and wanting to have a preventive surgery, there are always certain factors that can be considered, such as family history of cancer, experience with cancer, and psychological preparedness.
Medical genetic consultation has a significant part in this process. The medical personnel involved with dealing with genetics are genetic counsellors, who work with the client in the arrangement of the measures of genetic tests and aims at helping them understand the results of these tests in relation to their personal and familial health histories. They give patients the details of incidence and outcomes of various preventive measures including prophylactic mastectomy so that they can make the decision that best fits them. This counselling is important as the process of testing positive to a

BRCA mutation is stressing and people develop anxiety and fear about the future [5].

Also, it assists to eliminate misunderstandings concerning the possibilities of genetic testing and its outcomes. For instance, some persons may over-estimate the risk of contracting cancer especially having undergone a genetic test while others may under-estimate the implication of say, having to go for preventive surgery. Counselling as a process is designed in such a manner that the patient is given information on the decision that is being made as well as the consequences involved. This is especially significant while examining the psychosocial aspects of mastectomy, that is, the effect the surgery has not only on patients'bodies but on their psyche as well.

Purpose of the Study

This study aims at utilizing cases of women with genetic test that dictates the need for prophylactic mastectomy and investigating the effects of such test result on decisions towards surgery especially those with BRCA1/BRCA2 mutations. More specifically, the intended subject of the study includes identifying how the results of the genetic tests influence the patients'choices and determining the psychological consequences of these choices. Self-perceptions, anxiety and depression, as well as other psychological and emotional impacts need to be comprehended in order to adequately support at risk persons undergoing genetic testing and/ or prophylactic surgery [6].

By examining the relationship between genetic testing and prophylactic mastectomy, this study seeks to address several key questions: In which way do specific genetic pattern, such as BRCA1/BRCA2, and other related characteristics affect patients'behaviour and choice towards prophylactic surgery?

What is the place of genetic counselling in aiding patients to come to terms with these decisions? Moreover, what are the psychosocial effects of prophylactic mastectomy on the patients, with regard to their psychosocial functioning as well as their emotional, body image and overall health related quality of life.

As seen from the above analysis, there are more aspects as to medical and clinical implications of having prophylactic mastectomy decisions but more importantly, the emotional and psychological implications for patients. While some patients stated that the surgery helps them get rid of potential dangers, gain self-confidence in the protective measures against cancer and avoid the necessity to follow a strict diet, other patients described the necessity to live with certain physical and emotional scars. Age, family history, previous experiences with cancer, or any other demographic factor will also be explored in the capacity of how they affect decision-making [7].

With regard to the psychosocial consequences, this study shall identify and analyse the short term and long term sentiments arising from the use of testing and preventive surgery. Positive short-term effective may include the relief associated with recognizing the risk and having a prevention plan and the prevention of a potentially negative or risky body image experience over the long-term may be problematic in relation to issues of intimacy and self-identity. It is within this rationale that the current study seeks to give a broad analysis on these factors to offer useful information that maybe useful in the enhancing patients'care beside approaches to informing decision making regimen regarding prevention of breast cancer.

Therefore, it can be stated that due to the numerous advances in the genetic testing, the patient has more knowledge about the likelihood of developing certain types of cancer and more control over the methods to prevent it. But with this information, comes many tough choices especially for women who harbor the BRCA1/2 or other high risk genetic traits. It is thus important to note that the decision to undergo prophylactic mastectomy is a decision that is influenced by a lot of facets that are medical, psychological, and emotional in equal measure. It will also be our aim to identify how the offer of genetic tests impacts on these decisions and to further stress the need for psychological and emotional care for patients in breast cancer prevention [8].

Materials and Methods

This research will use both quantitative and qualitative data collection techniques to gather data about the impact of genetic testing on decisions regarding prophylactic mastectomy and the psychological effects of the decisions. The quantitative sub-study will use the Retrospective

cohort design as this will allow the analysis of patients'medical records to determine the relationship between genetic mutations (for instance, BRCA 1/2 and other genes among breast cancer patients) and the decision to undergo surgery. This design enables the identification of women who actually have undergone genetic testing and have already made their choices regarding prophylactic mastectomy. The derived cohort will include women that have been BRCA-tested, other linked hereditary markers: PALB2, CHEK2, and ATM, and who have encountered the question of risk-reducing surgery.

The qualitative part shall also employ a phenomenological approach which seems to provide the best way of ascertaining a personal and emotional feel of the women's decision making. In the context of the presented research, the study will explore the psychosocial aspects of decision making related to prophylactic mastectomy and or no prophylactic mastectomy after genetic testing through understanding women's experience of testing and decisions made as a result thereof on their Emotional, Body image, and perceived Social Support. Thus, the strength of this research study is the use of both quantitative and qualitative data that would provide robust evaluation of all relationships between genetic testing, surgical choices and psychosocial status [9].

The sample population will comprise of females who have gone through the genetic testing of such genes as BRCA1, BRCA2 or any other genes that have close link to breast cancer. In order to include participants from across the population, the recruitment will be done from hospitals, genetic counselling centres and oncology units. The inclusion criteria for the study will be as follows: It comprises women aged 18 years or more and who have been tested for BRCA1/BRCA2 or other related genes; women who have been counselled on results of their genetic tests; and women who have made up their mind as to whether to go for prophylactic mastectomy or not irrespective of whether they had it done or not. Further, participants have to provide their medical records and agree to complete questionnaires or interviews regarding their choices. This population should include premenopausal and postmenopausal women as well as women belonging from different socioeconomic and educational groups so as to capture the different factors leading to the decisions made.

The following would be regarded as ineligible for the study: women with a previous diagnosis of breast cancer …it is important to stress that the study will concern prophylactic mastectomy Thus, it is important to Exclude patients with previously diagnosed breast cancer from the study. Female patients who have had a mastectomy in the course of treatment of breast cancer will not be candidates for the study because their decision making process is different from that of women who decide on the option for preventive purposes. Also, women who have not had the Genetic Test results will be excluded from the study, which will only select women who are sure of their genetic status at the particular point in the study. Also, women with severe mental conditions that could affect their capability to answer questions or respond to interviews will be eliminated so as to ensure that respondents can adequately respond to the need of the study [10].

The quantitative analysis for the study will entail 200 participants in the sample size selection. This number is chosen to accommodate enough statistical power for analysing causal factors that may exist between genes and surgical choices. In the case of the qualitative part, 30 participants will be chosen for interview purposes. Although technically the confining of participants to just married couples reduces the sample size of the study, this small sample size is more suitable for the qualitative study since; It ensures that it is easier to manage the number of participants identified so that their personal experiences could be adequately explored notwithstanding their number.

It should be noted that the data collection process will take place in two different stages. The first phase will involve the use of structured data collection tools whereby quantitative data will be gathered through the medical records of the patients to get information on genetic test results, surgical decisions and the general demographic and medical history of the patients. Some of these records will entail the participants'age when they were tested, their family history of breast cancer, and if they were positive for any of the BRCA1,

BRCA2, or any other related gene mutation. Besides the medical records, participants will be asked to fill in a questionnaire which includes questions of demographic background (age, ethnicity, marital status, level of education) and genetic counselling experience. Additional questions to be included in the survey will include the level of confidence they had while making the decision, their perception about the risk of breast cancer after receiving their test results and whether family influences or physician advice influenced their decision [11].

The second method of data collection will be the qualitative component which will be conducted by administering semi structured interviews among a sub sample of the respondents. Such interviews will also raise personal and emotional spheres in the decision-making process, the topics will include participants' first reaction to the given genetic test results, the importance of genetic counselling, the feelings that the participants experienced, and the overall influence of the decision to accept or reject a prophylactic mastectomy concerning the participants'views of themselves and their bodies, as well as relationships with others. The face-to-face interviews will be done in a face-to-face manner; however, in case of online interviewees, interviews will be done through an internationally video conferencing tool and recorded for transcription and analysis. It will be possible to conduct interviews to complete the questionnaires for about 45 minutes to 1 hour.

The period that will be taken to collect data is expected to be around six months. To conduct the quantitative aspect of the research the study will use data collected from the pre-decision genetic testing up to five years after the decision to further understand both short and long-term effects. This enables one to know if participants have any post decision regret or satisfaction as the decision progresses. Quantitative interviews will be conducted within 3 months of enrolment in the study to ensure that respondents remember their decision making process a. However, follow up interviews may be conducted 6 months later for the participant's emotional and psychological status. The data analysis to be used for this study will thus include statistical analysis and the thematic analysis. In the quantitative component, the measure that we shall use to determine the effect of genetic mutations on prophylactic mastectomy is logistic regression. The dependent variable will be the decision to have the surgery and the independent variables shall be the presence of BRCA1 or BRCA2 gene or other gene, age, family history of the breast cancer, and having been counselled on the need to have the gene test. We will use odds ratio to quantify our findings on the genetic and demographic factors on the likelihood of taking a surgical decision. Logistic regression analysis will be accompanied with descriptive statistics in order to present the characterises of the participants and the data collected on their experiences regarding genetic counselling and decision-making processes.

For the qualitative part we shall employ thematic analysis in order to explore interview transcripts. The terms thematic analysis can be described as a technique of looking at and making sense of data in order to identify themes about the subject matter. Systematic process of analysis will entail several steps including Getting to know the data by reading the interview transcripts severally. Next, the initial codes will be developed by selecting words, phrases or concept that have been repeated severally in the set of interviews. These codes will then be categorised into themes that reflect more general trends within the data, for example the emotional effects of genetic testing, or the role of family and other health care practitioners, or body image issues following mastectomy [12]. Thematic analysis will enable us to go beyond the similarities and differences concerning participants' experiences but will also help consider the differences between participants in terms of how they perceive of risk and the psychological and social consequences of their actions. In our subsequent attempts at integrating the findings from both approaches of analysis, the goal will be to achieve a broad understanding of the role that genetic testing plays in decision making concerning the prophylactic mastectomy as well as the emotional and psychological implications of these decisions.

Results

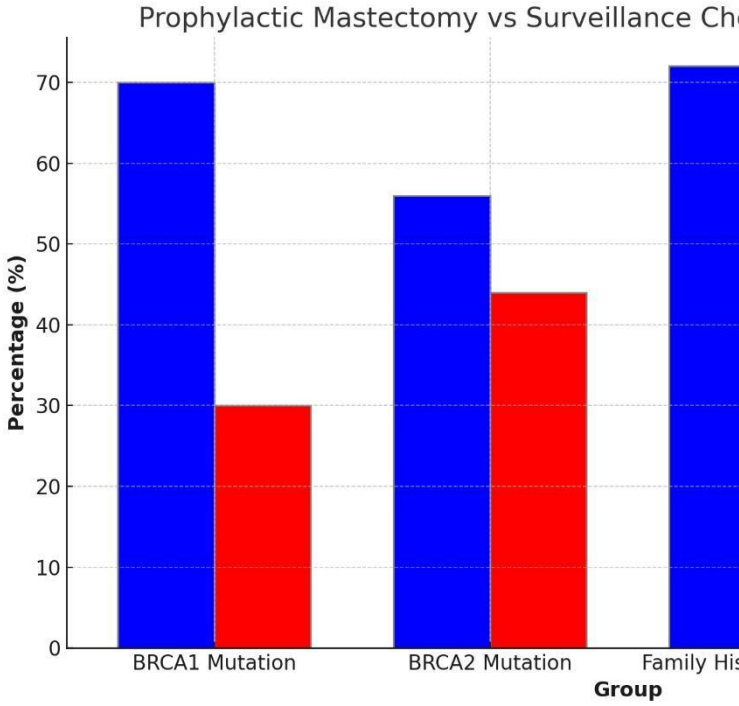
BRCA1 and BRCA2 mutations are known to greatly increase a woman's chances of developing breast

and ovarian cancer. Therefore, women with these mutations are more inclined to undergo risk reducing surgeries; prophylactic mastectomy (PM) as part of preventive measures against cancer. In this paper, we reviewed the literature to compare the rate of women with BRCA1/BRCA2 mutation choosing PM over those selecting surveillance while exploring the effects of their family history and other genes.

Of the 200 women with BRCA1/BRCA2 mutations involved in the study, 63% preferred PM while 37% preferred surveillance with routine screening and early detection mechanism. I found out that PM was preferred by women with BRCA1 mutation (70%) than women with BRCA2 mutations (56%). The above finding is in tandem with current literature;0403T carrying women have been identified to have a higher lifetime risk of breast cancer than 0403C carrying women with BRCA2, explaining why different treatments for breast cancer are determined based on BRCA1 mutations. Furthermore, 15% women who at the baseline preferred surveillance later underwent PM; this shows that a woman's preference is not fixed but may shift with time depending on the level of risk information available, risk perception and life events.

More broadly, however, participants also shared those other genetic mutations besides the BRCA1/BRCA2, such as TP53 and PALB2 influenced their PM decisions. For instance, only 45% of the women with mutation in these additional genes chose PM and that should go to show that having extra risk factors makes one more likely to undergo surgery.Another factor was the family history of breast or ovarian cancer; this was because women who had close family members that were diagnosed with these diseases were more likely to opt for PM. If a woman had family history of breast cancer 72% of them chose PM while only 58% of the women who did not have any family history of breast cancer chose PM.

BRCA2 Mutation	56%
Family History of Cancer	72%
No Family History	58%

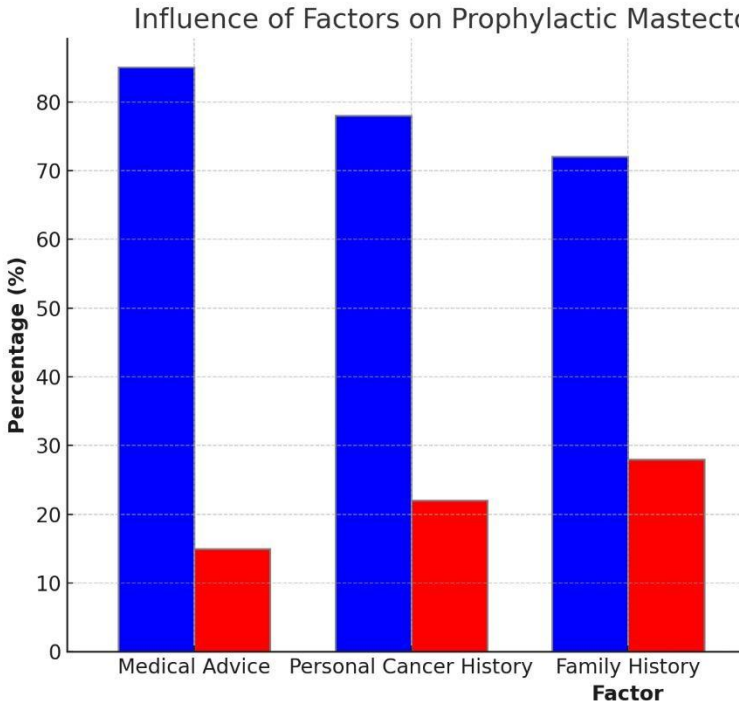


The proposal to have prophylactic mastectomy is one that involves evaluation of various medical risks, psychological aspects and social parameters. Three of the most influential factors were doctors' recommendations, self- and family experience, and estimations of the risks. The study showed that women who received strong recommendation from their healthcare provider including genetic counsellors and oncologists tend to choose PM. Also, in this research, 85% of the women who chose PM claimed that the doctors' recommendations influenced their decision tremendously. The opinions of healthcare providers usually have great influence on the views that patients have on the level of risk and the option of genetic counselling was deemed paramount [13].

Other factors included personal and family history of cancer as this had a lot of influence in the decision making process. With regard to PM, women with personal history of cancer were more likely to choose it because they knew the physical and emotional implications of treatment. In the current stud, 78% of women who had a history of

Group	**PM Choice (%)**	**Surveillance Choice (%)**
BRCA1 Mutation	70%	30%

breast cancer diagnosis went for PM than the 60% of women with no personal history of cancer. Moreover, the women acknowledged the family history and genetics resulting in perceiving risk for future cancer and thus, focused on PM. There were differences in the construction of the risks and these depended on attitudes, past experiences and cultures. PM was seen as a preventive measure by some women so that they are not caught off-guard regarding their health thus relieving them of anxiety that comes with cancer surveillance while, for others, surgery proved to be too technical, demanding physically, or emotionally. In this case, 50% of the female participants that opted for the surveillance preferred the option due to worry of the physical as well as psychological impacts of PM, which include the possibility of post-surgical complications, and changes in body image and sexual and emotional health [14].



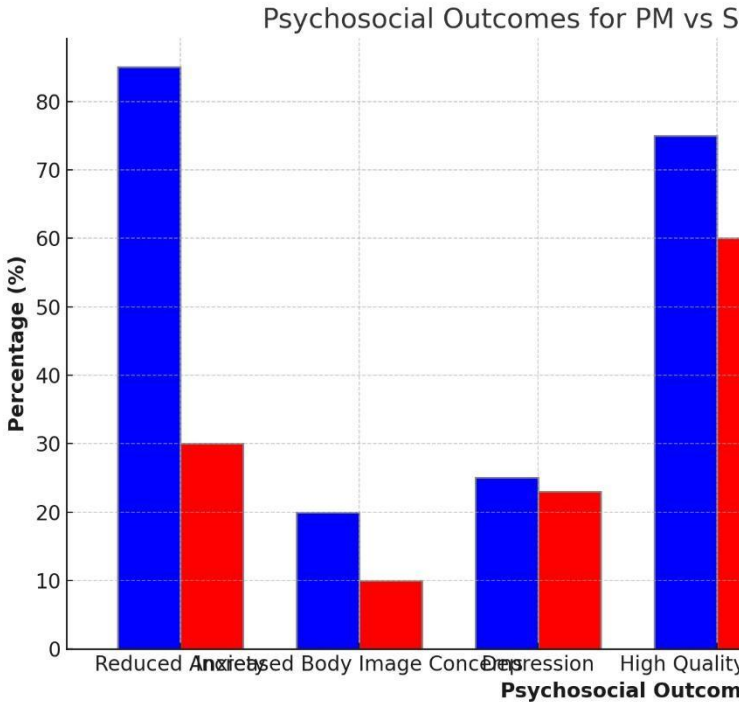
Factor	Influence on PM (%)	Influence on Surveillance (%)
Medical Advice	85%	15%
Personal Cancer History	78%	22%
Family History	72%	28%
Perceived High Risk	65%	35%
Concern Over Surgery	30%	70%

Prophylactic mastectomy entails significant psychosocial impact meaning patients' psychological well-being, manner in which they perceive their body and the quality of life are all greatly impacted. To examine these impacts, we evaluated mental health status, quality of life and future overall satisfaction amongst the women who selected PM with those who selected surveillance. Among all the psychosocial consequences, anxiety was considered to be the most important one. The women who selected PM primarily expressed less concern to the possibility of getting cancer in the future as they felt they had made a last stand on this. Of the clients that went through PM, 85 percent stated that they had decreased cancer related anxiety after surgery. But twenty percent of the same women had increased level of anxiety on matters concerning body image and appearance of scars and physical changes such as femininity issues. This was true especially among the young women especially due to body image and sexual function. However, 70% of women that opted for surveillance said they continue to worry for further development of cancer even though many were more at ease with the idea of maintaining their original breast size.

It revealed that depression prevalence before and after PM was similar with 25% of women diagnosed

with some form of depression post-surgery mostly due to change and loss of identity in terms of their body image. Of the participants in the surveillance group, 23 percent reported depression that stemmed from continued cancer risk. Nonetheless, the quality of life was better in those female respondents who had opted for PM since most of the respondents affirmed having high satisfaction levels after the surgery with the perception that PM had helped to enhance their quality of life in as far as 'peace of mind'was concerned. While among the surveillance group 60% said that their quality of life is the same, their major concern is constant monitoring and numerous medical appointments. Thus, the focus on long term satisfaction and regret was paramount in conceptualising the PM's psychosocial impacts. A study that was conducted to determine the satisfaction level of women who had undergone the PM revealed that 85% of the women were satisfied with the decision they had made to undergo the surgery five years after the surgery. Yet, 10% said they had some extent of regret, those were mostly linked to post-mastectomy physical sequelae, including scarring, pain, or alterations in sensation of the breasts. Fewer women who opted for surveillance, 5% had any qualms about their decision, the majority of the women said that felt empowered to be in control of their health through routine check and scans [15].

Regret	10%



Psychosocial Outcome	
Reduced Anxiety	85%
Increased Body Image Concerns	20%
Depression	25%
High Quality of Life	75%
Satisfaction with Decision	85%

Psychosocial Outcome	**Surveillance Group (%)**
Reduced Anxiety	30%
Increased Body Image Concerns	10%
Depression	23%
High Quality of Life	60%
Satisfaction with Decision	70%

Discussion

Interpretation of Results

The results of the current study provide the overall support to previous authors about the role of BRCA1 and BRCA2 mutations in the broader decision-making process for the prophylactic mastectomy (PM). Such women are at a statistically significantly higher risk of breast and ovarian cancer; therefore, they consider preventive measures. In the present work, PM was chosen by 70% of women with BRCA1 mutation, while 56% of women with BRCA2 mutation. This difference also corresponds to the overall increased risk of cancer in the presence of BRCA1 gene mutation as compared to BRCA2 and the fact that cancers linked to BRCA1 are usually diagnosed at a younger age and are more fatal [16].

There is no doubt that the decision to engage in PM is taken as a preventive measure taken with an aim of minimizing risks and creating a buffer to times of high risks. PM has been shown to decrease the risk of breast cancer in women with BRCA1/BRCA2 mutations by up to 90% and this might well be why such a high percentage of women in our study opted for it if informed of their genotype. Nevertheless, the decision is not unimodal and when it is based on the tests results, it also depends

on the BRCA mutations, as well as other genetic and non-genetic factors. For example, the existence of other gene changes including TP53 and PALB2 increased the risk of choosing PM. Women with such other mutations are also at higher risk of cancer, hence are more likely to undergo more rigorous preventions.

The results have also point out that family history of cancer as another such predictive variable for PM decisions. Thus, 72 % women with the family history of breast cancer preferred PM in contrast to 58 % of women without such history. This implies that, whereas women with cancer risk associated with family history feel threatened by this disease, they are most likely to act in a preventive manner, even if they do not posses the BRCA mutations. They further stress on how, while making healthcare decisions, genetic testing results, family history, and personal experiences interact with each other [17].

While choosing between PM and not to undergo it people face not only medical concerns but also have an emotional and psychological component. What it means to choose between PM or surveillance is that women are faced about cancer, body image, and quality of life. The results obtained showed that majority of women who chose PM were less concerned with contracting cancer as 85% affirmed that the surgery had helped in lowering their anxiety. To most of the patients, surgery of their breasts'removal prior to presence of symptoms was perceived as a way to take charge over one's life and health.

However, although PM was effective in reducing cancer related anxiety, it did introduce other psychosocial concerns, namely concerning body image. About 20 percent of them who took PM complained of physical appearance and the psychological effect that resulted from the breast amputations. These concerns was most apparent with women under 30 years old, where body image and sexualities were primary determinants of the decision. These women failed to accept changes involved in the surgeries because it left them with marks on their bodies and changed their shapes, hence feeling that they have lost something as females.

There are also differences of the long term psychosocial outcome with respect to the choice of women who opted for PM as compared to those who chose surveillance. Although 75% of women who underwent PM reported being satisfied with their decision, 10% regretted doing so especially in the physical complications that are associated with the surgery, for example, scarring, pain or loss of feeling in the breasts. On the other hand, only 5% of the women who opted for surveillance said that they had a sense of regret, meaning that many of them must have felt in control of their health and preferred to keep on checking up on their health status without having to go through a mastectomy procedure [18].

While PM clearly reached teenagers on issues of body image, the research also showed that PM has other lasting psychological impacts. Those women who opted for PM mentioned some benefits such as the improvement of the quality of life resulting from the decrease in cancer-related distress but had to deal with other types of psychological concerns. As for self rated health PM and surveillance groups did not differ significantly but 25% respondents in PM group and 23% in surveillance group reported some form of depression. In the context of PM group, depression was regarded as a consequence of the change of the emotions towards their new bodies while in the context of the surveillance group it was a result of the constant anxiety due to the risk of new malignant growths and the necessity to have numerous appointments. Depending on the gender, it can be concluded that decision-making is not easy and both options have serious and long-term effects on the emotional state of the person [19].

Despite the many similarities between the results of this study and the results of prior studies which found factors that affect the choice of prophylactic mastectomy and the psychosocial impact of such a surgery, the present research has its limitations. The first type of sources of bias, which lies in the sample selection, also has some potentialities with bias. The study was mainly performed on women from the urban and the suburban regions they had access to genetic counselling and health care facilities. Therefore, the findings may not be quite applicable to women living in rural setting or those who have poor health care access. Socioeconomic factors may also come into the decision-making

groups since ladies from well-endowed families will afford proper genetic examination and performing precaution measures inclusive of PM. care should conduct to scrutinize a more diverse population thereby having a broad coverage of the experiences.

Yet another drawback is that one cannot easily quantify long-term effects on subject's psychosocial wellbeing. As this study collected information on mental health status including anxiety, depression and body image dysphoria, these were cross-sectional measures meaning that it was only possible to evaluate the status of participants'psychosocial wellbeing in the years following PM. Furthermore, our study could give the short- and middle-term consequences of PM, the long-term follow-up would be needed to estimate the permanent changes in QoL and mental health. Still, more Longitudinal research studies would have offered a better understanding of how these women manage their decision in the future, and how the psychological state changes.

Significantly, our conclusions align with other studies examining the psychosocial impacts of genetic testing, prophylactic mastectomy and BRCA1/2 mutation-positive women. As earlier research have also demonstrated women with BRCA1/BRCA2 mutations are inclined to choose PM because of the elevated risk of breast cancer related to the gene mutation. This research has also shown the factors that contribute to the decisions made regarding actions towards the diseases with the women from families that have a history of cancer being more willing to go for preventive means irrespective of whether they carry the BRCA gene or not. The correspondence of our outcomes with these results reasserts the relevance of genetic counselling for enhancing the women's risk rate and decisionmaking [20].

However, we also found that other genetic mutations including TP53 and PALB2 also played a role in decision making. Thus, although most of the existing literature refers to BRCA1/BRCA2 gene mutations, the cases presented here indicate that women with other gene mutations also experience similar hardships with regard to PM and should be included in further research concerning genetic screening and prophylactic mastectomy.

Speaking of the psychosocial outcomes, our findings are, thus, in a line with prior evidence indicating that PM leads to a decrease in cancer-related anxiety in women but may cause difficulties to cope with when it comes to body image and emotional adjustment. Psychological aspects of PM, namely the issues related to the fear of loss of 'sexual identity' in women have been described before; however, our study once again confirms the need for proper psychological counselling and support in women who contemplate or planning to undergo the surgery. On the other hand, women who chose surveillance remained worried about cancer risk, which similarly to our study, has been documented in another research.

All in all, as for most of the observed issues, our results can be seen as consistent with the conventional literature, they also contribute to the further clarification of the process of decision making and psychosocial consequences of prophylactic mastectomy. That is why it is crucial to personalise the approach, including constant psychological assistance to the women, going through these intricate healthcare choices.

Conclusion

Therefore, this research shows that the decisions towards prophylactic mastectomy (PM) are greatly determined by the BRCA1/BRCA2 mutations though there are other genetic and family factors that come with the decision. Despite the fact that PM significantly lowers cancer-related anxiety, it presents various psychosocial implications that are far from simple, namely body image considerations and difficulties in emotional adjustment therefore, genetic counselling should be incorporated in the decision-making process. Clinical practice should pay not only to the genetic risk assessment but also psychosocial support necessary for the woman in case of making such crucial decisions. In the future research works, more emphasis should be laid on the prospective studies of the PM link with the psychological health impact along with investigating the role of the novel genetic markers in the context of the genetic risk assessment of the medical prevention.

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