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Learning to Live with Uncertainty**

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The Experiences of Women Possessing a Genetic Predisposition to Developing Breast  
Cancer: Learning to Live With Uncertainty

A thesis submitted in partial fulfillment of the requirements for the degree of Master's of  
Arts in Education, Educational Counselling

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## Abstract

Genetic testing to determine whether a woman carries a BRCA1 or BRCA2 mutation, which may entail an up to 80% lifetime risk of developing breast cancer, is relatively recent. As there is an apparent void in the literature investigating the experiences of these women, and few qualitative studies of this nature exist, six women identified as carriers were invited to participate in semi-structured interviews. The grounded theory method was utilized and through findings emerged two core categories: *Context Following the Receipt of the Test Results* and *Living with the BRCA Genetic Mutation: An Uncertain Conclusion to an Unending Process*. Specifically, following the receipt of their test result, participants reported changes in their self-perception and perception of others. Over time, participants described living with the BRCA gene mutation and its medical consequences. Particularly significant findings consisted of participants feeling less like a woman, feeling stigmatized, and living with unending uncertainty.

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The Lived Experience of Women Possessing a Genetic Predisposition to Developing  
Breast Cancer: A Grounded Theory Study

**Background and Rationale for this Study**

Breast cancer is the most common malignancy affecting human females (Hartman et al., 2007). This disease consists of a malignant tumor, which starts from breast cells. The term malignancy signifies that the cancer cells have the ability to spread from the affected tissues to other areas of the body such as further organs or lymph nodes. While various types of breast cancer have been discovered, certain have been identified as more prevalent than others (American Cancer Society, Inc., 2007).

Although the incidence of breast cancer has decreased over the past few decades, it is still the most prominent cancer affecting Canadian women, with approximately 22,000 new diagnoses each year (Canadian Cancer Society, 2007). The Public Health Agency of Canada (PHAC) (2008) reports that in 2001, breast cancer represented about 19% of all new cases of cancer in women. While this disease does affect a small percentage of men, it is roughly one hundred times more common in women, with only approximately 1% of breast cancer cases being male (American Cancer Society, Inc., 2007; PHAC, 2008). Throughout her lifetime, a Canadian woman has a one in nine chance of developing breast cancer. Moreover, recent statistics demonstrate that one in twenty-seven women will eventually die from breast cancer in Canada, with about 5,000 women dying each year from this disease (Canadian Cancer Society, 2007). This evidence demonstrates that even in a country where advanced medical screening facilities exist in addition to advanced treatment options, breast cancer continues to have a

devastating impact on the female population (Canadian Cancer Society/National Cancer Institute of Canada, 2007).

### **Risk Factors**

Nearly 80% of women who develop breast cancer tend not to have a family history (Alberta Cancer Board and Foundation, 2007). Generally, the most common factor leading to breast cancer is that of being a woman and that of aging (particularly reaching the age of 50) (Canadian Cancer Society, 2007). The PHAC (2008) reports that over 75% of women who were diagnosed with breast cancer in 2001 were over the age of 50. Nevertheless, various other risk factors to developing this disease have been identified.

These risk factors consist of a previous family history of breast cancer, a personal history of breast cancer, race, age of onset of menarche, an abnormal breast biopsy, previous radiation (particularly of the chest area), and treatment with diethylstilbestrol (American Cancer Society, Inc., 2007).

Certain lifestyle risk factors also exist. These mainly consist of using birth control pills, having no children or giving birth later in life (after the age of 30), utilizing Post-Menopausal Hormone Therapy, not breast-feeding, consuming more than one glass of alcohol per day, being overweight or obese and not exercising (American Cancer Society, Inc., 2007).

While having certain risk factors increases a woman's chances of developing breast cancer, many women who possess these risk factors do not develop the disease. Conversely, many women who develop breast cancer possess no identifiable risk factors (American Cancer Society, Inc., 2007). Therefore, possessing many risk factors does not

entail that an individual will develop breast cancer. This stems from the fact that researchers are still unsure of how these risk factors cause cells in the breast to become cancerous (American Cancer Society, Inc., 2007).

Although women cannot change many of their risk factors, they are nonetheless able to make certain lifestyle changes in order to help prevent the development of breast cancer. In addition, the impact of breast cancer can be minimized by screening, early detection and appropriate genetic testing in individuals known to be at increased risk. The latter may identify individuals who are eligible for specific cancer prevention strategies (such as chemoprevention drugs or preventative surgery) to reduce the risk of cancer.

### **Screening and Genetic Testing**

In order to detect breast cancer before any symptoms have appeared, physicians and researchers recommend various screening methods. It has been demonstrated that the extent to which a breast cancer has spread, as well as its size when found, are two very important factors in determining the prognosis, or chances of survival, of the individual (American Cancer Society, Inc., 2007).

Various non-invasive breast cancer screening methods exist. For instance, the American Cancer Society, Inc. (2007) recommends that women over the age of 40 have an annual mammogram, or x-ray of the breast. The PHAC (2008) reports that when women aged 50-69 years adhere to mammographic screening, mortality from breast cancer can be reduced by nearly 30%. The American Cancer Society, Inc. (2007) also suggests that women starting in their twenties should receive a clinical breast exam, a manual examination of the breast by a medical doctor or nurse, every 3 years during their regular examination by their family physician. It has also been proposed that women

belonging to this age group should administer a breast self-exam once a month, or as regularly as possible, in order to become familiar with their breasts and report any suspicious changes to their family physician (American Cancer Society, Inc., 2007).

While the above-mentioned screening methods are very helpful and effective in detecting breast cancer in women at general popular risk for the development of breast cancer, additional strategies have been developed to identify and more intensively screen those women who are considered at higher risk for developing the disease.

One method available since the mid nineties that is clinically offered to a subset of women at higher risk is that of genetic testing (Di Prospero et al., 2001). Genetic testing involves the molecular screening of specific genes to identify gene changes (mutations) that are known to cause an elevated risk of developing breast cancers. More specific to breast cancer, women possessing mutations of the BRCA1 or BRCA2 genes have been identified as having a higher susceptibility to develop breast and ovarian cancer. It has been found that a woman carrying a BRCA1 and or BRCA2 mutation has an up to 80% chance of developing breast cancer throughout her lifetime (American Cancer Society, Inc., 2007). Moreover, this entails that carriers' children have a 50% percent chance of inheriting the BRCA mutation (Di Prospero, 2001).

Statistics demonstrate that approximately 3-8% of all breast cancers are attributable to the BRCA1 and BRCA2 mutations, and only about 1% of women in the general population tend to possess this mutation (PHAC, 2008). Generally, a woman's family doctor may suggest that she undergo genetic testing for the BRCA1 and BRCA2 mutations if she possesses a previous family history of breast cancer that is believed to be genetic. Physicians often consider the age of onset of these cancers within the family

before they offer a woman the option to undergo genetic testing. If it is determined that a woman may possess a minimum 10 to 15% chance of having the mutation, she may be eligible for genetic testing (Weber, 1996).

Although genetic testing can provide knowledge to a woman about her risk of developing breast cancer, it is nevertheless a debated subject due to the potential psychological implications of such a diagnosis (Di Prospero et al., 2001; Lerman, Seay, Balshem, & Audrain, 1995). So as to better comprehend these potential implications, I will examine the psychological implications of being a cancer sufferer, and will consequently review the psychological impacts of genetic testing.

### **Psychological Implications of Living with Cancer**

Research indicates that physicians no longer view cancer as a cessation of daily activities and an automatic death sentence, but rather as a curable illness with an increasingly good prognosis. More specifically, death rates due to major chronic illnesses have been reduced from 794 for every 100 thousand individuals in 1984, to 622 for every 100 thousand individuals in 2001 (Samson & Siam, 2007). In addition, the death toll in Canada for all cancers combined in both sexes has dropped from approximately 191 per 100 thousand in 1984, to 173 per 100 thousand in 2004 (PHAC, 2007). Consequently, physicians have encouraged those individuals living with chronic illnesses, including cancer, to learn to adapt rather than prepare to die (Samson & Siam, 2007). What this suggests is that a rising number of individuals who have cancer or who have had cancer are dealing with the psychological as well as social impacts of this disease every day, while living a relatively 'normal' life.

Previous research, which has been conducted on breast cancer survivors, indicates

that length of time of remission does not necessarily entail that a woman is less concerned about the cancer and its recurrence (Polinski, 1994). More specifically, Polinski (1994) conducted a quantitative inquiry examining the physical, psychological, and functional status of  $N=223$  breast cancer survivors through the use of mailed-in questionnaires. The researcher found that along with the negative physical aspects of having had breast cancer, approximately 70% of the women stated that they were nervous about recurrence of the disease, anxious about medical checkups and possessed a negative body image. As these women had been on average cancer-free for about 8 years, these findings demonstrate the long-lasting physical, as well as psychological effects of breast cancer, and the fear of its recurrence (Polinski, 1994).

A differing study was conducted by Samson and Zerter (2003), who examined spirituality in the psychosocial adaptation of cancer survivors. In this qualitative study,  $N=4$  cancer survivors (with a minimum of 5 years remission) recalled their experiences with cancer. These researchers found that the cancer survivors generally described themselves as uncertain, vulnerable, helpless, and without control of the experience. However, with personal growth, the participants gained more control and placed more importance on their values and beliefs. Consequently, more emphasis was placed on their spirituality, as they wanted to dedicate themselves to something meaningful.

When reviewing this study, the various psychological implications of being a cancer patient are evident, even when one survives and is invited to recall the experience.

On the other hand, a qualitative inquiry conducted by Shanfield (1980) examined the psychological impact(s) of having had various types of cancer. Twenty participants who had been in remission ranging from 1-33 years were interviewed regarding their

experiences with cancer, living in remission, etc. The researcher found that the cancer survivors were deeply affected by their experience. More specifically, they continued to check for growths or symptoms of the disease, continued to experience concerns about their mortality, and maintained a feeling of vulnerability (Shanfield 1980). On the other hand, participants felt that their support systems were of great importance throughout and following their bout with cancer.

The participants from this study present descriptions of their emotions similar to emotions reported by Samson and Zerter's (2003) participants. For instance, the theme of vulnerability was considered significant to both groups of cancer survivors. Nevertheless, the publication date of Shanfield's (1980) study must be taken into account when considering these results, as cancer treatment and research has dramatically changed over the past three decades.

Through the use of a meta-analysis, Gotay and Muraoka (1998) examined 34 previous researches on the topic of long-term cancer survivors' quality of life (QOL). A long-term survivor was defined as an individual who had survived 5 or more years after having received an adult-onset cancer diagnosis. Researchers found that although the QOL amongst cancer survivors varied greatly, generally, a large portion of long-term cancer survivors "continue to experience negative impacts of cancer and/or treatment on their daily lives, resulting in decrements in QOL, well beyond the completion of therapy" (Gotay & Muraoka, 1998, pp. 664). More specifically, sexual satisfaction/functioning and psychological functioning were often found to be of concern to survivors; regardless of their diagnosis or of the treatment they had received.

This study's results are in line with the findings that were reported in the studies

previously reviewed in this section (Polinski, 1994; Samson & Zerter, 2003; Shanfield, 1980). In general, it seems that cancer survivors tend to continue to experience overall negative effects even years after they have been diagnosed with or have been in remission from cancer.

Cimprich, Ronis, and Martinez-Ramos (2002) conducted a study in which similar constructs were examined. More specifically, they performed a quantitative study investigating whether QOL in breast cancer survivors was affected by factors such as age at diagnosis and years of survival. A total of  $N=105$  participants at least 5 years post diagnosis completed a mail-in questionnaire which measured QOL. In order to compare specific age ranges, the researchers sent out questionnaires to women belonging to particular age strata (less than 45 years old, 45 to 65 years old, and older than 65 years old). Through results it was found that the QOL of long-term breast cancer survivors is significantly affected by age at diagnosis. Specifically, the researchers found that younger women's (less than 45 years of age) social well-being was more affected compared to the other age groups, whereas older women (over 65 years of age) tended to experience the poorest physical well-being. The women who received their diagnosis at a middle age (between 45 and 65 years) experienced much better physical and social outcomes than the younger and older women.

Throughout this study, the researchers speculated that women in particular age strata have specific biological, social, and personal goals, and that these goals can be disrupted by significant life events such as the diagnosis of breast cancer, thereby negatively impacting their QOL (Cimprich et al., 2002).

This study provides interesting information with regards to life stages and the

effect of a significant event such as cancer on an individual's life and QOL. Furthermore, its results could be applied to the experiences of women undergoing genetic testing for BRCA1 and BRCA2 mutations, since being identified as a carrier of this genetic mutation may constitute a significant life event for these women. Consequently, such an event could have an impact on a woman's life stages, and as a result her QOL.

The results presented in these studies seem highly applicable to the experiences of women who have been identified as carriers of a genetic mutation predisposing them to develop breast cancer. Like the cancer survivor, the genetically predisposed woman possesses a higher chance than the average population to develop cancer (American Cancer Society, Inc., 2007; Graham, Ramirez, Love, Richards, & Burgess, 2002). Furthermore, she is more likely to engage in preventative measures such as lifestyle changes, advanced and regular screening and possibly preemptive surgeries, similarly to women with a family history (Kash, Holland, Halper, & Miller, 1991).

### **The Psychological Impacts of Genetic Testing for Breast Cancer**

Previous research that has been conducted on the psychological implications of genetic testing for mutations of the BRCA1 and BRCA2 genes has found that those who discover they possess the mutation tend to experience many negative effects. For instance, Lerman et al. (1995) conducted a quantitative study in which they examined the interest and expectations of genetic testing in a large number of women ( $N=105$ ) with first-degree relatives suffering from breast cancer. Women were interviewed by telephone and questioned about their interest in genetic testing, the reasons for which they would undergo the procedure, as well as their potential reactions to a positive or negative test result. It was revealed through findings that most participants (91%) were

interested in undergoing genetic testing. Furthermore, the interviewed women generally stated that their children's welfare, taking better care of oneself, and greater use of cancer-screening tools were all motivations to take part in testing. On the other hand, the majority of women anticipated that a positive test result would lead to increased anxiety and depression, and would also negatively affect their quality of life (Lerman et al., 1995).

Despite these researchers' findings, it is important to note that this study was undertaken only a few years after the conception of genetic testing for breast cancer. Therefore, these women may not have been as knowledgeable about the risk factors or implications of such tests, thereby potentially affecting results.

A more recent quantitative study on the psychosocial implications of genetic testing for breast cancer was conducted by Di Prospero et al. (2001). Twenty-four women who had tested positive for a BRCA1 or BRCA2 mutation were invited to participate in a focus group ( $n=8$ ), or to answer a mailed-in questionnaire ( $n=16$ ). These women's perception and worry about cancer, whether they communicate with their immediate family, their outlook regarding prevention and surveillance, their satisfaction with received services, their need for support, and their satisfaction with undergoing genetic testing, were all examined. The researchers determined that in general, participants experienced increases in cancer perception risk, as well as worry about cancer. In addition, many participants had faith in advanced cancer surveillance (97%) and believed that they would undertake personal prevention techniques such as lifestyle changes.

Overall, participants had shared the results of their genetic test with immediate family, however certain reasons stated for not having shared this information were the

age of the family member(s), a desire not to spread fear, and lack of closeness with the relative (Di Prospero et al., 2001). Moreover, while 40% of women indicated that they would be interested in forming a support group, and all of the women indicated that they were satisfied with the services they received, they nonetheless believed that the wait time for test results was too lengthy. It was also found that no participant regretted her decision of having undergone genetic testing (Di Prospero et al., 2001).

The results obtained from this study suggest that while carriers experienced negative psychological effects such as increased worry and increased need for support from similar others, they nevertheless maintained that their decision to be tested was optimistic and were satisfied with the overall experience.

On the other hand, as this study was conducted with a relatively small sample ( $N=24$ ) and participants consisted of a relatively small, homogeneous group, results should be generalized with caution.

Similar results were obtained through two separate studies conducted by Dorval et al. (2000), and Croyle, Smith, Botkin, Baty, and Nash (1997). These researchers also examined the psychological and or emotional reactions to BRCA1 testing on women with a family history of breast cancer. Both studies found that women tended to experience more negative emotions such as anger, worry, and distress, following the receipt of results, which indicated they possessed the genetic mutation (Croyle et al., 1997; Dorval et al., 2000). Furthermore, researchers Dorval et al. (2000) found that women who were carriers of the mutated gene generally experienced more negative emotional reactions to results than they had anticipated.

Differing results were obtained through a quantitative study conducted by Schwartz et al. (2002). One hundred and eighty-six women with a family history of breast cancer underwent genetic testing for a BRCA1/2 mutation. Carriers ( $n=43$ ) were invited to have family members tested as well, of which 38% possessed a definite mutation. Participants were contacted through telephone interview both one and 6 months following notification of results to ascertain their perceived risk and psychological distress level. The majority of carriers as well as affected relatives were not found to have experienced adverse psychological reactions. Furthermore, relatives who had received a negative test result indicating they did not have the mutation experienced a decrease in cancer-related distress (Schwartz et al., 2002). This suggests that those who learn that they do not possess the mutation may actually benefit from the testing.

The results from this study must nonetheless be interpreted in light of their limitations. For instance, researchers claim that the sample was relatively homogeneous; being mostly Caucasian, wealthy, and well educated. These characteristics may have affected results. In addition, the overall rate of attrition for those who tested either negative or had inconclusive results was more elevated than for those who tested positive. These factors may have contributed to the researchers' findings as those who abandoned the study may have experienced less distress than those who continued participation, thus affecting the comparison of distress between the three groups (Schwartz et al., 2002).

Comparable results were compiled by van Oostrom et al. (2003). These researchers, having conducted a 5-year longitudinal study with women either testing positive ( $n=23$ ), or negative ( $n=42$ ) for carrying the BRCA1 or 2 mutations, determined that carriers did not experience more distress than non-carriers overall. Nevertheless, both

groups experienced a heightened sense of anxiety and depression when compared to five years previous. An interesting finding is that individuals possessing certain characteristics tended to present more distress than those without such traits. More specifically, participants who had experienced changes in familial relationships, less communication with family about the test, and those who had doubts with regards to the genetic test's validity, tended to experience more distress (van Oostrom et al., 2003).

It is important to note that a large proportion of participants ( $n=21$ ) had a prophylactic mastectomy (PM), and  $n=12$  had undergone a bilateral salpingo oophorectomy (BSO) after they were identified as carriers of the genetic mutation (van Oostrom et al., 2003). These factors appear to have affected findings, because researchers indicated that those having undergone a PM reported a reduction in cancer risk fear. Hence, the overall lower level of distress after obtaining confirmation of carrying the BRCA1 or 2 mutations may have been due to the PM, and not the actual test results.

Similar results to those obtained by van Oostrom et al. (2003) and Schwartz et al. (2002) were also obtained by researchers Hamilton, Williams, Skirton, and Bowers (2009), Beran et al. (2008), as well as Lim, Macluran, Price, Bennett, Butow, and kConFab Psychosocial Group (2004). Particularly, these researchers each conducted longitudinal studies in which they examined the psychological effects of receiving a positive test result for the BRCA1 or BRCA2 gene mutation. Women possessing the BRCA1 or BRCA2 gene mutation were found to either not experience additional distress following testing (Hamilton et al., 2009), or experience remittance of distressing symptoms over time (Beran et al., 2008; Lim et al., 2004).

Additional psychological implications of being identified as a carrier of the BRCA1 or BRCA2 genetic mutation, which have been reported in certain studies, is that of developing an altered self-concept, and living with uncertainty (d'Agincourt-Canning, 2006; Dagan & Goldblatt, 2009; Esplen et al., 2009; McConkie-Rosell & DeVellis, 2000).

More specifically, d'Agincourt-Canning (2006) conducted a qualitative study in which she examined the subjective interpretations of self and self-identity of  $N=39$  participants, of whom 28 were BRCA1 or BRCA2 positive. In general, following the receipt of their genetic test result, participants were found to become more aware of certain aspects of themselves, particularly their physical selves (or embodied selves), their social selves, and their familial-relational selves. Furthermore, participants generally tended to view their test result as initially negative, but as a chance to become proactive in detecting and preventing the development of cancer. On the other hand, certain participants ( $n=3$ ) perceived their test result as a sure indication of the future development of cancer. Finally, some participants described their test result as having propelled them into a state of uncertainty, feeling as though they were not necessarily sick, but not necessarily healthy either (d'Agincourt-Canning, 2006).

This study's results must be interpreted with certain factors in mind. Specifically, although most participants were not previously affected by cancer, certain were, which may have influenced results. Furthermore, the age-range of participants spanned nearly forty years, from around 25 years to 60 years, and although most participants were women, some men were also included. Finally, not all 39 participants had received a

positive test result indicating the possessed a BRCA1 or BRCA2 genetic mutation, potentially affecting results (d'Agincourt-Canning, 2006).

Similarly, researchers Dagan and Goldblatt (2009) conducted a qualitative study in which they examined the experiences of  $N=17$  women possessing Ashkenazi mutations in the BRCA1 and/or 2 genes. Through a phenomenological analysis, these researchers found that 4 dominant themes emerged: focusing on illness or surgery, family, knowledge of possessing the gene, and their mother's experiences. In addition, the researchers described that these women felt as though they were caught between health and sickness, leading them to feel vulnerable and to feel that they possessed an unstable future.

Certain limitations must be considered when reviewing this study. More specifically, the women participating in the study were all recruited from a medical centre in Israel, and all possessed the Ashkenazi mutations in the BRCA1 and/or 2 genes. However, the BRCA 1 or 2 genetic mutation is much more common amongst Ashkenazi Jewish women (Caldes et al., 2002). Therefore, their experiences may not be as transferable to other populations, as the frequency of this genetic mutation is much more common amongst these women, potentially affecting their perception of receiving a positive test result.

In a differing study that was conducted by Esplen et al. (2009), the researchers devised a scale intended to measure self-concept in BRCA1 or BRCA2 genetic mutation carriers. Through two separate studies, these researchers conceived of a 17-item scale which was found to have three specific factors: vulnerability, stigma, and mastery.

This study's findings imply that women undergoing testing and receiving a positive test result indicating they possess a mutation of the BRCA1 or BRCA2 genes

may encounter feelings such as vulnerability or even stigma. Although these findings seem very revealing, they must nevertheless be considered in light of certain limitations. For instance, the scale's test-retest reliability was not determined, entailing that results should be interpreted with caution until further testing can be implemented (Esplen et al., 2009).

In general, it appears that while women who are identified as carriers of the BRCA1 or BRCA2 mutations do not suffer significant psychological distress, they nevertheless experience distressing emotions, such as symptoms of depression and anxiety. Also, it seems as though certain factors such as communication with family and closeness with family or relatives tend to affect whether a carrier will disclose such information, and whether this will result in additional negative emotions. Furthermore, the long-term psychological effects of genetic testing appear to become less pronounced over time. Finally, some women describe that the receipt of a positive test result has affected their self-concept, or the manner in which they perceive themselves.

Another potential implication of undergoing genetic testing for BRCA1 or BRCA2 mutations may be the possible stigma associated with receiving these results and potentially developing a life-threatening illness.

### **Stigma**

Crocker and Major (1989), claim stigmatized individuals are those that “by virtue of their social membership in a social category are vulnerable to being labeled as deviant, are targets of prejudice or victims of discrimination, or have negative economic or interpersonal outcomes” (pp. 609). These researchers also add that people who are

stigmatized are “devalued not only by specific ingroups but by the broader society or culture” (Crocker & Major, 1989, pp. 609).

Goffman (1963) further distinguishes between *discredited* and *discreditable* stigmas. An individual with a discredited stigma is one who believes that his or her stigma is evident to others immediately. On the other hand, one with a discreditable stigma believes that his or her stigma is not instantly known by others, nor is it instantaneously perceivable by those present.

Hence, stigmatized individuals are those who due to a particular circumstance in their lives or their social membership in a certain group, experience prejudice and are discriminated against by the society at large due to their membership in this particular social category. Moreover, an individual with a discredited stigma is one who is stigmatized, because of an immediately visible ailment. Conversely, an individual with a discreditable stigma possesses a stigmatizing ailment which can be concealed, and that can also be revealed by this individual when they feel compelled to do so (Goffman, 1963; Rosman, 2004).

Goffman (1963) discusses the concept of *managing information*, referring to those afflicted with a discreditable stigma. For instance, he presents the example of a mental patient in a hospital, as compared to an ex-mental patient. The mental patient in hospital is faced with a discredited stigma in the sense that those visiting him are aware of his condition. On the other hand, the ex-mental patient encounters a very different situation. More specifically, “it is not that he must face prejudice against himself, but rather that he must face witting acceptance of himself by individuals who are prejudiced against the persons of the kind he is revealed to be” (Goffman, 1963, pp. 42). Therefore,

the ex-mental patient, or the discreditable individual, must decide whether to display, to tell, to let on, or to lie about the condition that makes him or her considered stigmatized (Goffman, 1963).

Stigma has been found to lead to rejection, which consequently leads to the development of negative self-perceptions. In addition, those with concealable (or discreditable) stigmas are more likely to demonstrate negative self-perceptions (Frale, Platt, & Hoey, 1998).

Previous research conducted on the topic of stigma demonstrates the negative consequences of being a member of a stigmatized group. For instance, Frable et al. (1998), examined the self-esteem of  $N=86$  Harvard undergraduates who either possessed a concealable stigma such as being gay, having a low income, or being bulimic, compared to those with a visible stigma. Participants, who were asked to record various aspects of their daily lives, were found to report lower self-esteem and increased negative affect if they possessed a concealable stigma.

On the other hand, findings presented by Weiner, Perry, and Magnusson (1988) suggest that the perceived stability and controllability of different stigmas elicit differing reactions from others. Through two separate studies, these researchers presented participants with 10 different stigmas that were either classified as physical or mental-behavioral, and the reactions to these stigmas were classified as either anger, pity, or helping judgments.

Results of the first study indicated that the participants,  $N=59$  first-year University students, perceived the physical stigmas (for instance, blindness, cancer, paraplegia) as onset-uncontrollable, and elicited reactions such as pity and judgments to help.

Conversely, those stigmas which were considered mental-behavioral (such as having the Acquired Immune Deficiency Syndrome (AIDS), having suffered through child abuse or obesity), were perceived as being onset-controllable and elicited reactions such as anger and judgments to neglect.

Similarly, the results of the second study revealed that participants, of a somewhat larger sample ( $N= 320$ ), maintained the same perceptions as the participants in the first experiment, even when a cause for the stigmas was provided. For instance, the researchers presented a scenario where an individual develops cancer either because they were unknowingly living in a toxic area, or because they were working with a cancer-causing agent without wearing the proper protective equipment (Weiner et al., 1988). While those with cancer were considered to possess a physical, and onset-uncontrollable stigma, the reactions to this disease nevertheless elicited pity, an overall undesirable emotion. On the other hand, it must also be considered that this study is relatively dated, and its results should consequently be interpreted with caution.

The previously reviewed studies' results seem to indicate that those who are stigmatized tend to experience more negative emotions and have poorer self-esteem. In addition, it appears that individuals presumably not afflicted by stigma feel compelled to pass judgment on those possessing an undesirable difference.

### **Stigma and Disease**

Previous research which has been conducted on the stigma of possessing certain diseases or disorders such as AIDS, mental illness, sexually transmitted diseases (STDs), etc., tends not to provide consistent findings with regards to the reported effect of stigma on participants (Camp, Finlay, & Lyons, 2002; Nack, 2000; Santana & Dancy, 2000).

For instance, Camp et al. (2002) examined self-esteem and stigma in a group of women ( $N=10$ ) suffering from stigmatizing mental health problems. Through the use of open-ended interviews, the researchers asked participants how they believed others understood mental illness, if they themselves agreed with these understandings, how mental illness was considered a part of their identities, and if the women engaged in negative self-evaluations regarding their stigmatizing condition. The researchers found that although the participants perceived society as possessing negative evaluations of mental illness, felt that their mental illness was considered a salient part of their identity, and applied society's negative representations of them to themselves, they nevertheless did not consider society's representations of them to be valid. More specifically, the researchers concluded that these women did not consider the stigma of mental illness as something they were responsible for, but rather due to those who engage in stigmatization.

However, it must be mentioned that although these women did not consider their illness as a fault of their own, nor did they place the blame for the stigma of mental illness onto themselves, they nevertheless felt stigmatized by society, and even engaged in certain avoidance and selective disclosure behaviors.

A dissimilar study conducted by Nack (2000), examined the manner in which  $N=28$  women with chronic STDs ranging from 19 to 56 years of age, "managed the impact of stigma on how they saw themselves as sexual beings" (p. 95). Through the use of conversational, unstructured interviews, the researcher found, amongst many findings, that all but one of the 28 participants felt significantly transformed by their STD diagnoses and utilized differing methods to attempt to manage her stigma. These stigma

management strategies each had their own impacts on the manner in which these women viewed themselves sexually. Furthermore, while 9 participants stated that they were able to reveal their diagnoses to certain friends and family, the majority of the women did not feel as comfortable. Although these women eventually accepted their stigmatizing condition, they nonetheless experienced a variety of negative emotions before reaching this accomplishment. Moreover, as their stigma was generally imperceptible to most individuals, these women found that the stigma only penetrated their sexual identity (Nack, 2000).

In light of these findings, certain limitations of this study must still be considered. For example, the main researcher had significant emotional involvement in the topic of STDs, as she herself had previously been infected, which may consequently have biased her results. However, it must also be noted that the researcher did provide a detailed explanation of this ordeal and may henceforth have taken this factor into consideration throughout the completion of her research.

Santana and Dancy (2000) conducted a similar study in which they examined the stigma of AIDS in a group of Haitian-American women ( $N=11$ ), mainly recruited from a Haitian-American women's association. The researchers conducted a qualitative cross-sectional study based upon one focus-group interview. Participating women were asked whether the AIDS epidemic had affected Haitians' lives in the United States (U.S.), how they have been affected, which type of labels have been used by others to describe Haitians, and so on. It was found that many facets of Haitian-American women's lives were affected by the stigma of Haitians being labeled as 'AIDS carriers' by the dominant society. More specifically, these women stated that their self-esteem and intimate

relationships were both affected. Moreover, they experienced self-doubt, rejection by the dominant society, as well as rejection from Haitians. For instance, a woman recounted that when someone became ill, the rest of the Haitian society assumed that they were AIDS carriers, regardless of their symptoms, and tried to avoid this individual at all costs in order not to be associated with the virus.

This study presents significant findings in that it clearly supports Goffman's (1963) theory on stigma, particularly the rejection of stigmatized individuals by the dominant society. However, it must be considered that these findings are only transferable to a specific population; that of Haitian-American women who partake in a community association. Therefore, findings should be interpreted with caution.

### **Stigma and Cancer**

Similarly to many individuals who possess concealable stigmas such as homosexuality, low income and bulimia, the discredited and at times discreditable stigma of being a cancer sufferer was largely overlooked in the past (Frable et al., 1998; Klawiter, 1999). Klawiter (1999) claims that due to the symbolic nature of a woman's breasts, which are generally considered a source of vitality and sexual power, the stigma associated to breast cancer has consequently been intensified.

Since it has been found that certain aspects of cancer sufferers or survivors and women possessing mutations of the BRCA1 or BRCA2 genes experiences are similar, it seems appropriate to examine literature in which stigma was investigated amongst cancer sufferers.

Previous literature describes the stigmatizing experience of being a cancer patient undergoing treatment. For instance, Wilson and Luker (2006) found that individuals with

cancer tend to feel more comfortable and accepted when spending time in the cancer-centre where they receive their treatment, compared to the outside world. They also discovered that cancer patients tend to feel admired and uneasy when in the company of outsiders, and consider the hospital a 'safe' place where they do not feel stigmatized (Wilson & Luker, 2006).

On the other hand, certain researchers have found that cancer patients, particularly women, with breast or lung cancer, feel much stigma when they begin treatment for cancer, and develop alopecia (hair loss) (Rosman, 2004). For instance, many women associate hair loss with the label of 'cancer patient', and even though certain women attempt to cope with hair loss with the use wigs, they continue to experience stigma. Rosman (2004) has identified this occurrence as a discreditable (not immediately visible) rather than discredited stigma.

Individuals experiencing a more permanent change to their bodies, such as women undergoing a mastectomy (removal of the breast), have been found to experience strong negative emotions. For instance, Rancour and Brauer (2003) investigated the experience of a woman undergoing a mastectomy for recurrent breast cancer. They proposed the use of a journal to aid the woman to cope with the situation. Nevertheless, throughout the journal, it became evident that this woman had experienced fear and devastation having undergone such a disfiguring treatment. This surgery was particularly distressing since she had recently been treated for a previous breast cancer. Rancour and Brauer (2003) add that many patients who undergo body-altering operations grieve the losses related to these significant changes. They name such operations: stigma-producing anatomic alterations, similar to those operations experienced by women undergoing a

mastectomy.

Conversely, Fife and Wright (2000) conducted a study which involved the comparison of the stigma experienced by individuals who had the Human Immunodeficiency Virus (HIV)/AIDS ( $n= 130$ ) and those who had cancer ( $n= 76$ ). They specifically attempted to discern whether stigma differs across illnesses, and if individuals affected by certain illnesses were impacted differently due to the differing illness. The researchers also examined four dimensions of perceived stigma: those of social isolation, internalized shame, financial insecurity, and social rejection.

Through the use of questionnaires, Fife and Wright (2000) found that individuals with HIV/AIDS and cancer both felt stigmatized. Although those with HIV/AIDS reportedly experienced stronger feelings of stigma than those with cancer, the researchers stated that overall; the nature of the illness had little direct effects on self-perception.

This study should however be considered in light of its limitations. For instance, there was a significant difference between the size of the HIV/AIDS group and the cancer group. Furthermore, the HIV/AIDS group was comprised of mostly men (123 out of 130), thereby potentially skewing results.

### **Summary**

The results reported in the previous studies contend that individuals who are suffering from and or being treated for cancer tend to feel stigmatized by the general population.

In the same vein, results obtained from research that has been conducted with cancer survivors tend to communicate that survivors often experience negative psychological effects due to their diagnosis (Gotay & Muraoka, 1998; Polinski, 1994;

Shanfield, 1980; Samson & Zerter, 2003). Furthermore, the time-period during which a woman is diagnosed with breast cancer has been found to determine how she might be affected psychologically (Cimprich et al., 2002).

Upon reviewing studies which have been conducted on the psychological impacts of genetic testing for breast cancer, it seems as though most women, whether asked to imagine or having actually received a positive test result, either foresee experiencing or actually experience negative psychological effects due to the receipt of a positive test result (Beran et al., 2008; Croyle et al., 1997; Di Prospero et al., 2001; Dorval et al., 2000; Lerman et al., 1995; Lim et al., 2004). Nevertheless, varying results have been obtained, indicating that future research must be conducted in order to determine whether receiving a positive test result from BRCA1 or BRCA2 mutation testing leads to negative psychological effects (Hamilton et al., 2009; Schwartz et al., 2002; van Oostrom et al., 2003).

Similarly, it appears that BRCA1/2 genetic testing may have an effect on a woman's self-concept. However, this construct has been understudied, and more research is needed to improve understanding (d'Agincourt-Canning, 2006; Esplen et al., 2009; McConkie-Rosell & DeVellis, 2000).

On a different note, researchers examining the topic of stigma and various diseases tend to report that those afflicted with stigmatizing conditions, whether mental or physical, are often heavily impacted psychologically (Camp et al., 2002; Nack, 2000; Santana & Dancy, 2000).

Furthermore, findings from research that has been conducted specifically on cancer and stigma indicate that cancer patients often feel stigmatized by non-cancer

sufferers. These feelings have been found to persist even when the cancer sufferer cannot be distinguished from non-cancer sufferers (Fife & Wright, 2000; Rancour & Brauer, 2003; Rosman, 2004; Wilson & Luker, 2006). However, one must note that there is an apparent lack of qualitative studies examining the experience of stigma in cancer sufferers.

Although the topic of cancer and various constructs has been thoroughly researched, it is also essential to further understand the potential implications of BRCA1 and BRCA2 genetic testing, as approximately 6 million individuals worldwide possess this mutation (PHAC, 2008). Since one's breast cancer risk level can be relatively quickly determined following genetic testing, physicians and patients tend to strongly consider pursuing this course of action, particularly as early detection of breast cancer is imperative to ensure a good prognosis (American Cancer Society, Inc., 2007). However, the potential psychological implications of undergoing genetic testing should also be considered when choosing to face BRCA1/2 genetic mutation testing. Although research has been conducted to examine psychological effects in women who have undergone BRCA1 or 2 gene mutation testing, very few qualitative studies have been conducted to investigate the potential experience of stigma in carriers of the genetic mutations.

Consequently, in the current research, I chose to examine whether women who have tested positive for mutations of the BRCA1 or BRCA2 genes experienced stigma. In order to gather an in-depth understanding of these women's lives, a qualitative inquiry was undertaken.

## **Objectives and Research Questions**

The main objective of this study was to better understand the experiences of women who tested positive for a mutation of the BRCA1 or BRCA2 genes, and the manner in which they experienced stigma in their daily lives due to their test result. The following research questions served as a guide for this qualitative research study:

- 1) How do women who have been identified as carriers experience the initial news of receiving a positive test result?
- 2) How do women who possess mutations of the BRCA1 or BRCA2 genes experience stigma, and how has this impacted their lives?

## **Contributions to Knowledge**

As previously mentioned, the experiences of women having been identified as carriers of the BRCA1 or BRCA2 genetic mutations have been understudied. Hence, the aim of the current study is to examine the lived experience of women who have received a positive test result indicating that they possess mutations of the BRCA1 or BRCA2 genes. Through this research it is hoped that researchers, physicians, and therapists may develop a better understanding of the subjective experiences of these women.

## **Method**

### **Rationale for a Grounded Theory Methodology**

In order to further comprehend the experiences of women who have been genetically tested and found to possess mutations of the BRCA1 or BRCA2 genes, as well as to explore whether these women feel stigmatized, a qualitative study utilizing a grounded theory methodology was conducted.

Grounded theory methodology was selected so as to better understand and give meaning to the various aspects of these women's experiences (Zogran, 2008). This method also allows researchers to conceptualize participants' experiences through the development of a conceptual model. More specifically, this model enables the researcher to characterize data and various emerging concepts through the method of constant comparison analysis (Glaser & Strauss, 1967).

Grounded theory is appealing as it is considered an "ever-developing entity, not as a perfected product" (Glaser & Strauss, 1967, pp. 32). This suggests that as the theory is further supplemented and developed, it will become more rich and dense, thereby potentially better describing and encapsulating the experiences of individual participants (Glaser & Strauss, 1967).

Grounded theory, while initially conceived of for sociological research, has also been widely utilized in psychology. Specifically, as grounded theorists work with natural language rather than numbers, this method can be more easily applied by psychologists studying psychotherapy (Rennie, 1998). Grounded theory permits researchers to access a certain aspect of human experience that is virtually inaccessible when utilizing quantitative methods attempting to achieve the same results. For instance, concealed facets of clients' experiences during therapy have been revealed by utilizing this method, thereby strengthening psychologists' motivation to consider the grounded theory method when they feel constricted by the natural science canons (Rennie, 1998; Rennie, Phillips, & Quartaro, 1988).

Although grounded theory methodology was utilized in this study, due to the small number of participants, saturation (later described in detail) was not achieved. Therefore,

analysis was halted following the conceptual organization of data, also known as *conceptual ordering*, which is a method devised by Strauss and Corbin (1998) that allows researchers to conceptually organize their data without the final step of devising a theory. As more participants are recruited and the analysis of further interviews is undertaken, saturation should be achieved, leading to a theoretical representation of the data.

### **Grounded Theory Methodology Defined**

Grounded theory methodology is a discovery-oriented qualitative approach, which was originally conceived by Glaser and Strauss in 1967 (Rennie, 1998). This particular research methodology was considered innovative as a qualitative research method because it involved reversing the typical order of research steps, contrasting the then popular logico-deductive method (Walker & Myrick, 2006). Specifically, rather than postulating a hypothesis, which is thereby tested, grounded theorists instead firstly immerse themselves in their data to 'discover' a theory that emerges from the data itself (Glaser & Strauss, 1967).

Although after its initial conception grounded theory became increasingly widespread, the two founding researchers eventually began to disagree about many fundamental aspects of the approach (Walker & Myrick, 2006). Glaser claims he has maintained a style more similar to the original grounded theory, initially conceptualized by Glaser and Strauss. On the other hand, Strauss has paired himself with researcher Juliet Corbin, and has implemented various changes to the original method (Heath & Cowley, 2004; Rennie, 1998). According to Rennie (1998), the rift between Glaser and Strauss stems from "a fundamental disagreement about what amounts to the logic of justification of the methodology" (p. 102).

Heath and Cowley (2004), claim that qualitative research or analysis is a cognitive process, and that each individual has their own cognitive style. In other words, although each method in grounded theory may have its own strengths and weaknesses, the method that best suits one's cognitive style should be implemented. Therefore, for the purposes of this study, the original Glaser and Strauss (1967) method was utilized, as it emphasizes certain aspects of grounded theory which best cohere with my fundamental research beliefs such as induction and theory emergence (For a more descriptive and detailed explanation on the differences between Glaserian and Straussian grounded theory methodology, see Heath and Cowley, (2004), and Rennie, (1998)).

**Data Collection.** The process of undertaking a grounded theory research, in accordance to Glaser and Strauss (1967), involves *theoretical sampling*, a process whereby the researcher “collects, codes, and analyzes his data and decides what data to collect next and where to find them, in order to develop his theory as it emerges” (Glaser & Strauss, 1967, pp. 45). Therefore, the data collection process is dependent upon or controlled by the emerging theory. Unlike most research methodologies, decisions in grounded theory methodology are not founded upon a predetermined theoretical framework. Glaser and Strauss (1967) stress that *theoretical sensitivity*, or the ability to devise a theory as it emerges from data, is lost if the researcher entrusts him or herself to one particular predetermined theory.

**Data Analysis.** Researchers conducting grounded theory research engage in *constant comparison analysis*, which involves the joint coding, and analysis of data. More specifically, data is compared to other data, and categories and codes are derived from their commonalities (Rennie, 1998). The relationship amongst categories is

examined, and those that can be grouped together are united to form higher-order categories. Eventually, the goal is to form a core category, which encompasses all others. Once it is deemed that new data no longer supplements or adds meaning to the emerging findings, data collection ceases and the classification is considered saturated (Rennie, 1998). It should be noted that according to the Glaser and Strauss method, the process of coding is meant to be a fluid process with no distinct steps, contrary to the version later brought forth by Strauss and Corbin (Heath & Cowley, 2004).

As previously mentioned, due to the number of participants and interviews, saturation was not achieved. Specifically, the analysis was concluded after conceptual ordering until further interviews can be conducted so as to achieve saturation and the development of a grounded theory.

Conceptual ordering involves the “organization of data into discrete categories...according to their properties and dimensions, then the utilization of description to elucidate those categories” (Strauss & Corbin, 1998, pp. 54-55). Strauss and Corbin (1998) further clarify that the process of conceptual ordering is considered a precursor to theorizing. Although the researchers stress that theorizing is the ultimate goal of grounded theory, conceptual ordering is considered an acceptable conclusion to research prior to the development of a theory (Davieson, O’Callaghan, Grocke, 2008).

## **Participants**

**Eligibility Criteria.** In order to be eligible for this study, participants had to be women who had undergone genetic testing for breast cancer. Specifically, these women must have received a positive test result indicating that they possessed a mutation of the BRCA1 or BRCA2 genes.

As the study was conducted in a city where most individuals could speak either French or English, only women who could speak either of these two languages were invited to participate. This measure was implemented so as to ensure that unnecessary frustration did not ensue, particularly considering the sensitivity of the research topic.

For the purposes of this study, only women who had received positive test results at least 6 months prior to the interviews or more were invited to participate. This precaution was intended to firstly allow women to thoroughly examine and consider the implications of this sensitive news. Secondly, this also hopefully enabled women to get through the initial shock and potential disbelief associated with the receipt of the test result.

Women who had elected to undergo close monitoring by their physicians and specialists were considered for this study. More specifically, women who had opted to undergo a PM were not included as this measure has been hypothesized to reduce cancer-related fears, and these women may therefore have encountered different experiences than women who chose to conserve their breasts (van Oostrom et al., 2003).

Finally, women who possessed a previous psychiatric history, for instance major depression, were not invited to participate in this study as the risks may have outweighed the benefits.

**Age Range.** Inspired by the findings of researchers Cimprich et al. (2002), women between the ages of 20 and 45 years were invited to participate in this study. These measures were instilled in order to examine the experiences of women who may have similar social, personal and biological goals. Consequently, this also entails that

women aged younger than 20 years were not included, as their experiences could have been significantly different, particularly as they are terminating puberty.

**Recruitment.** Women invited to participate in this study were recruited from a large hospital offering comprehensive services to women at high risk for the development of breast cancer in the federal capital of Canada. This particular site was selected as its location was in proximity to where the current research was being conducted, as well as due to the specialized and advanced breast cancer screening facilities offered at this hospital.

**Demographics.** The average age of the six women interviewed was 38.5 years (range 31 to 44 years). The average age of participants when they received their positive test result indicating that they possessed a mutation of the BRCA1 or BRCA2 gene was approximately 35.5 years (range 30 to 39 years). All six participants were married, and each had at least one child (range of 1 to 3 children, average of 2 children) (See Table 1). The average age of participants' children was 8.75 years, and their ages spanned from 20 months to 19 years. Four out of the six participants had at least one daughter (range from 1 to 2, average of 1.75).

Four of the six participants possessed at least one university degree, and the remaining two participants had attended community college. Four of the six participants reported that they resided in an urban setting, and two were from a rural area.

None of the participants reported having taken part in any drug trials for the treatment of breast cancer. In addition, only one woman had had a prophylactic surgery (oophorectomy) following the receipt of her test result. Each participant stated that she had been receiving specialized care since the receipt of her test result; referring to the

care she received at the specialized clinic where she had been recruited. Only one of the six participants reported having sought additional care through the means of a homeopathic doctor.

All six participants had a family history of breast and/or ovarian cancer. Specifically, at least 2 family members were affected in each of the participants' families, and all six participants stated that they had experienced the death of at least one family member due to either breast or ovarian cancer. Family members were reported to be: mothers, sisters, maternal or paternal aunts, maternal or paternal grandmothers, and great aunts. All six participants possessed at least one other family member who had also been identified as having a mutation of the BRCA1 or BRCA2 genes, and most of the participants (four) reported that either their mothers and/or sisters had also been identified as possessing the genetic mutation.

Table 1

*Demographic Information*

Demographic Variable	Ratios and ages
Average age	38.5 (range 31-44) years
Average age at receipt of positive test result	35.5 (range 30-39) years
Average age of children	8.75 (range 20m-19) years
Urban: Rural home	4:2
College: University	2:4
Sought additional care	1/6

While all six participants considered their spouse and specialists/physicians to be a part of their support networks, few reported that they considered their children, neighbours, or work colleagues as social support. Other individuals who were commonly stated as being part of these participants' social support network were their friends, family doctor, and family.

Participants were treated in accordance to the ethical guidelines of the Canadian Counselling and Psychotherapy Association. In addition, this study was approved by the Ethics Committee of the University of Ottawa as well as the Ottawa Hospital Research Ethics Board.

### **Measures**

**Demographics.** In order to collect pertinent information for the study, participants were invited to complete a demographic questionnaire (See Appendix A). Specifically, information such as age, date and age when the news of the positive test result was received, previous breast-cancer related family history, marital status, number of children, race, occupation, support structure (for example, spouse, family, friends), and level of education, was collected. This information was collected so as to allow researchers and readers to further understand the context of the experiences of these women.

### **Procedures in Conducting a Grounded Theory Study**

As this study was conducted utilizing a grounded theory methodology, a literature review was initially undertaken with the purpose of guiding the topic of study. It was revealed through the review of the literature that there was a void in the domains of qualitative research and genetic testing for breast cancer susceptibility. Furthermore, the

potential impact of stigma on these women had been scarcely investigated. Consequently, the current study's objectives and two research questions, which served as a guide, were formulated.

The physicians from the specialized risk assessment clinic at the previously mentioned hospital agreed to include interested patients from their practice in the study. These physicians determined the number of women in their practice who possessed BRCA1 or BRCA2 genetic mutations and who fit inclusion criteria. Once this was determined, these physicians instructed a designated nurse to print stickers with eligible patients' contact information and affix them to individual pre-stamped envelopes that I had previously prepared. These envelopes each contained: a letter addressed to the individual patient from their physician; a short summary of the research objectives, methodology, and inclusion criteria; and a last form in which the patient was asked whether they would like to hear more about the study. Patients were then invited to return this last form indicating whether or not they would like to be contacted (See Appendix B). Each envelope also contained another pre-stamped envelope with the correct return address so that the patient was able to send the form back to the appropriate location free of charge.

Envelopes, which were returned to the previously mentioned hospital, were collected by one of the two physicians and read by the above-mentioned nurse who then notified me when she received a response indicating that a patient wanted to hear more about the study.

Twelve letters were initially mailed to women who were between the ages of 30 and 40 years, and who fit all other inclusion criteria. However, since only three women

responded to this initial invitation to participate, a reminder letter was sent out to the nine women who had not responded to the initial invitation (See Appendix C). From this second attempt to contact participants, one response was received. Following this attempt, the age range was amended to include women who were between the ages of 30 and 45 years. Letters inviting patients to hear more about the study were therefore mailed to eight women between the ages of 41 and 45, and who fit inclusion criteria. Two responses were received from this third attempt, leading to a total of six participants.

I contacted each patient who reported wishing to hear more about the study by telephone. During our conversation, I presented myself as the primary researcher of this study, and provided her with information regarding the research questions, methodology, and procedures of the study. In addition, I answered any questions she may have had regarding the study and its procedures. I then asked her to consider whether she would like to participate in the study.

Once the woman agreed to participate, we selected a time, which was most convenient for her to participate in the interview. The interviews took place at the previously described institution in a private room in the clinic. This helped ensure privacy and confidentiality, and also consisted of a familiar environment for the participant.

For the purposes of this study, semi-structured interviews were conducted. This particular interview type was selected as it allows the interviewer to follow-up with further questions and probes, which then enables the researcher to base future questions on the information gathered from initial interviews (Brugha, Bebbington, & Jenkins, 1999). Interviews each lasted approximately 1 hour and 10 minutes.

Before each interview, an informed consent and confidentiality agreement was presented to the participant (See Appendix D). This informed consent form also contained a separate section in which the participant was asked to sign if they accepted to be contacted for further follow-up interviews and/or questions, in the event that additional information would be required. Once the document was signed, the participant was then asked to complete the demographic questionnaire in order for me to better comprehend each individual's background and the context in which participants described their experiences. Following the completion of the questionnaire, the interview began. All interviews were digitally recorded in order to ensure accurate recollection of the participant's experiences, as well as to facilitate coding and analysis of data.

The tape-recorded interviews were stored in a secure location to which only I had access so as to ensure confidentiality. Furthermore, in order to maintain complete anonymity, each interview (and the resulting transcript) was given a pseudonym.

After the completion of every interview, it was transcribed verbatim onto a computer into a Word document. Immediate transcription helped to ensure that accurate information was gathered. Moreover, immediate transcription also served as a memory refresher and consequently facilitated the formulation of additional questions for upcoming interviews (Rubin & Rubin, 2005). Only I, the primary researcher, had access to this data, which was also protected by a security password.

Consistent with the grounded theory method, data collection and analysis was undertaken concurrently (Glaser & Strauss, 1967; Rennie, 1998). In other words, the information gathered from the initial interviews, as well as its following analysis, helped shape future interview questions and guide further data collection in order to better

understand the participants' experiences. Finally data was conceptually organized, or organized into distinct categories, depending on its properties.

### **Development of Interview Protocol**

The development of the interview protocol (See Appendix E) was guided by information gathered from the literature, as well as from reports brought forward by other professionals. Research amongst cancer sufferers, demonstrates that they experience stigma due to their condition (Fife & Wright, 2000; Rancour & Brauer, 2003; Wilson & Luker, 2006), and that even when their condition is not detectable, for instance, when they wear a wig, they continue to experience stigma (Rosman, 2004). As women who test positive for mutations of the BRCA1 or BRCA2 genes do not have a condition which is immediately visible to others, I thought it would be of interest to examine whether they also felt stigmatized as their medical condition cannot necessarily be detected.

The compilation of these research questions was also influenced by the account of one committee member who described the experience of a young woman who was at high risk for the development of breast cancer. This woman was experiencing a great deal of distress, as she explained that her friends had begun to treat her very differently since she had received this news- they did not wish to introduce her to potential dates, as they no longer considered her suitable for this role. As this was a very powerful statement, it was decided that the concept of stigma should be further examined amongst women who had received a positive test result.

Further questions were created after unearthing research findings which indicated that after their receipt of positive test results, women have been found to experience greater levels of anxiety, depression, and worry and they have also been found to choose

not to disclose their test results to certain individuals in order not to spread fear (Croyle et al. (1997); Di Prospero et al., (2001); Dorval et al. (2000); Lerman et al., 1995; van Oostrom et al. (2003)).

Finally, previous results obtained by Wilson and Luker (2006) who found that cancer patients tend to feel more at ease around similar others prompted the investigation of whether women who have received a positive test result for BRCA1 or BRCA2 mutations undergo similar experiences.

### **Methodological Rigor**

Methodological rigor for this study was established utilizing the criteria brought forth by Lincoln and Guba (1989), Glaser and Strauss (1967), and Sandelowski (1986). These researchers describe four criteria as being essential in judging the trustworthiness of qualitative research. More specifically, they discuss the concepts of credibility, transferability, dependability, and confirmability.

The first criterion was that of credibility or truth value. More specifically, the criterion of credibility concerns whether the presented experiences of participants are faithful to their actual lived experiences (Sandelowski, 1986). So as to increase the credibility of this research, I engaged in prolonged engagement in the topic of women at high risk for the development of breast cancer (Lincoln & Guba, 1985). More specifically, in order to fully immerse myself in this topic, I dedicated a day, prior to beginning data collection, during which I shadowed a physician at a clinic for women at high-risk for the development of breast cancer. I carefully listened to these women's stories and experiences, in order to better comprehend the issue at hand, and to better formulate my research questions.

Similarly, in order to increase credibility, I also engaged in persistent observation, which entailed focusing in detail on the attributes that were most relevant to the issue being researched (Lincoln & Guba, 1985). Finally, credibility was increased through constant comparison analysis, which entails that one constantly compares data, and modifies further interview questions accordingly (Glaser & Strauss, 1967).

The second criterion was that of applicability or transferability of the research. Transferability concerns whether the findings can 'fit' or apply to situations or circumstances independent of the study situation (Sandelowski, 1986). Throughout this research, transferability was increased through the rich description of data provided through extensive questioning of participants regarding their experiences, as well as the coding and analysis of the data (Lincoln & Guba, 1989).

The third criterion, which was utilized to increase the trustworthiness of this study, was that of dependability. The dependability, or the consistency of a study, can be increased through providing a clear and detailed sequence for future researchers to follow. This would allow future researchers to conduct a similar study where comparable findings should be achieved (Sandelowski, 1986). Dependability of the current study was increased by formulating and carefully following detailed procedures, which have been included in the final version of the research.

The final criterion utilized for establishing trustworthiness was that of confirmability or neutrality. Confirmability is considered to be "the extent to which data and interpretations of the study are grounded in events rather than the inquirer's personal constructions" (Lincoln & Guba, 1985, pp. 324). In order to increase this study's confirmability, grounded theory methodology was utilized. This particular method of

qualitative analysis entails that the obtained findings are grounded in the data itself, hence more clearly reflecting the experiences of the participants (Hamberg, Johansson, Lindgren, & Westman, 1994).

Additionally, confirmability was increased through a close revision of the raw data codes derived from the interviews, and their subsequent interpretation, by my thesis supervisor, which provided a second neutral opinion. The ability to judge and understand the coding and interpretation of data of another researcher is an alternative manner in which one can increase a research's confirmability (Hamberg, Johansson, Lindgren, & Westman, 1994).

## Results

**A note about the use of identifiers.** In order to provide as much lucidity as possible for the reader, participants are referred to as pseudonyms throughout this section.

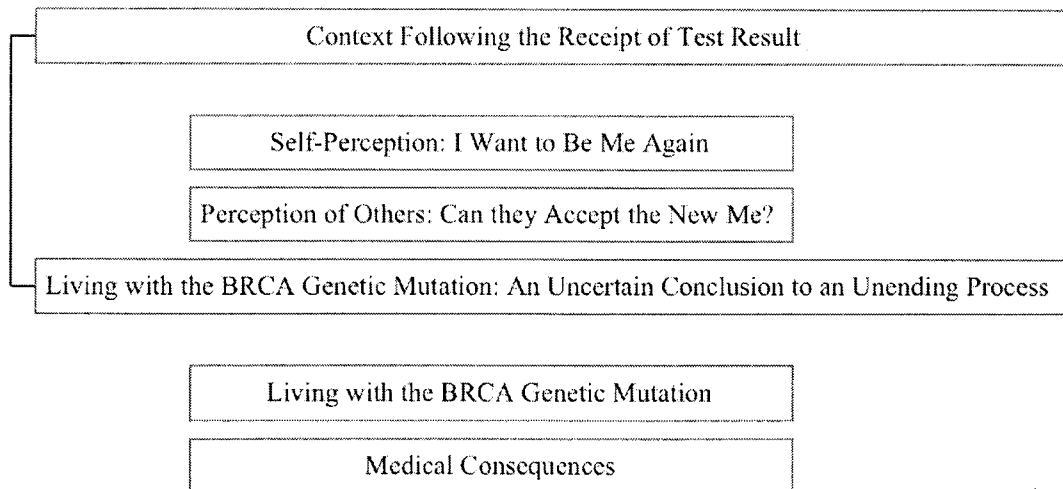
### Themes Identified

Through the analysis of six interviews, 126 raw data themes were identified, leading to the formation of two core categories, each containing distinct major themes, categories, and sub-categories (See Figure 1). For the purposes of this study, *core categories* will consist of the highest order classification, whereas *major themes* will be the second-highest order classification. *Categories* will consist of the third-highest level of classification, while *sub-categories* will be the fourth level of classification, which also encompasses the *raw data themes*, or codes.

More specifically, the first core category that was identified consisted of the *Context Following the Receipt of the Test Results*, which contained the following two major themes: Self-Perception, and Perception of Others. The second core category that

was identified was: *Living with the BRCA Genetic Mutation: An Uncertain Conclusion to an Unending Process*. The two major themes identified for this core category were Living with the BRCA Genetic Mutation, and Medical Consequences.

Each core category, major theme, categories, and sub-categories will be described in detail throughout the results section, with examples for each category being presented. Due to the large volume of raw data themes, each is not always mentioned or described, however those mentioned represent the category and sub-category accurately. Furthermore, verbatim excerpts will be presented alongside the raw data themes, so as to provide further support for the data, meaning for the reader, as well as to provide examples of participants' lived experience.



*Figure 1.* Two core categories derived from the data, as well as their respective major themes.

### **Context Following the Receipt of the Test Results**

Participants described that they had perceived the experience of receiving their positive test result as very significant event in their lives. Specifically, receiving a positive test result indicating that they possessed a mutation of the BRCA1 or BRCA2 genes generally affected participants' self-perception, and their perception of others (see Figure 2).

Participants' self-perception was found to be affected on a cognitive, emotional, and behavioural level. Specifically, emotions were found to have become increasingly negative, whereas cognitions and behaviours each had adaptive, and protective aspects.

Participants' perception of others was found to complement the descriptions they provided for their emotions, cognitions and behaviours, in that these also demonstrated a variety of negative aspects. Particularly, participants described that while those surrounding them could be viewed as a source of help and support, they also felt that these individuals were at times a source of distress.

**Self-Perception: I Want to Be Me Again.** Throughout the initial period following the receipt of their positive test result, participants described changes to their self-perception. Specifically, changes were reported in participants' cognitions, emotions, and behaviours.

Cognitions were reported to have adaptive and protective aspects, in that they either helped participants face reality, or sheltered their minds from the truth. Conversely, emotions appeared to be unbalanced; particularly, they were described to be solely negative. Finally, similarly to cognitions, behaviours were dichotomous in that they had

adaptive and protective aspects. For a detailed description of the raw themes belonging to each category and sub-category in the major theme of Self-Perception, see Appendix F.

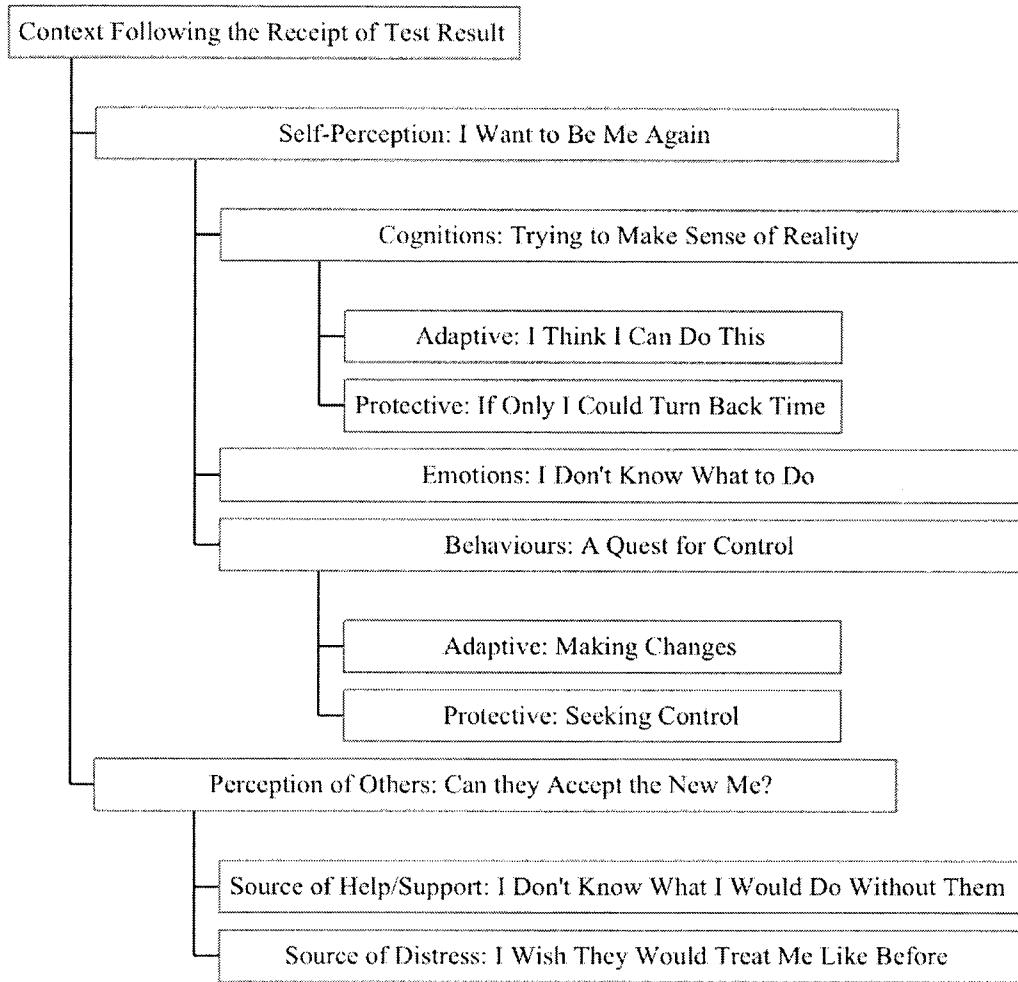


Figure 2. Core category (Context Following the Receipt of Test Result), as well its 2 major themes, 5 categories, and 4 sub-categories.

*Cognitions: Trying to Make Sense of Reality.* Following the receipt of their test results, participants generally engaged in adaptive or protective cognitions. Adaptive cognitions were those considered to help participants face and make sense of their new reality, whereas protective cognitions had the goal of sheltering the participants' minds from the stress and uncertainty surrounding the receipt of the test result.

Adaptive cognitions assisted participants in giving meaning to the new uncertain reality that came forth following the receipt of their test result. For instance, certain participants described being thankful for having received this test result, because since they were now aware, they could receive the medical care necessary in order to help prevent, or quickly detect, the development of cancer in the future.

On the other hand, certain participants stated that they hoped or wished to help others through the means of becoming an advocate for other carriers of the BRCA1 and BRCA2 genetic mutations, or for those who had already developed breast or ovarian cancer.

For instance, Linda described that since she would have appreciated additional support immediately after having received her test results, she would be willing to provide such support to others. She stated:

There could have been someone like right there that day? Like I would volunteer, you know, to do something like that. I would volunteer to...to just be on wait, you know, waiting there when someone's getting their results or you know...knowing that someone might be getting that that day. You could just say like, you can leave here and call that person right now. And talk to them, and set up a meeting with them.

Moreover, some participants reported that discovering that they possessed the BRCA1 or BRCA2 genetic mutation helped them feel empowered, as this allowed them to be proactive rather than passive.

Kiara described why discovering the news about her test result led her to feel empowered, rather than to perceive herself in a negative manner. She explained:

...as far as just knowing? Then no. I think if anything, it empowered me more to do something with myself instead of just, you know, floating along and thinking I'll be alive 'till I'm 80 something.

Although many participants described utilizing adaptive cognitions, many also engaged in protective thinking so as to guard their minds from the difficulties which are associated with this process. For example, one protective cognitive skill utilized by some participants was to consider ignorance as bliss. In other words, participants described that rather than know whether they possessed the genetic mutation, they wished that they had never received the results. Hence, they would not have known whether they possessed this predisposition to developing breast cancer, which would have allowed them to live their life without the stress and anxiety associated with testing, screening, and prophylactic surgeries.

For example, Linda described why she felt that she wished she had not received her test results. She explained:

No. I hadn't intended to find out...Ummm, not... for awhile. Maybe not until I was done having kids, because I didn't want... I didn't want the pressure of like- should I have them sooner? Closer together? Should I... I didn't want to be considering, you know, they've already started to talk to me- you know, when

you're done having your kids, you should have your ovaries removed, and that sort of thing. And, I didn't want to deal with that?

In addition to desiring that they could be unaware of their test result, participants also described that receiving this test result had made them contemplate thoughts they had not necessarily entertained in the past. Specifically, one participant described contemplating that one should have control over developing breast cancer. She stated that although she knew that she possessed the BRCA1 or BRCA2 genetic mutation, she nevertheless believed that if she ever did develop breast cancer, she would most likely feel as though this was something she was responsible for, and could have controlled. In particular, she stated that in the event that she develops breast cancer, she would most likely attribute it to not having exercised enough, or having eaten too many fatty foods.

Similarly, some participants described that this experience had led them to seriously consider what it would be like to develop breast cancer, and the effects this may have on them. In other words, the receipt of the positive test result, which leads to an increase in the potential of developing breast cancer, resulted in certain participants thinking that breast cancer could actually become part of their daily life.

Other protective cognitive methods included avoidance, justification, and minimizing. Avoiding entailed that a participant was evading a situation, such as a prophylactic surgery or even testing itself, even though she was aware of the potential benefits. Similarly, participants sometimes felt the need to justify actions or statements, when they felt that most individuals might not deem these appropriate. Finally, participants often also engaged in minimizing the importance of their test result, and it's meaning, rather than allowing their true emotions to emerge.

Many of these cognitions became evident when Avril described why she had waited to be tested, rather than being tested when she was aware of the genetic mutation in her family. She explained:

So, yeah. I guess, I guess, I think I've known about it but it's been very much like back burner stuff. And also I was having children, I was breastfeeding, and, I didn't want to do any testing until that [process]; that part of my life was over. Because I- there was nothing I was going to be able to do in terms of, like prophylactic surgery or, or even I can't even get, be properly, couldn't be properly screened when I was breast feeding, so, I waited until I was pretty much at the end of that and then I've started into the screening process.

In summary, participants described cognitive changes, which were both adaptive and protective, following the receipt of their test result. These cognitions, which appear to have influenced their self-perception, allowed them to become more familiar with their new reality, and sheltered their minds from becoming overwhelmed by the uncertainty that surrounded this period in their lives.

***Emotions: I Don't Know What to Do.*** Due to the receipt of the test result, participants often described an altered emotional state, often focusing on unpleasant emotions to illustrate their feelings. These feelings were reported as being as straightforward as guilt, or as complex and remarkable as feeling less like a woman. Participants described feeling guilt, for instance, due to potentially passing on their genetic mutation to their children, as well as regret for having been tested, as they felt they could have avoided this stressful and uncertain part of their lives.

On the other hand, some participants described that the experience of receiving the positive test result indicating that they possessed a mutation of the BRCA1 or BRCA2 gene, led them to contemplate things which would make them feel less like a woman. More specifically, Avril described the manner in which she perceived herself following the receipt of her test result. She stated:

...it makes you contemplate things that would make you feel much less like a woman, which is [an] oophorectomy, like getting your ovaries removed, you know, getting your breasts removed, but I mean that's obviously a pretty dramatic. That would; the thought of that having to happen certainly...impacts the way you think about yourself. And I guess in that sense... that that's; that that's something I have to take seriously; that I have to think about... I guess...I feel different about myself in that way?

Other participants, such as Kiara, who explained how she would feel if she would undergo a PM, also described the potentiality of feeling less like a woman. She stated:

Um... I think that, now I was discussing this with my husband; if I would've had a surgery, the prophylactic mastectomy done, I would've felt um, more self-conscious, um, about having done that type of surgery? I would've been, um...I think maybe that's why I'm reluctant to do it, is that the... the whole physical aspect of it- I would look different. That I would always want to, in a change room, hide myself, because I wouldn't look the same...

Similarly, the climate surrounding the receipt of the test result appears to have resulted in participants experiencing a variety of emotions, which illustrated the uncertainty that permeated this experience. For instance, participants described

themselves as feeling more vulnerable, generally to cancer, since the receipt of their test result. Likewise, participants felt more uncertain, and insecure regarding themselves and their health, following the receipt of this news.

Participants also explained that they felt powerless to their newly discovered genetic mutation, which had so greatly impacted their lives. For example, while describing her reaction to being identified as a carrier of a BRCA1 or BRCA2 genetic mutation, Carlie described her struggle with this feeling of powerlessness:

I'm very healthy, and I don't feel like I'm very healthy anymore? Even though I am. (laughs) You know, I'm a very healthy person; I run, I'm active. But I feel sometimes like I'm a ticking time bomb kind of? You know, that there's something in me that's just gonna... go. At any moment. And [there is] nothing I can do about it. So it's kind of disempowering because there's... I, you know, regardless of how much I exercise or eat, or take care of myself, it's just a genetic thing, so, you feel somewhat, uh...like it's completely out of your control.

In addition, participants also reported feeling a multitude of fears associated with the actual receipt of the test result, and its impending consequences, such as fear of their options, fear for their children, and fear of prejudice. For instance, participants often described being afraid of their options, which generally consisted of having their breasts and/or ovaries removed. This option was frequently presented to women by their physician following the receipt of their test result, and resulted in them experiencing varying degrees of fear.

Kiara described a meeting with her physician after she had received her positive test result. She reported her fears in having any prophylactic surgeries.

I question a little bit more on having your, your ovaries removed, and having to take, um, hormone replacement, and how that could affect...Early menopause, and all that, and I thought at [this age] I'm not ready to do the menopause. And I think that scared me more than having breast cancer. Um, I thought that's such a huge life change. That um, I, I wasn't ready for that. Uh, so I've kind of backed off from doing any type of surgery at the moment.

Participants often also stated that they were experiencing fear for their children, which often involved contemplating whether their children would also possess the BRCA1 or BRCA2 genetic mutation, thereby also predisposing them to develop breast cancer.

Additional fears presented by a number of participants were those of fearing other's reactions, and fears of prejudice to her possessing the genetic mutation. Certain participants described that they were hesitant to share their test result with others because they feared the manner in which others would react. For instance, participants feared whether others would pity them, or judge that they were not competent in their job due to an emotional weakness caused by the test. Furthermore, certain participants feared that they would experience prejudice due to their test result, either through others treating them differently, or difficulties with insurance companies in other provinces or countries. These fears of prejudice were sometimes also directed towards the participants' children.

Specifically, when Hannah described her parents' views of a cousins' illness, this led her to voice her fears of prejudice for her children's future. She stated her fears of:

...whether this will become a problem for [her] kids. Will someone not want to marry them because of the possibility of them passing on a gene to, to children,

you know, like is that gonna be a problem for them... you know, you wouldn't want them to choose somebody that they would not have otherwise married because, because of this, you know there's so, and I have no idea what to think.

In conclusion, participants generally reported feeling various disagreeable emotions due to their test result, which led to an altered perception of self. A particularly salient emotion and fear that was mentioned was that of feeling less like a woman. These altered emotions, as well as cognitions, seem to have influenced participants' behaviours following the receipt of this news.

*Behaviours: A Quest for Control.* As a consequence to receiving the positive test result indicating that they possessed a mutation of the BRCA1 or BRCA2 genes, participants often engaged in a variety of behaviours with the attempts of regaining control and seeking to reclaim a sense of normality in their everyday lives. Specifically, participants generally utilized either adaptive or protective behaviours to achieve this. Adaptive behaviours often consisted of behaviours, which appeared to help participants feel more in control of their reality, whereas protective behaviours involved precautionary behaviours that seemed to allow participants to regain a sense of normality.

Adaptive behaviours generally entailed that a participant was actively trying to make changes in her life that would allow her to regain some sense of control. Particularly, certain participants sought additional information by learning more about their options and preventative methods, allowing them to make the appropriate choice for them. Other participants described planning for the future, for instance by updating their will, or determining who would be their children's caregiver if they and their spouse would be unable to care for them.

In the same vein, some participants engaged in proactive behaviours, by changing their lifestyle, and engaging in further research so that they could make the best decisions for themselves for the future.

For example, Kiara described the changes she made to her home following the receipt of her positive test result. Furthermore, her illustration also demonstrated that the participants' proactive behaviour was not only adaptive for themselves, but sometimes also for their children. Kiara stated:

I basically went home, and we cleaned out the cleaners. My husband and I we researched, uh the chemicals that are in, um, our products, to see, you know the; there's so many products, chemicals that imitate hormones, especially estrogen... I thought well, with the girls, they're [young], I should start now. 'Cause for me, in my case, I'm [older], whatever happened in the past, I can't change. But, I knew I could do something for them.

Other adaptive behaviours that were discussed by participants were those of living in the moment, and taking opportunities. Specifically, certain participants stated that they tried to live in the moment, so that they could appreciate every day and focus on the positive. Similarly, some participants stated that they made an effort to take more opportunities, so that they could ensure that they achieved what they would like, regardless of what happened in the future.

For instance, Hannah described her and her husband's views of taking opportunities. She stated:

[My husband and I] always sort of felt like you know, if you were happy, you should just do...you should uh; I guess you know, even then, I sort of felt that,

you never know? What's coming. And that if you're happy you should just, you know, go with that and, uh, do the things you want to do. So, um, that I guess, this [test result] has sort of...helped me to sort of realize; to sort of solidify that for myself?

Although many participants engaged in adaptive behaviours, many also described utilizing protective behaviours to cope, following the receipt of their test result.

Protective behaviours were typically behaviours that helped participants maintain a sense of normality following the receipt of their test result. More specifically, although these behaviours had shielding properties, they nevertheless allowed participants to maintain a grip on reality at this stage of their lives.

A particularly widespread protective behaviour described by participants was that of attempting to control the future. Particularly, some participants hoped that by making certain changes in their lives, for instance with regards to diet, or by sheltering their children from the news of the test result, that they could maintain the life that they had prior to receiving the test result.

For example, Avril described why she did not feel that telling her children about the genetic predisposition would necessarily benefit her children until much later in the future. She explained:

And I feel like, you know, children whose parents have no genetic predisposition also get cancer, so if it happens, then we'll deal with it then, but I don't, I don't feel like there's, there's any advantage, for discussing with them right now. And I will discuss it with them later because obviously they're going to have to make decisions about genetic testing, so, but I, that would be much later.

Similarly, certain participants explained that they were afraid to form closer bonds with others, for fear of loss. For instance, a participant mentioned that while she appreciated support, she did not want to join any support groups, or meet anyone who may also possess the genetic mutation, in case these people would develop cancer, which would result in her experiencing additional loss.

In short, following the receipt of their positive test result, participants engaged in both adaptive and protective behaviours. These behaviours allowed participants to feel more in control of their reality, as well as regain a sense of normality, following this significant change in their lives. The dichotomy observed in participants cognitions and behaviours following the receipt of their test result, also appears to have been present in their perception of others.

**Perception of Others: Can they Accept the New Me?** A modified perception of others appeared to have emerged following the participants' receipt of their positive test result. More specifically, participants generally either viewed others as a source of help and support, or as a source of distress. Others were often perceived as a source of help/support when they were able to offer something to the participant. On the other hand, others were also seen as a source of distress, as participants perceived them as threatening to their secret, and stigmatizing towards themselves.

These modified perceptions of others mirrored the previously mentioned altered cognitions and behaviours reported by participants. Similarly as these processes tended to have adaptive and protective properties, participants' perception of others became equally distinct following their receipt of their positive test result. For a detailed description of

the raw themes belonging to each category in the major theme of Perception of Others, see Appendix G.

*Source of Help/Support: I Don't Know What I Would Do Without Them.* When participants perceived others as a source of support, they judged others as able to help them cope and live with their test result. The support described by participants often stemmed from a participants' spouse, friends, and other BRCA1 or BRCA2 genetic mutation carriers. More specifically, participants explained that their husbands were a significant part of their support structure. For instance, they generally attended the appointment during which participants were given the news that they possessed the genetic mutation predisposing them to develop breast cancer, as well as further follow-up appointments for screening. On the other hand, some participants explained that they considered their friends as support, as they were very understanding of the test result, and were present and empathetic during times of need. Similarly, participants tended to perceive other BRCA1 or BRCA2 genetic mutation carriers, whether part of their families or not, as providers of support. Participants generally described that these individuals were better able to understand the meaning behind possessing this genetic mutation.

For instance, Brigitte described her special bond with a co-worker who had breast cancer and had also received a positive test result indicating she possessed the BRCA1 or BRCA2 genetic mutation. Brigitte explained:

So I, as soon as she, as soon as she was diagnosed with breast cancer I actually felt a bond with her. And then to find out after; because when she would talk to me I already knew all this stuff, and of course she, she didn't know I was BRCA2

positive. Um, until pretty much when she was through all her chemo and radiation and then, and then she started talking about getting tested and stuff and that's when I told her; 'cause like I said I don't tell people that I'm (laughs); I don't tell people... Well I thought that she could- yeah exactly. I thought she would understand...

Participants also appeared to perceive a great deal of support being provided to them through the medical team. They often described that their physicians, and at times nurse at the clinic, were a very important source of support, as he or she provided them with care and a listening ear.

In general, participants viewed others as a source of support or help when they perceived them as being able to help them cope and support them through this time of need. While participants described perceiving others as a source of support, they also delineated instances when they perceived others as a source of distress.

***Source of Distress: I Wish They Would Treat Me Like Before.*** When participants perceived others as a source of distress, they generally saw them as threatening to their secret, as well as different from themselves. More specifically, participants often described perceiving others as insensitive to their problem, or as pressuring them to take a different course of action. For instance, participants perceived external pressure from family or physicians to undergo prophylactic surgeries.

In addition, many participants reported perceiving others as judgmental or misunderstanding towards their having been tested, and towards their choices regarding surgery and treatment. Similarly, certain participants described that they perceived their families as ignorant towards their beliefs about testing and treatment.

For instance, Brigitte, who had sisters who were also tested, described the manner in which her family viewed her and her sisters' choice of undergoing testing. She explained:

I think it's important. I have to tell you that a lot of people in my family think it's not important, and a lot of people haven't been tested in my family. So... but it's funny, my sisters and I; immediately. Like, there was no question.

Additional perceptions of others described by participants were those of perceived pity, avoidance from family, and insensitivity. Specifically, participants sometimes explained that they believed others would pity them if they would tell them about their positive test result. Similarly, some participants described that they sometimes felt that their family avoided them because of the test result, or their choices resulting from the test. On the other hand, certain participants believed that their physician or family member were insensitive, because they suggested that participants should undergo prophylactic surgeries, or be more active in their treatments.

For instance, Carlie described an incident where she perceived that her physician was behaving insensitively when he suggested that she should undergo a PM. She stated:

I found it, it was actually quite shocking to me because I was; had just had a baby and was nursing (laughs), and he basically said, you should consider having a double mastectomy, and I, I just found it very odd, because my baby was like a month old, you know, sitting beside me... (laughing). But it was a man. He wouldn't think of those kinds of things...

To summarize, participants not only perceived others as a source of help and support following the receipt of their test result, but they also at times perceived others as

a source of distress. However, the contrast between these two perceptions appeared to have been present due to participants' perception, regardless of the behaviour of others. In particular, while some participants did not find the suggestion of undergoing prophylactic surgeries as insensitive or pressuring, others were deeply disturbed and upset by these recommendations. Hence, it appears factors other than others' behaviour may have been responsible for the perception of participants with regards to others.

### **Living with the BRCA Genetic Mutation: An Uncertain Conclusion to an Unending Process**

The period following the receipt of a positive test result indicating a woman possesses a mutation of the BRCA1 or BRCA2 genes appears to have had consequences on participants' self-perception as well as their perception of others.

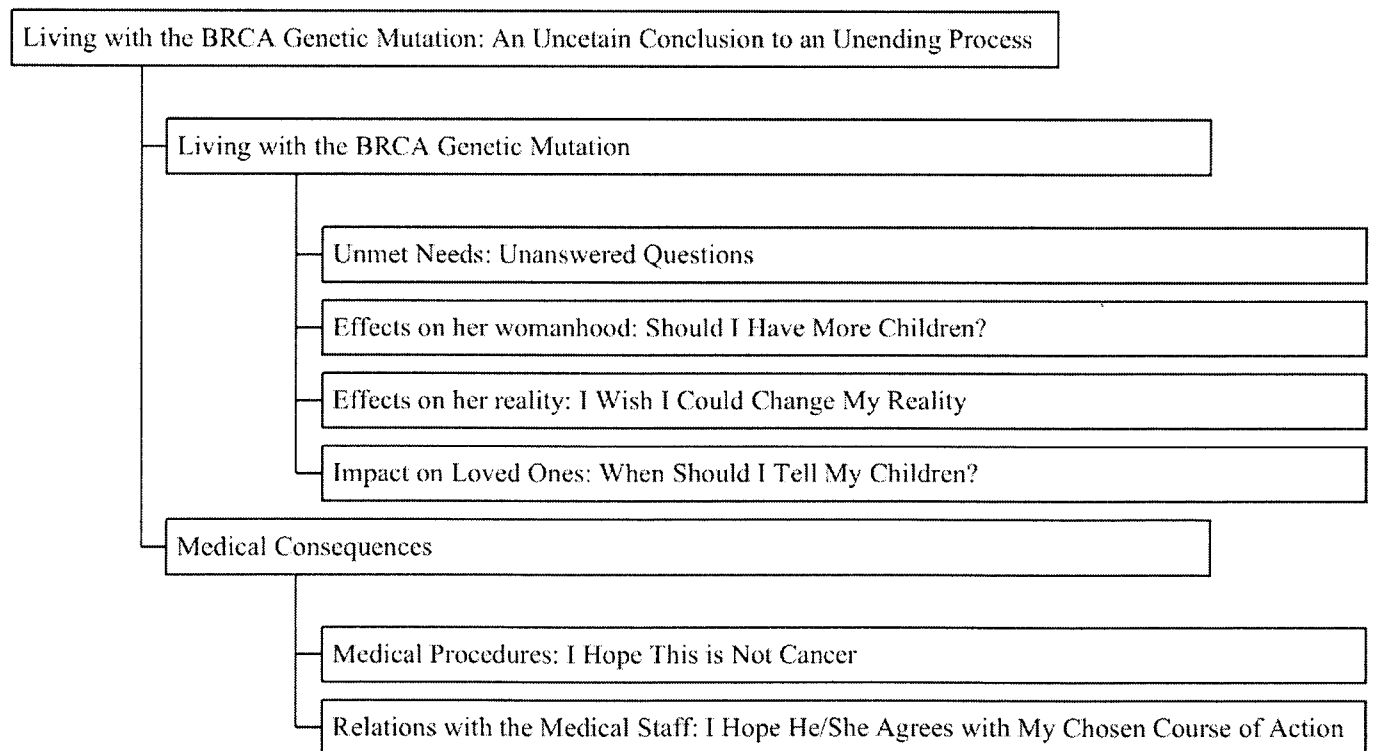
After the initial period of receiving this result, participants eventually learned to live with this news and its consequences. Nevertheless, it appears that no conclusion could be foreseen to this stressful and uncertain period in participants' lives, as the only potential termination to the uncertainty would be that participants develop breast or ovarian cancer.

Nonetheless, participants were able to reflect on the consequences of Living with the BRCA Genetic Mutation, as well as the Medical Consequences associated with this. Participants described that living with the BRCA gene mutation led them to feel that they had unmet needs, that their womanhood was affected, and that their reality was also affected. Not only did living with the test result affect participants over time, but it also affected their loved ones.

Finally, as a consequence to receiving and living with their test result, participants' interactions with the medical staff became more prevalent, and underwent changes. Similarly, knowing that they possessed the gene also led participants to undergo certain medical procedures (see Figure 3).

**Living With the BRCA Genetic Mutation.** The uncertainty associated with living with the BRCA genetic mutation led participants to experience a multitude of consequences. Specifically, these consequences took the form of: unmet needs, effects on the participant's womanhood, effects on her reality, and impacts on loved ones.

For a detailed description of the raw themes belonging to each category in the major theme of Living with the BRCA Genetic Mutation, see Appendix H.



*Figure 3.* Core category (Living with the BRCA Genetic Mutation: An Uncertain Conclusion to and Unending Process), as well its 2 major themes, and 6 categories.

*Unmet Needs: Unanswered Questions.* Living with the BRCA genetic mutation often resulted in participants realizing that they had many unmet needs due to this experience. Particularly, various participants described that since they had received their test result, even if they had been aware of the result for years, they were searching for guidance through the medical staff, or through research.

For instance, Avril described an experience of searching for guidance through her physician, as she was unsure of how to proceed living with the genetic mutation and its potential consequences in the future. She explained:

[I'm] looking into my options of you know, if I'm going to look at getting an, uh, ovarian removal; what is that going to look like. You know, just sort of maybe if I get a mastectomy, like, what are the; what are my risks, for that. I, I've started the ball rolling with those things, like I'm signed up to go to a menopause clinic out at [another hospital], or, which should be, should be happen; I mean I; when I, when I talked to [my specialist] about it it was supposed to be happening in a couple of months or something, so... Just try to lay some groundwork for making decisions...

Furthermore, some participants explained that throughout this experience they were receiving inadequate support, wishing that more support were available to them, or that they could discuss their medical options with additional individuals.

For example, Kiara explained that living with the test result has led her to feel that many individuals do not share her views on prevention, and even though she sought support, she continued to feel as though additional support was needed. She stated:

Um, there's a, Facebook has a, a bunch of different groups where people blog, and they just talk about their different experience, and so on... Um, I've read some of them, and yeah, some of them, uh, I could see where we have similarities about, how we want to change our lifestyle, and, some of them I just don't seem to, to get. You know they... To relate to them. Um, I would like to meet other women who have, um, this gene mutation, just to find out what they're doing, you know- people that are local, um, maybe if there's a support group somehow that we can discuss the different, um... options together, and decide, you know, just to have a support group, but um, that's yeah.

In summary, participants described that living with the BRCA genetic mutation made them feel as though they had unmet needs, due to this experience. These unmet needs generally consisted of a lack of guidance, and inadequate support, which participants continued to search for years following the receipt of their test result.

*Effect on her Womanhood: Should I Have More Children?* In addition to unmet needs, certain participants described that living with the positive test result for the BRCA1 or BRCA2 genetic mutation had also had an effect on their womanhood. In other words, participants re-questioned certain fundamental characteristics that are typically considered as part of being a woman. Specifically, some participants thoroughly considered a desire to conceive after testing, and the ethics of childbearing following testing.

For instance, one participant contemplated that she wanted another child, as a consequence of receiving her test result. This desire was reported to have persisted for years following testing. Similarly, this led to a consideration of whether conceiving of a

child following testing was ethical, as the parent would be aware of the possibility of passing on the genetic mutation to their unborn child.

The desire to conceive, years after receiving and living with her test result, was described by participant Hannah, who after being recommended to have a hysterectomy, thought:

...if I'm going to be having to have an operation like that anyway, maybe first I would like to have another child. And, that was something that I really hadn't wanted up until then... So it was sort of surprising to suddenly; that was... I felt that that would [be] sort of a positive outcome of this whole...uh, discovery?

Hence, the experience of living with the BRCA genetic mutation had a significant impact on certain women, in that it affected their desire to conceive, and made them consider whether it was ethical for themselves or other carriers to bear children knowing that they possessed the potential to pass on the genetic mutation.

***Effect on Participants' Reality: I Wish I could Change my Reality.*** In addition to having effects on the participants' womanhood, living with the BRCA genetic mutation was also found to have had effects on some participants' reality. Particularly, living with the BRCA genetic mutation sometimes forced participants to face reality, made them wish they could change their reality, and sometimes led them to experience relief knowing the truth about their reality.

For instance, there were times when participants described that living with this genetic mutation forced them to face reality, even though they may not have wanted to, for instance having to cope with potential scares as a result of screening. Similarly, many participants explained that they wished that they could change their reality, of knowing

and living with this test result. On the other hand, certain participants described relief at knowing the truth about the test result and facing reality, as it allowed them to be prepared for potential outcomes in the future.

Avril, who explained the potential impact of this test result on her family, described the wish that she could change her reality. She stated:

I mean and a lot of that to with like my children. I don't, I don't want it to like continue down the line, you know, it's like, you know when you get to be a parent it's like it's bad enough that you have to sort of bear the consequences of your genetic history, but then to have also passed it along is, not a great feeling.

In conclusion, participants described that living with the BRCA genetic mutation had affected their reality, and views of reality. In particular, this experience at times forced them to face reality, made them wish they could change their reality, and at times led participants to experience relief knowing the truth about their reality.

***Impact on Loved Ones: When Should I Tell my Children?*** In addition to having effects on the participant, living with the test result was also described as having had consequences on loved ones. In particular, participants described that receiving and accordingly living with this test result had had consequences on the manner in which they would raise their children, had impacted other family members, led to familial discord, and raised many questions regarding the potential testing of their children once they reached adulthood.

Specifically, participants often explained that ever since they had discovered that they possessed the genetic mutation, this experience had impacted the manner in which they would raise their children in the future. For instance, participants described that they

would, or had been, teaching their children to eat more healthily, so as to hopefully prevent the development of breast or ovarian cancer in their future.

This experience sometimes also impacted other family members, such as causing participants' families to worry about their health. On the other hand, for certain participants, a consequence of receiving and living with this news was that of familial discord. For example, some families did not agree with the concept of genetic testing, believing that nature should take its course, while others did not agree with the course of action certain participants chose, so as to try and prevent the development of cancer in their everyday lives.

For some participants, living with the positive test result raised many questions regarding the potential testing of their children once they became adults. Participants struggled with when testing should take place, and whether they should wait to tell their children of the test result until after they had their own children. Often, participants attempted to protect their children from the future, by keeping the secret of the test result.

Brigitte, the only participant who said she had told her children of the test result, explained that while she told them, it was very casually. She described:

I've told them, but I've told them very lightheartedly because they're young and I don't want them to worry about it yet. I don't; I just don't think that's necessary, they can learn about it as they get older. Um, my older daughter is; she takes it a little more serious than my younger daughter... She's not overly worried about it, but that's partially because I, I don't let her be yet... I wouldn't want to know when I was [her age].

Hence, living with a positive test result indicating that a woman possesses a mutation of the BRCA1 or BRCA2 genes not only impacted participants in their everyday lives, but also impacted their loved ones, especially their children and immediate family.

**Medical Consequences.** Living with the news of possessing a mutation of the BRCA1 or BRCA2 genes was generally considered an uncertain process with differing outcomes for these participants. Furthermore, participants described that living with the test result generally engendered both medical procedures, as well as affected relations with the medical staff.

***Medical Procedures: I Hope This is Not Cancer.*** Medical procedures became a sure result of living with the positive test result, and generally entailed either the removal of participants' ovaries, or potential scares.

More specifically, one participant reported having had a prophylactic oophorectomy as a precaution to reduce her chances in developing breast cancer. On the other hand, multiple participants had experienced what they often described as a scare, or potential detection of cancer, following routine examinations, which became commonplace following the receipt of their test results.

For instance, Linda described her experience with having been told by her physician that she needed additional testing because he was concerned about a previous test result. She stated:

Right now I'm awaiting to have an MRI biopsy on something that they found, and, I really hope that it's nothing, and we can just go on after that... I was here just last week for a mammogram and an ultrasound and they told me they didn't

find anything, but I still have to have this MRI... Well now I'm just sitting on my hands.

Similarly, Carlie described a comparable experience to that of Linda's. She described the manner in which her life now included frequent testing and screening, and her feelings regarding a potential scare she once had. She explained:

Emotionally now, I don't even think about it...between tests. And then for about 2 weeks before the tests, I'm nervous (laughs), and thinking about it again. I'm thinking, oh, I haven't been doing my examinations (laughing). And then, after the tests; I had a one scare uh, but, something showed up, but I'm now learning that because we're such high risk, they check every possible little thing.

The experiences of these women appear to describe the uncertainty associated with living with the BRCA genetic mutation, which now permeates their lives. This uncertainty was particularly evident when participants described potential scares they experienced following routine screenings, and the manner in which this often made them feel helpless; hoping to receive the news that they would continue to be cancer-free.

***Relations with the Medical Staff: I Hope He /She Agrees with My Chosen***

***Course of Action.*** The increase in medical procedures that participants underwent while living with the BRCA1 or BRCA2 genetic mutation also affected the number of interactions they had with the medical team that surrounded them. Furthermore, living with this test result also ensured that these participants encountered additional new medical professionals, such as surgeons, or cancer specialists. These interactions were at times deemed negative, and other times positive. Specifically, some participants

described that they were frustrated with the medical team, while others explained that they felt that they were treated with superior care.

One participant, Hannah, described an experience when she felt frustration while speaking to a physician who did not seem to agree with her course of action with regards to surgical procedures. She said:

I would have to go to the doctor for various things, and one doctor I saw in the clinic, sort of said to me; I explained the situation, he sort of said 'oh, and you haven't had the hysterectomy already?!'. And he was, he was totally shocked- that I hadn't already had it done, and seemed to have felt that as soon as I had known that I had this, that I should have immediately run out...But that, that was sort of an interesting reaction.

On the other hand, some participants described that they felt that being a part of this clinic made them feel as though their physicians and the medical staff treated them with superior care. Linda described this experience:

...the only place where I'm affected by it is here, and, I get treated better...So I'm not complaining about that. No, you know, I have a cousin who's desperate to get the testing done, 'cause she's thinking, yeah, I want to get front of the line, so...

In summary, as a result of living with the test results, participants' relations with the medical team increased. Particularly, these increased interactions were sometimes judged positively, while others were viewed negatively. Nevertheless, medical interactions became commonplace for these participants, as the uncertainty surrounding the development of cancer continued.

## Discussion

The purpose of this study was to better understand the experiences of women who tested positive for a mutation of the BRCA1 or BRCA2 genes, as well as to comprehend the way in which they experienced stigma in their daily lives due to their test result.

Through the current research findings, it is implied that women who possess mutations of the BRCA1 or BRCA2 genes undergo a very significant experience when receiving the test result. Furthermore, they find that this experience is continuous and fraught with uncertainty, as it appears to have no certain ending.

Although women who possess this genetic mutation are not sick, they are intended to continue with their daily lives even though they must undergo specific screening procedures and medical appointments on a regular basis in order to hopefully quickly detect, and henceforth prevent the development of cancer.

This chain of events appears to be rather disconcerting to these women, as they are aware that they are healthy, yet they possess the potential to become very ill with a disease that is rather stigmatized, cancer. More specifically, these fears appear to permeate BRCA gene mutation carriers' lives, as they begin to contemplate having cancer and the repercussions surrounding this possibility.

### **Context Following the Receipt of the Test Results**

**Self-Perception and Perception of Others.** After undergoing genetic testing and receiving a positive test result indicating they possessed a mutation of the BRCA1 or BRCA2 genes, participants appear to have been transformed. More specifically, they experienced transformations to their self-perception, as well as their perception of others.

Rather than perceive themselves as they had previous to the test result, participants began to perceive themselves as someone who had the potential to become ill at any moment. Their outlook on life, their goals, and their priorities were all altered due to this test result. Their cognitions became either adaptive or protective, and generally facilitated their ability to face the reality that they possessed this genetic mutation, while sometimes allowing them to protect their minds from the difficulties of facing this truth.

On the other hand, their emotions regarding what they perceived as a new version of themselves became increasingly negative. They began focusing on what might have been if they had not decided to undergo testing, or what might be such as if they did develop cancer. Participants also described that being aware of this test result had made them contemplate things that would make them feel less like a woman, such as losing their breasts and/or ovaries. These potentially significant surgeries were perceived as a loss of the participant's womanhood, in that if they would take place, they would make her look different, feel different, and be unable to conceive of a child.

Participants' behaviours were consistent with this altered perception of themselves, as their new goals were now to be proactive and attempt to control the development of cancer through eating healthy, exercising, and being screened. However, participants also attempted to engage in behaviours that would allow them to feel more in control of this tumultuous experience, and regain a certain sense of normality in their lives. For instance, by attempting to control the future, and attempting to keep the secret of their test result:

Following the receipt of their test result, participants underwent transformations in the manner in which they perceived others, in addition to themselves. As they now

perceived themselves differently, it seems as though they felt others also perceived them in a different light. Although they often felt that others were a great source of help and support, they also often perceived them as judgemental, misunderstanding, pitying, rejecting, and avoiding.

**Findings in Comparison to Previous Research.** The experiences reported by the women in the current study are significant in that they closely resemble the experiences of individuals who have been diagnosed with cancer. Particularly, in a seminal article, Taylor (1983) devised a theory of cognitive adaptation to describe the experiences of individuals who are faced with life-threatening events, such as cancer. She explained that individuals' adjustment centers around three general themes. Those of: seeking meaning in the experience, regaining mastery over the event and the individual's life, and attempting to reinstate self-esteem through self-enhancement.

Although these themes were devised after having interviewed  $N=78$  women who had breast cancer, these women's experiences nevertheless seem to closely relate to the experiences that were described by the women in the current study, even though they had not received a diagnosis of cancer (Taylor, 1983). As was reported in Taylor's (1980) study, participants in the current study attempted to give meaning to their experience, for example, through attempting to find the source of their gene mutation, and describing the implications that this test result had had on their lives.

More specifically, similarly to the women in the Taylor (1983) study, the participants in the current study often explained that they were very surprised to discover that they possessed the BRCA1 or BRCA2 genetic mutation. Although they were aware of the 50% chance of inheriting this mutation, they nevertheless were very shocked to

uncover that they also possessed the gene mutation if their sisters had been identified as carriers, for instance. Participants in the current study also often continued to provide explanations of where they believed their gene mutation may have originated, and who had suffered from cancer in their families.

On the other hand, participants in the current study gave meaning to their experience through describing that they now attempted to take more opportunities, would live in the moment, and that their priorities had been modified; findings that were also reported by Taylor (1983).

Taylor (1983) described that women, who had been diagnosed with breast cancer, attempted to regain mastery of this event and their lives by attempting to gain control. For example, Taylor (1983) stated that many of the participants in her study believed that they could prevent the recurrence of cancer through a change in diet or exercise, or through acquiring additional information. These findings coincide with those obtained in the current study, in that the current study's participants often explained that in order to help prevent the development of cancer, they had begun exercising, eating more healthy, and even eradicating chemicals around their home. Furthermore, many participants in the current study sought additional information through research and approaching new professionals.

Although the women in the current study did not engage in self-enhancement in the same manner as those in the Taylor (1983) study, the current participants nevertheless did attempt to make their plight look less difficult than other BRCA1 or BRCA2 gene mutation carriers'. More specifically, one participant in the current study who was asked about having children following testing, for instance, described that she was very glad

that she did not have to deal with this ethical conundrum and seemed to pity other BRCA1 or BRCA2 carriers that did. On the other hand, some participants described that they considered themselves fortunate because they had a good support system, unlike some other women who possessed the BRCA gene mutation. Finally, some participants stated that they did not feel stigmatized, but if they were to undergo any prophylactic surgeries, that this status would surely change.

Taylor (1983) states that these downward social comparisons have the ability to help individuals affected by life-threatening events to feel more positively about their current situation, as they think that others are less fortunate than them.

Taylor (1983) concludes her paper by stating that the themes of meaning, mastery, and self-enhancement each necessitate a certain amount of illusion. Particularly, the causes attributed to developing cancer, or in the current study- receiving a positive test result, are often illusory. Furthermore, the manner in which individuals attempt to gain control over developing cancer, through changing their diets for instance, are often unrealistic. Finally, even if no individual with whom one can compare themselves exists, which was often the case in the current study, downward comparisons are nonetheless still made with a person who is made up (Taylor, 1983). She explains that these illusions allow those who are faced with life-threatening events to adapt in a beneficial way.

The striking similarities between the study conducted by Taylor (1983), and the current study, demonstrate that the experiences of women who have been identified as possessing the BRCA1 or BRCA2 genes, may in some cases closely resemble those of women who have already been diagnosed with cancer. However, the significant difference between these two studies is that the women in the current study did not

possess cancer, and had an up to 85% chance of developing this disease over their lifetime.

Studies, which have been conducted on BRCA1/2 gene mutation testing and its effect on women's self-concept, have generated comparable results to those of the current study. Particularly, researchers d'Agincourt-Canning (2006) and Esplen et al. (2009) conducted two separate studies in which they examined the construct of self-concept in BRCA1/2 gene mutation carriers, and discovered that these women can have an altered self-concept following genetic testing.

While the study conducted by d'Agincourt-Canning (2006) was qualitative, it nevertheless involved participants who were from varying age groups (25-60 years), women who had not tested positive for a BRCA1/2 gene mutation, and women who had previously had cancer. These factors may have affected results.

On the other hand, the study conducted by Esplen et al. (2009) involved the construction of a scale intended to measure self-concept in BRCA1/2 gene mutation carriers. However, this study was quantitative in nature, and test-retest reliability had not been established, indicating that further testing is required in order to ensure that similar results would be yielded when administering the test to other BRCA1/2 gene mutation carriers.

Previous studies, which have been conducted on the psychological impacts of genetic testing for breast cancer, have also yielded similar results to those found in the current study. More specifically, Croyle et al. (1997), Di Prospero et al. (2001), Dorval et al. (2000), and Lerman et al. (1995) reported that participants who were asked to consider receiving, or who had actually received a positive test result indicating that they

possessed the BRCA1 or BRCA2 genetic mutation, anticipated to feel or actually experienced a variety of negative emotions. Participants in these studies and in the current study reported to have experienced emotions such as increased depression, anxiety, worry about cancer, anger, and distress.

However, while many participants in the current study described experiencing regret following the receipt of their positive test result, it appears that this experience is not necessarily common amongst all women who receive the positive test result (Di Prospero et al., 2001).

More specifically, the participants ( $N=24$ ) who participated in a quantitative study conducted by Di Prospero et al. (2001) stated that they were satisfied with their experience of testing, and that they did not regret the decision of being tested.

On the other hand, participants in the current study often mentioned being proactive in attempting to prevent and quickly detect breast cancer. Similar findings were reported in a study conducted by Di Prospero et al. (2001), whose participants stated they would be engaging in lifestyle changes as a method of preventing the development of breast cancer.

Although many negative emotions and preventative behaviours were reported to have been experienced by other BRCA mutation carriers who participated in the previously mentioned studies, it seems as though these women did not experience additional fears such as: fear of other's reactions, fear of prejudice, fear of a child's resentment, fear of spilling her secret, and fear of making changes. Similarly, participants in the previously mentioned studies did not describe experiencing altered cognitions such as wanting to be treated like others, wanting to be in control, avoidance, justifications,

and wanting to help others, such as the participants in the current study (Croyle et al., 1997; Di Prospero et al., 2001; Dorval et al., 2000).

Similarly, no participants in the previously reviewed studies examining the experience of BRCA gene mutation carriers were found to have stated that this experience made them contemplate feeling less like a woman (d'Agincourt-Canning, 2006; Croyle et al., 1997; Di Prospero et al., 2001; Dorval et al., 2000; Esplen et al., 2009; Lerman et al., 1995). This finding is particularly significant as it has serious implications, particularly if these feelings were found to be widespread amongst other BRCA gene mutation carriers.

Conversely, as in a study conducted by Di Prospero et al. (2001), participants in the current study had generally shared their test results with family. Furthermore, similarly to participants in Di Prospero et al. (2001)'s study, the current study's participants also hesitated to share these results depending on the age of the family member(s), as well as depending whether they believed this news would induce fear in the relative.

Further similarities were observed between results obtained by Di Prospero et al. (2001) and the current study. More specifically, as in the study conducted by Di Prospero et al. (2001), participants in the current study also expressed that they appreciated support from similar others.

Interestingly, participants in the current study often perceived others as a source of distress, feeling their families, friends, and even physicians were treating them in a manner that they perceived to be different than prior to the receipt of their test result. This finding, alongside the changes in cognitions, emotions, and behaviours, suggests that the

participants in this study may have been suffering from stigma. More specifically, these women at times described feeling judged, misunderstood, rejected, avoided, and so on, by individuals who were aware of their positive test result, such as their physician, friends, and family. In these instances, participants may have been dealing with a discredited, or visible stigma (Goffman, 1963).

However, participants also described instances where they were frightened for themselves or their children, in that they wished to keep their test result a secret, for fear of discrimination, being pitied, or being treated differently. Since participants' positive test result could not be perceived by others unless they chose to inform them, in these instances, the possession of their test result became a discreditable stigma, or one that is imperceptible to others (Goffman, 1963).

These findings were similar to those obtained by Camp et al. (2002), in that they investigated stigma and self-esteem in women suffering from stigmatizing mental health problems, a discreditable stigma. These women, similarly to the women in the current study, engaged in certain selective disclosure behaviours, as well as avoidance, so as to manage this stigma.

On the other hand, dissimilar results than those obtained in this study were observed in a research conducted by Wilson and Luker (2006). These researchers found that cancer patients tend to feel uneasy when not in the hospital, which they consider a safe environment. It follows that, cancer patients in the oncology ward can be identified as individuals who are suffering from cancer. Similarly, participants in the current study who were being screened and meeting with their specialist physician in a designated ward were also identifiable as female patients who were at high risk for the development of

breast cancer. However, unlike the patients in the study presented by Wilson and Luker (2006), the women in the current study generally felt as though they had been stigmatized by their physician or by the medical staff at one time at the hospital, rather than feel as though the hospital was a safe haven.

Nevertheless, the findings reported for the two previously mentioned studies involved the experiences of patients who had mental health problems and cancer, not a BRCA1 or BRCA2 genetic mutation, and their experiences may therefore not necessarily be interchangeable.

A study conducted by Esplen et al. (2009), on the construction of a scale intended to measure self-concept in women who have undergone BRCA1/2 gene mutation testing, did include a construct identified as 'stigma' in the completed scale. However, it is unclear whether the women in the current study also described the concept of stigma in the same manner, or how they would score on Esplen et al. (2009)'s self-concept scale, as the current study was qualitative and involved open-ended questions.

Similarly, in a qualitative study conducted by Kenen, Arden-Jones, and Eeles (2006), these researchers describe that the young BRCA1 and 2 gene mutation carriers who participated in their exploratory study described feeling lonely, feeling different, feeling isolated, and devastated, following the receipt of their test result. In addition, these women utilized silence and verbal discretion with regards to communicating the fact that they possessed this genetic mutation.

Although the researchers from the Kenen et al. (2006) study do not necessarily identify the experiences of the young participants as stigma, many of the experiences described by the young women in this study could be identified as stigmatization, or fear

of stigma. Despite having certain similarities with the results obtained in the current study, the Kenen et al. (2006) study's limitations must be taken into consideration. Specifically, the women in this study received genetic testing as a result of having been diagnosed with breast cancer. Even though these women were relatively young when they received their positive test result, the fact that they had been previously diagnosed with breast cancer may have affected the manner in which they perceived their experience, making the results less comparable to the current study's.

Interestingly, a study conducted by van Oostrom et al. (2003), which was conducted with BRCA gene mutation carriers, found that participants who experienced additional distress had reported less communication with family about the test, and changes in familial relationships. These results are applicable to the women in the current study since some described that they had experienced avoidance from family and familial ignorance, since the receipt of their positive test result. Hence, it is possible that the lack of familial support may have affected the manner in which these women coped with the receipt of their test result.

This discovery that BRCA gene mutation carriers may be suffering from stigma is particularly noteworthy, as the women in the current study were not suffering from cancer, nor had they ever suffered from this disease. The women in this study only had a more elevated chance of developing breast and/or ovarian cancer, yet their social behaviour was similar to the behaviour of those who are already afflicted by cancer.

In summary, participants in the current study experienced changes in their self-perception, as well as their perception of others. Furthermore, while certain aspects of their experience were similar to those experienced by other BRCA1 or BRCA2 gene

mutation carriers', certain novel findings were uncovered, such as feeling less like a woman, and the experience of a variety of fears. Finally, it seems as though the participants in the current study experienced some form of stigma, similarly to patients with cancer.

### **Living with the BRCA Genetic Mutation: An Uncertain Conclusion to an Unending Process**

**Living with the BRCA Gene Mutation and Medical Consequences.** The findings obtained in this study imply that following the initial receipt of their positive test result, BRCA gene mutation carriers generally experience changes in their self-perception and their perception of others. Furthermore, it seems as though these women experience the receipt of their test result as though they had already been diagnosed with cancer, in addition to feeling as though they are members of a stigmatized group. However, through results, it is also suggested that these women learn to live with the news of the positive test result, as well as its consequences.

It was suggested through findings that living with this gene mutation generally impacted these women through unmet needs, by affecting their womanhood, by affecting their reality, and by impacting those they loved. For instance, after reflecting on their lives since receiving the news of the positive test result, participants felt as though this experience had left them with certain unanswered questions, and left them feeling unfulfilled. On the other hand, through living with the BRCA genetic mutation emerged the development of certain previously undesired aspirations, such as feeling a need to conceive.

These experiences in turn affected the manner in which participants viewed reality. For example, these women were at times forced to face reality, and often wished they could change their reality. Finally, it seems as though all of these effects on participants while living with this genetic mutation tended to impact their loved ones, either through the manner in which they treated others, or their families' fears or frustration with them for being influenced by this situation.

On the other hand, through results it was suggested that living with the BRCA genetic mutation also engendered a variety of medical consequences such as medical procedures and impacts on these women's relations with the medical staff. Particularly, living with the BRCA1 or BRCA2 genetic mutation entailed that these women underwent additional screening in order to detect breast and/or ovarian cancer at an early stage. This supplementary screening consequently led to increased relations with the medical staff, as well as potential medical procedures such as prophylactic surgeries or discovering an abnormality which had to be further examined.

Nevertheless, although women who have received a positive test result indicating that they possess a mutation of the BRCA1 or BRCA2 genes are aware of their test result, and consequently their approximate risk level of developing breast or ovarian cancer, according to the current study's findings, these women do not appear to foresee a conclusion to this uncertain process. Particularly, although their new everyday lives appear to become more consistent, with increased screening, medical procedures, and effects on themselves and others, it seems as though this new normality is nonetheless permeated with varying levels of uncertainty, as the fear of developing cancer, further

screening, and potential surgeries are always a looming possibility in these women's lives.

**Findings in Comparison to Previous Research.** Although previous studies have been conducted to examine the experiences of women who possess mutations of the BRCA1 or BRCA2 genes, there is a lack of studies examining the experiences of women who have been living with the news of this result for six months or more (Beran et al., 2008; Croyle et al., 1997; Di Prospero et al., 2001; Dorval et al., 2000; Hamilton et al., 2009; Lerman et al., 1995; Lim et al., 2004; Schwartz et al., 2002; van Oostrom et al., 2003). Furthermore, there is a lack of qualitative studies examining the experiences of these women. Hence, comparison to other studies may pose certain challenges.

For instance, while researchers Di Prospero et al. (2001) conducted a study in which women possessing BRCA1 or BRCA2 genetic mutations reported wishing for additional support, similarly to the women in this study, the time at which participants were interviewed post-testing was not clearly delineated. Hence, it is difficult to determine whether these results truly support the experiences reported by women in the current study.

On the other hand, researchers van Oostrom et al. (2003) conducted a longitudinal study with women who possessed a BRCA1 or BRCA2 genetic mutation, and determined that women who had received their test result and had maintained family relationships tended to experience less distress than those who had experienced changes. These results could be applicable to the women in the current study in that some also described having experienced familial discord with family members since living with their test result.

However, distress level was not measured in the current study, hence comparisons should be made with caution.

Additional longitudinal studies, which were conducted by Schwartz et al. (2002), Hamilton et al. (2009), Beran et al. (2008), as well as Lim et al. (2004) describe conflicting results. Particularly although certain researchers found that women possessing the BRCA1 or BRCA2 gene mutation did not experience additional distress following testing (Hamilton et al., 2009; Schwartz et al., 2002), others described that these women experienced negative psychological effects, but that they experienced remittance of distressing symptoms over time (Beran et al., 2008; Lim et al., 2004). Nevertheless, distress level was not measured in the current study; hence comparisons to these studies should be made with caution.

Although very few studies have been conducted on the experiences of young women who possess mutations of the BRCA1 and BRCA2 genes, similar findings to those uncovered in the current study have been found. Specifically, researcher Allison Werner-Lin (2007, 2008) conducted two qualitative studies on the experiences of young women (aged 22-36) who possessed mutations of the BRCA1 or BRCA2 genes. Through findings it was revealed that, similarly to the current study, young women who receive and are living with a positive test result for the BRCA1 or BRCA2 gene mutation often feel as though they must engage in childbearing as soon as possible, prior to having any prophylactic surgeries.

This study's findings are similar to those in the current study, as some women from the current study expressed a desire to conceive following the receipt of their

positive test result. In particular, this desire to conceive entailed that the woman could have her child prior to developing cancer or engaging in any prophylactic surgeries.

Despite the similar age-range and desires of the women from the Werner-Lin (2007, 2008) studies and those of the women in the current study, certain significant differences also exist between the two samples. For instance, unlike in the current study, the time since receipt of test results was not made evident in one of the Werner-Lin (2007) studies. Furthermore, not all participants were married or in a committed relationship in the Werner-Lin (2007, 2008) studies, potentially affecting the desires and responding of participants.

Unlike in the current study, very few of the previously reviewed studies found that participants felt as though their lives were permeated with uncertainty, now that they were aware of their BRCA genetic mutation (d'Agincourt-Canning, 2006; Dagan & Goldblatt, 2009). Despite similarities in results with the current study, d'Agincourt-Canning (2006) did not examine the experiences of participants six months or more following the receipt of their test results, which makes comparisons to the current study's findings more difficult. On the other hand, the participants in the Dagan and Goldblatt (2009) study possessed Ashkenazi mutations in the BRCA1 and/or 2 genes. Specifically, as Jewish Ashkenazi women have a greater susceptibility to possessing this mutation, this may have affected their perception of receiving a positive test result, and may make results of this study less transferrable to individuals who do not possess Ashkenazi mutations in the BRCA1 and/or 2 genes (Caldes et al., 2002).

Interestingly, it has been found that one of the motivational factors of women who undergo testing for the BRCA1 or BRCA2 genetic mutation is to reduce the uncertainty

associated with their breast cancer risk level. Furthermore, patients being questioned regarding the uncertainty surrounding their breast cancer risk level as well as their ability to cope with a positive test result for a BRCA1/2 gene mutation, have been found to describe reduced feelings of uncertainty following genetic testing (Baty, Dudley, Musters, & Kinney, 2006). Nevertheless, these findings may not be comparable to those of the current study, as these constructs were not specifically examined through the current study's qualitative analysis. Furthermore, Baty et al. (2006) did not examine uncertainty in participants 6 months or more following the receipt of their test result. In other words, uncertainty levels may have been affected over time; hence these results may not be comparable to the current study's findings.

Further research indicates that there is often a great deal of uncertainty associated with the decision-making of women who are choosing whether to undergo Prophylactic Mastectomies or oophorectomies, which is a course of action often offered to women who have been identified as carriers of the BRCA1 or BRCA2 genetic mutations (Klitzman & Chung, 2010). However, the uncertainty associated with prophylactic surgery and BRCA1/2 gene mutation testing needs further examination. More specifically, the study conducted by Klitzman and Chung (2010) did not specify between patients who were cancer sufferers, cancer survivors, BRCA1/2 mutation carriers, or those who had received inconclusive or negative results, making it difficult to determine whether any of these factors influenced responding of participants, and consequently, obtained results.

Uncertainty in patients suffering from cancer has been more thoroughly researched than uncertainty in women who are living with BRCA1 or BRCA2 genetic

mutations. For instance, researchers Shaha, Cox, Talman, and Kelly (2008) performed a meta-analysis of 40 studies pertaining to uncertainty in breast, prostate, and colorectal cancer. In general, they uncovered that uncertainty can be reinforced by three different sources, in cancer patients. Specifically, the authors stated that these sources include “information related issues after cancer has been diagnosed; decisions about treatment and the effect of cancer-related uncertainty, over the disease trajectory, on the lives of patients and their families” (Shaha et al., 2008, pp. 65). However, the researchers mentioned that it is unclear which of these sources is more important or influential.

These findings could be applied to the present study in that the women in the current study felt uncertainty surrounding the medical consequences of living with their genetic test result. In addition, the women in the current study also described uncertainty pertaining to their families and children, with respects to possessing this genetic mutation.

In addition to research on uncertainty in relation to cancer, uncertainty in illness in general has also been more thoroughly examined, and its effects have also been conceptualized. In particular, in a frequently cited article, Mishel (1988) devised of a model of perceived uncertainty in illness. In this article, he describes uncertainty in illness as:

The inability to determine the meaning of illness-related events. It is the cognitive state created when the person cannot adequately structure or categorize an event because of the lack of sufficient cues. Uncertainty occurs in a situation in which the decision maker is unable to assign definite value to objects or events and/or is unable to predict outcomes accurately (Mishel, 1988, p. 225).

This definition, which was intended to describe uncertainty in illness, appears to also apply to women who are living with a positive test result indicating they possess mutations of the BRCA1 or BRCA2 genes. Specifically, like the ill person who is unable to properly predict the outcome of their illness, these women also seem to feel as though they cannot predict the outcome of their life now that they are aware of and are living with a BRCA1 or BRCA2 genetic mutation. According to Mishel (1988), this inability to predict the outcome of one's illness, or in the current study, potential illness, propels the patient into a state of uncertainty.

Mishel (1988) explains that if a patient does not create a cognitive schema, or their own interpretation for their illness, they may consequently experience uncertainty. The cognitive schema is composed of three specific components: symptom pattern, event familiarity, and event congruence. These components can in turn be influenced by two variables: cognitive capacity and structure providers. Mishel (1988) describes cognitive capacity as the information-processing capabilities of an individual. On the other hand, he states that an individual's structure providers consist of the resources which are available to a person such as social support, education level, and credible authority.

Mishel (1988) explains that uncertainty results from an inability to form a cognitive schema for the illness. Nevertheless, he describes that an individual's appraisal of uncertainty can either be one of danger or as an opportunity, and that coping skills can assist individuals in adapting to uncertainty in illness.

Since women who are aware of and living with a BRCA1 or BRCA2 genetic mutation do not present symptoms of possessing this genetic mutation, have not necessarily encountered a similar experience, and appear unsure what to expect, they tend

to experience uncertainty. According to Mishel (1988), this would entail that these women have trouble creating a cognitive schema of their situation, which leads them to perceive it as uncertain and unable to be properly defined.

This particular finding is quite significant in that it suggests that women possessing mutations of the BRCA1 or BRCA2 genes must learn to live with uncertainty for an undeterminable amount of time. More specifically, since no ending can be foreseen to possessing the genetic mutation, the only options available to these women consist of prevention through prophylactic surgeries, medication, and screening. Nevertheless, these options do not preclude these women from developing cancer (Di Prospero et al., 2001).

Furthermore, this is a particularly blunt reality for the women in the current study, as most had decided not to undergo any prophylactic surgeries, or participate in any medical trials. Hence, their lives will continuously be permeated with the uncertainty surrounding their potential to develop breast and/or ovarian cancers due to their BRCA1 or BRCA2 genetic mutation.

In summary, while certain studies have examined the experiences of women with BRCA1 or BRCA2 genetic mutations, very few have investigated these experiences months following the receipt of the test result (Beran et al., 2008; Hamilton et al., 2009; Lim et al., 2004; Schwartz et al., 2002; van Oostrom et al., 2003). Furthermore, few qualitative studies have been undertaken to examine the experiences of these women. Finally, studies which have uncovered that these women must learn to live in an uncertain new world, in which no satisfying ending is perceived to be in reach, appear to be virtually nonexistent (Baty et al., 2006; Klitzman & Chung, 2010).

### **Implications for the Medical Team**

The current research has many implications for the medical team who provides test results, cares for, and provides support to women possessing the BRCA1 or BRCA2 genetic mutation. Although it would be imprudent to suggest that all women who have received this positive test result experience this occurrence in the same manner as the women in the current study, the obtained findings could nevertheless be helpful in implementing certain changes in the care and support provided to these women.

More specifically, the findings obtained through the current study implied that women receiving a positive test result, indicating that they possess a mutation of the BRCA1 or BRCA2 genes, experience changes in their self-perception as well as their perception of others. It appears that these women's self-perception is affected cognitively, emotionally, and behaviourally.

Hence, through these findings, it could be suggested that the medical team be more attentive and allot more time to their patients, following the receipt of this news. For instance, studies in which physicians allocate additional time for cancer patients have found that this course of action tends to alleviate patients' anxiety levels (Takayama, Yamazaki, & Katsumata, 2001). Based upon the current study's results, it follows that a similar course of action could potentially help alleviate some of the distress experienced by these women following the receipt of their test result.

On the other hand, it would also be important to address the potential stigma associated with receiving the positive test result. Particularly, the participants in the current study described feeling different aspects of stigma, such as feeling judged, misunderstood, rejected, avoided, and so on, following the receipt of their test result.

Although no other study to date has examined this topic amongst these women, it would nevertheless be important for genetic counsellors to mention this topic as a potential consequence of receiving a positive test result. In addition, it would be significant for physicians and nurses to follow-up and address the stigma associated with possessing this genetic mutation if participants felt similarly to the participants of the current study.

Furthermore, the finding of feeling less like a woman would also be important to manage through additional discussions and support provided by physicians and nurses at the clinic. Particularly, it would be essential to determine whether these feelings are widespread amongst patients, and if so, how to help them cope with this significant emotion.

Although very little research has been conducted on women who have lived with a test result indicating that they possess a BRCA1 or BRCA2 genetic mutation for over six months, it would nevertheless also be important for physicians and nurses to consider that the impacts of the test result continue even years after the receipt of results, particularly the uncertainty associated with living with this test result. Therefore, discussing the impacts of living with this genetic mutation, specifically the impacts this has on the patient and their families, would be essential in further assisting these women cope with this experience. Furthermore, the medical staff should also consider the implications of living with the perpetual uncertainty of possessing this genetic mutation.

### **Implications for the Field of Counselling**

Counsellors could generally be very important in the process of helping BRCA gene mutation carriers receive their test results, and continue to live with its consequences and impacts.

In particular, many participants in the current study, as well as in previous studies, mentioned that they would appreciate additional support through the means of support groups for women possessing the BRCA1 and BRCA2 genetic mutations (Di Prospero et al., 2001). Furthermore, some women in the current study also described that they would have appreciated additional support during the meeting with the genetic counsellor who provided them with their test results.

Counsellors could be involved in this process by meeting with patients who are receiving their test results, as well as providing continuing support as long as is deemed necessary by the patient and their physician. Furthermore, counsellors could also organize and facilitate group discussions run through the clinic that patients attend, in order to help patients better adjust and feel less isolation.

### **Limitations**

Despite this study's revealing findings, they must be considered in light of certain limitations. Firstly, although grounded theory methodology was utilized to identify themes and analyze the data obtained through this study, due to the reduced number of participants, saturation of data was not achieved. Therefore, the compilation and analysis of data was halted at the step of conceptual ordering, the precursor to devising a theory. Consequently, the interpretation of the data obtained through this study did not constitute a grounded theory.

Secondly, the goal of this research was to better comprehend the experiences of women who have been identified as possessing a mutation of the BRCA1 or BRCA2 genes. As this is a very specific medical condition, which was recently discovered, and which has potentially differing outcomes for each individual, findings may not be

transferrable to other individuals who have been identified as possessing differing gene mutations, other women at high risk for breast cancer, or patients with cancer (Di Prospero et al., 2001).

Thirdly, as the sample size was relatively small, and consisted of a very specific subset of the BRCA gene mutation carriers who attended the high risk clinic where they received their services (i.e. age range and no PM), it is possible that the findings obtained from this study do not necessarily represent the experiences of other BRCA gene mutation carriers who have chosen a different course of action.

Finally, even though certain significant findings emerged through the analysis of the experiences of the current study's participants (i.e. feeling less like a woman, and stigma), further research is needed in order to better comprehend these phenomena.

### **Implications for Future Research**

This study provided a glimpse of the experiences of women possessing a mutation for the BRCA1 or BRCA2 genetic mutation. Future research will be essential in order to better understand the experiences of these women, particularly after six months following the receipt of their test results. Furthermore, there is a need for additional qualitative research to be conducted on the topic of BRCA gene mutation carriers' experiences.

The current study's finding regarding the stigma associated with receiving a positive test result indicating one possesses a mutation of the BRCA1 or BRCA2 genes should be further investigated. A challenge that may be faced in future studies is the manner in which to examine stigma. Specifically, most of the women in the current study stated that they did not feel stigmatized even when provided a definition of stigma, yet their experiences clearly delineated the definition of stigma provided. Hence, examining

this construct amongst these women should be approached carefully, as it may pose difficulties in future studies as well.

In addition, the current study should be replicated utilizing a larger, and more diverse sample, in order to better represent the experiences of women from more diverse backgrounds. Furthermore, since the current study did not achieve saturation, and hence the development of a grounded theory, it would be of interest to continue or add to the current research results so as to attain this step.

It would also be of interest to further examine the finding of the uncertainty associated with living with the BRCA genetic mutation, so as to better comprehend the consequences this genetic test result has on recipients over time. More specifically, future research on the uncertainty associated with receiving this test result could examine the uncertainty experienced by these women longitudinally, as the persistence or experience of uncertainty over a long period of time has not been previously examined.

The feelings of uncertainty and stigma should also be examined amongst women who possess mutations of the BRCA1 and/or BRCA2 genes, and who have previously had cancer, as well as in women who have not had cancer, but who have opted to undergo prophylactic surgeries, such as a prophylactic mastectomy. Since the majority of the women in the current study had chosen not to undergo any prophylactic surgeries, and none had been previously diagnosed with cancer, it would be of interest to investigate whether feelings of uncertainty and stigma are also present amongst women who have had differing experiences, yet still possess the BRCA genetic mutation.

Finally, it would be of great interest to examine the finding of feeling less like a woman, amongst other women who have also received a positive test result indicating

they possess a BRCA genetic mutation. Furthermore, it would be of interest to examine this finding in different age groups, for instance in women aged 20-35, and from 36-45, in order to further understand whether this feeling changes with age. The implications of this finding would be essential to manage through counselling and medical care.

### **Conclusion**

Receiving a positive test result indicating one possesses a mutation of the BRCA1 or BRCA2 genes appears to be a significant experience in a woman's life. The current study examined the experiences of women who possessed mutations of the BRCA1 or BRCA2 genes, and their lives some time following the receipt of their test result. The findings described may allow researchers, family, friends, and medical staff to better comprehend the phenomena experienced by these women following the receipt of this test result, as well as their lives years later. It is hoped that this research will lead to effective interventions for these women, particularly in the domain of support and care, so that they may benefit from this experience both emotionally and psychologically.

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Appendix A

**Demographic Questionnaire**

- 1) Your current age: \_\_\_\_\_
- 2) Your age when you were identified as possessing a mutation of the BRCA1 or BRCA2 genes: \_\_\_\_\_
- 3) Your mother tongue: \_\_\_\_\_

4) Marital status: (Please indicate with a checkmark)

Married \_\_\_\_\_ Separated \_\_\_\_\_ Divorced \_\_\_\_\_  
Widowed \_\_\_\_\_ Single \_\_\_\_\_

- 5) Children: Do you have any children? Yes \_\_\_\_\_ No \_\_\_\_\_  
Expecting? Yes \_\_\_\_\_ No \_\_\_\_\_

If you have any children, please indicate (his/her):

Age \_\_\_\_\_ Sex \_\_\_\_\_  
Age \_\_\_\_\_ Sex \_\_\_\_\_  
Age \_\_\_\_\_ Sex \_\_\_\_\_  
Age \_\_\_\_\_ Sex \_\_\_\_\_  
Age \_\_\_\_\_ Sex \_\_\_\_\_

- 6) Education: Your level of education.
  - a) Less than High School \_\_\_\_\_
  - b) High School \_\_\_\_\_

- c) Community college \_\_\_\_\_
- d) University (Undergraduate degree) \_\_\_\_\_
- e) University (Graduate degree) \_\_\_\_\_
- f) Other \_\_\_\_\_

7) Employment:

- a) Present occupation \_\_\_\_\_ (Since \_\_\_\_\_)
- b) Previous occupation \_\_\_\_\_ (Number of years \_\_\_\_\_)
- c) Retired? Yes \_\_\_\_\_ (Number of years \_\_\_\_\_)  
No \_\_\_\_\_

8) Does your family live in:

- a) A rural setting \_\_\_\_\_
- b) An urban setting \_\_\_\_\_

9) Are you currently enrolled in a clinical trial of a new drug treatment for breast cancer prevention? Yes \_\_\_\_\_ No \_\_\_\_\_

10) Have you been receiving any other specialized care since you initially received your positive test result? Yes \_\_\_\_\_ No \_\_\_\_\_

11) How many times per year do you come to the clinic? \_\_\_\_\_

12) Previous breast-cancer related family history:

Who in you family has previously been diagnosed with breast cancer?

Family Member	Diagnosed with Breast Cancer? (Yes/No)	Number of Women affected:	Living (Yes/No)	Living with no recurrence? (Yes/No)	Deceased (Yes/No)	Deceased from cancer? (Yes/No)
Mother						
Sister						
Maternal Aunt						
Paternal Aunt						
Maternal Grandmother						
Paternal Grandmother						
Great Aunt						

13) Have any men in your family ever been diagnosed with breast cancer? If so, please indicate how many \_\_\_\_\_

14) Has anyone else in your family received positive test results for possessing the BRCA1 or BRCA2 mutation?

Yes \_\_\_\_\_ No \_\_\_\_\_

If yes, please indicate your relation to this(these)

individual(s): \_\_\_\_\_

15) Please indicate with a checkmark who you consider to be part of your support structure:

a) Spouse/partner \_\_\_\_\_

b) Children \_\_\_\_\_

- c) Family \_\_\_\_\_
- d) Friends \_\_\_\_\_
- e) Work Colleagues \_\_\_\_\_
- f) Neighbours \_\_\_\_\_
- g) Family Doctor \_\_\_\_\_
- h) Specialists/physicians \_\_\_\_\_

### Questionnaire démographique

- 1) Votre âge: \_\_\_\_\_
- 2) Votre âge quand vous avez reçu le résultat positif indiquant que vous aviez une mutation des gènes BRCA1 ou BRCA2: \_\_\_\_\_
- 3) Votre langue maternelle: \_\_\_\_\_

- 4) État civil : (S'il vous plaît indiquer avec un crochet)

Mariée \_\_\_\_\_ Séparée \_\_\_\_\_ Divorcée \_\_\_\_\_

Veuve \_\_\_\_\_ Célibataire \_\_\_\_\_

- 5) Enfants: Avez-vous des enfants? Oui \_\_\_\_\_ Non \_\_\_\_\_

Êtes-vous enceinte? Oui \_\_\_\_\_ Non \_\_\_\_\_

Si vous avez des enfants, s'il vous plaît indiquer (sa/son):

Âge \_\_\_\_\_ Sexe \_\_\_\_\_

Âge \_\_\_\_\_ Sexe \_\_\_\_\_

Âge \_\_\_\_\_ Sexe \_\_\_\_\_

Âge \_\_\_\_\_ Sexe \_\_\_\_\_

Âge \_\_\_\_\_ Sexe \_\_\_\_\_

- 6) Scolarité: S'il vous plaît indiquer avec un crochet quel est le dernier diplôme que vous avez reçu.

a) Moins que l'école secondaire \_\_\_\_\_

b) École secondaire \_\_\_\_\_

- c) Collège \_\_\_\_\_
- d) Université (1er cycle) \_\_\_\_\_
- e) Université (2e et/ou 3e cycle) \_\_\_\_\_
- f) Autre \_\_\_\_\_

7) Emplois:

- a) Occupation actuelle: \_\_\_\_\_ (Depuis \_\_\_\_\_)
- b) Occupation antérieure: \_\_\_\_\_ (Nombre d'années \_\_\_\_\_)
- c) Retraitée? Oui \_\_\_\_\_ (Nombre d'années \_\_\_\_\_)  
Non \_\_\_\_\_

8) Est-ce que votre famille vit:

- c) En région \_\_\_\_\_
- d) Dans un centre urbain \_\_\_\_\_
- e) \_\_\_\_\_

9) Participez-vous actuellement dans un traitement expérimental pour la prévention

du cancer du sein? Oui \_\_\_\_\_ Non \_\_\_\_\_

10) Avez-vous reçu des soins spécialisés supplémentaires depuis que vous avez reçu

vos résultats positifs? Oui \_\_\_\_\_ Non \_\_\_\_\_

11) Combien de fois par année venez-vous à cette clinique? \_\_\_\_\_

## 12) Antécédents familiaux de cancer du sein dans votre famille:

Qui dans votre famille a déjà été diagnostiqué avec le cancer du sein dans votre famille?

Membre de la famille	Diagnostiquée avec un cancer du sein? (Oui/Non)	Nombre de femmes affectées:	Vivantes? (Oui/Non)	Vivantes sans cancer? (Oui/Non)	Décédées? (Oui/Non)	Décédées du cancer? (Oui/Non)
Mère						
Sœur						
Tante maternelle						
Tante paternelle						
Grand-mère maternelle						
Grand-mère paternelle						
Grandes tantes						

13) Es-ce qu'il y a des hommes qui ont déjà été diagnostiqués avec un cancer du sein dans votre famille? Si oui, s'il vous plaît indiquer combien \_\_\_\_\_

14) Es-ce qu'il y a quelqu'un d'autre dans votre famille qui a déjà reçu un résultat positif pour la mutation des gènes BRCA1 ou BRCA2?

Oui \_\_\_\_\_ Non \_\_\_\_\_

Si oui, s'il vous plaît indiquer votre relation : \_\_\_\_\_

15) S'il vous plaît indiquer qui vous considérez faire partie de votre réseau de support:

a) Mari/conjoint(e) \_\_\_\_\_

- b) Enfants \_\_\_\_\_
- c) Famille \_\_\_\_\_
- d) Amis \_\_\_\_\_
- e) Collègues de travail \_\_\_\_\_
- f) Voisins \_\_\_\_\_
- g) Médecin de famille \_\_\_\_\_
- h) Spécialistes (médecins) \_\_\_\_\_

Appendix B

Dear (patient name),

I am writing to inform you of a research study being conducted at the Ottawa Hospital's Women's Breast Health Center (WBHC). This study, entitled *The Lived Experience of Women Possessing a Genetic Predisposition to Developing Breast Cancer: A Grounded Theory Study*, is being conducted with women who have been identified as carriers of BRCA1 or BRCA2 mutations. You are being contacted as a woman who is eligible to participate in this study. You are not obligated to participate. Also, whether or not you choose to participate will not affect the care you usually receive at the WBHC.

If you might be interested, I invite you to read more about the study on the next page where you will find a short summary and a form that you can complete if you decide that you would like to be contacted by the researcher to hear more about the study.

Thank you very much for your time and consideration,

Dr. Audley Bodurtha  
MD, FRCPC  
Surgeon  
Medical Director, Women's Breast Health Centre

Dear (patient name),

I am writing to inform you of a research study being conducted at the Ottawa Hospital's Women's Breast Health Center (WBHC). This study, entitled *The Lived Experience of Women Possessing a Genetic Predisposition to Developing Breast Cancer: A Grounded Theory Study*, is being conducted with women who have been identified as carriers of BRCA1 or BRCA2 mutations. You are being contacted as a woman who is eligible to participate in this study. You are not obligated to participate. Also, whether or not you choose to participate will not affect the care you usually receive at the WBHC.

If you might be interested, I invite you to read more about the study on the next page where you will find a short summary and a form that you can complete if you decide that you would like to be contacted by the researcher to hear more about the study.

Thank you very much for your time and consideration,

Dr. Shailendra Verma  
MD, FRCPC  
Medical Oncologist

## Permission to Contact Patients Form

**Title:** The Lived Experience of Women Possessing a Genetic Predisposition to Developing Breast Cancer: A Grounded Theory Study

**Investigators:**

Julia DiMillo

(Principal Investigator)

Dr. Eva Tomiak, MD

(Responsible Principal Investigator)

Dr. André Samson, PhD.

(Co-Investigator)

**Description:**

We are asking for your permission to contact you to hear more about a research study being conducted by Julia DiMillo for her master's degree in Educational Counselling, at the University of Ottawa. The primary researcher is being supervised by Dr. André Samson (PhD), a professor at the University of Ottawa in the Faculty of Education.

The purpose of this study is to better understand the experiences of women who have tested positive for mutations of the BRCA1 or BRCA2 genes. More specifically, the researcher would like to examine whether these women experience stigma, and the effect this might have on their day-to-day life.

Women who have mutations of the BRCA1 or BRCA2 genes and who are between the ages of 20 and 35 years are eligible to participate.

As a participant in this study, you will be asked to participate in one interview lasting about 90 minutes and to complete a short demographic questionnaire.

Your decision to participate in this study is completely voluntary, and the decision not to participate in the study will not have any impact on the usual care you receive at the clinic.

We are currently only asking for your permission to be contacted to hear more about the study and ask that you return this form completed and signed if you are interested. If you indicate that you are interested to hear more about the study, a researcher will contact you by telephone and will describe the research in more detail. You will then have the chance to decide whether or not you wish to participate.

This research project has been approved by the Ottawa Hospital Research Ethics Board (OHREB). If you have any questions about your rights as a research participant, this research, you may contact the Chairman of the Ottawa Hospital Research Ethics Board at

If you have any questions about this study, please contact:  
Julia DiMillo (Principal Investigator)

Thank you for your time and consideration.

## Permission to Contact Patients Form

**Title:** The Lived Experience of Women Possessing a Genetic Predisposition to Developing Breast Cancer: A Grounded Theory Study

**Investigators:**

Julia DiMillo  
(Principal Investigator)

Dr. Eva Tomiak, MD  
(Responsible Principal Investigator)

Dr. André Samson, PhD.  
(Co-Investigator)



I would like to be contacted to hear more about this study.

If you indicated that you would like to hear more information about this study, please complete the following information. PLEASE PRINT CLEARLY

Name: \_\_\_\_\_

Telephone Number:

Day: (\_\_\_\_) \_\_\_\_\_

Evening: (\_\_\_\_) \_\_\_\_\_

\_\_\_\_\_  
Signature

\_\_\_\_\_  
Name (Please print)

\_\_\_\_\_  
Date

\*\*\*Please return this form in the included envelope when complete.

Thank you for your time and consideration.

Chère (nom de la patiente),

Je vous écris pour vous informer d'une étude prenant place au Centre de santé du sein de la femme de l'Hôpital d'Ottawa. Cette étude, intitulée *L'expérience des femmes qui possèdent une mutation des gènes BRCA1 ou BRCA2 : Une recherche utilisant une théorie ancrée*, est entrain d'être menée avec des femmes qui ont reçu un résultat indiquant qu'elles possèdent une mutation des gènes BRCA1 ou BRCA2. Je vous contacte car vous êtes une femme éligible à participer dans cette recherche. Cependant, vous n'êtes pas obligée de participer, et les soins usuels que vous recevez à la clinique ne seront pas affectés, n'importe votre décision de participer.

Si vous pensez que vous pourriez être intéressée, je vous invite à lire un peu plus au sujet de l'étude sur la prochaine page. Vous y trouverez un court résumé de l'étude ainsi qu'un formulaire que vous pourrez compléter si vous décidez que vous aimeriez être contacté par la chercheure principale pour apprendre un peu plus au sujet de l'étude.

Merci beaucoup pour votre intérêt et votre considération,

Dr. Audley Bodurtha  
MD, FRCPC  
Chirurgien  
Directeur Médical, Centre de santé du sein de la femme

Chère (nom de la patiente),

Je vous écris pour vous informer d'une étude prenant place au Centre de santé du sein de la femme de l'Hôpital d'Ottawa. Cette étude, intitulée *L'expérience des femmes qui possèdent une mutation des gènes BRCA1 ou BRCA2 : Une recherche utilisant une théorie ancrée*, est entrain d'être menée avec des femmes qui ont reçu un résultat indiquant qu'elles possèdent une mutation des gènes BRCA1 ou BRCA2. Je vous contacte car vous êtes une femme éligible à participer dans cette recherche. Cependant, vous n'êtes pas obligée de participer, et les soins usuels que vous recevez à la clinique ne seront pas affectés, n'importe votre décision de participer.

Si vous pensez que vous pourriez être intéressée, je vous invite à lire un peu plus au sujet de l'étude sur la prochaine page. Vous y trouverez un court résumé de l'étude ainsi qu'un formulaire que vous pourrez compléter si vous décidez que vous aimeriez être contacté par la chercheure principale pour apprendre un peu plus au sujet de l'étude.

Merci beaucoup pour votre intérêt et votre considération,

Dr. Shailendra Verma  
MD, FRCPC  
Oncologue Médical

## Formulaire de demande pour contacter des patients(es)

**Titre :** L'expérience des femmes qui possèdent une mutation des gènes BRCA1 ou BRCA2 : Une recherche utilisant une théorie ancrée.

**Investigateurs:**

Julia DiMillo (Chercheuse Principale)	Dr. Eva Tomiak, MD (Co-Investigatrice)	Dr. André Samson, PhD. (Co-Investigateur)
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**Description:**

Nous vous abordons pour demander votre permission pour vous contacter pour apprendre un peu plus au sujet d'une étude étant conduite par Julia DiMillo pour satisfaire les exigences de sa maîtrise en counselling éducationnel. La chercheuse sera supervisée par le Dr. André Samson, professeur de l'Université d'Ottawa, faculté d'éducation.

L'objectif de cette recherche est de mieux comprendre les expériences vécues par les femmes qui ont reçu un résultat positif indiquant qu'elles ont une mutation des gènes BRCA1 ou BRCA2. Plus particulièrement, la chercheuse aimerait mieux comprendre l'expérience de ces femmes face au stigma, et l'impact que cela pourrait avoir dans leur quotidien.

Des femmes possédant une mutation des gènes BRCA1 ou BRCA2 et qui sont âgées entre 20 et 35 ans, sont éligibles à participer dans cette étude.

Comme participante dans cette étude, vous serez invitée à participer dans un entretien qui durera environ 90 minutes et de compléter un court questionnaire démographique.

Votre décision de faire partie de cette recherche est complètement libre et volontaire. Si vous décidez de ne pas participer à cette recherche, cette décision n'aura aucun impact sur les soins que vous recevez régulièrement à la clinique.

En ce moment, nous vous demandons seulement pour votre permission de vous contacter pour en apprendre plus au sujet de l'étude. Nous vous prions de retourner ce formulaire signé si vous êtes intéressée. Si vous indiquez que vous aimeriez apprendre d'avantage au sujet de l'étude, une chercheuse vous contactera par téléphone et décrira l'étude en plus de détails. Vous aurez ensuite la chance de décider si vous aimeriez participer.

Ce projet a été approuvé par le Conseil d'éthique en recherche de l'Hôpital d'Ottawa. Si vous avez des questions concernant vos droits comme participante ou cette recherche, vous pouvez contacter le Conseil d'éthique en recherche de l'Hôpital d'Ottawa

Si vous avez des questions concernant ce projet, contactez s'il vous plaît :

Julia DiMillo  
Chercheuse principale

Merci beaucoup pour votre intérêt et votre considération.

## Formulaire de demande pour contacter des patients(es)

**Titre :** L'expérience des femmes qui possèdent une mutation des gènes BRCA1 ou BRCA2 : Une recherche utilisant une théorie ancrée.

**Investigateurs:**

Julia DiMillo  
(Chercheure Principale)

Dr. Eva Tomiak, MD  
(Co-Investigatrice)

Dr. André Samson, PhD.  
(Co-Investigateur)

Oui, j'aimerais apprendre d'avantage au sujet de l'étude.



Si vous avez indiqué que vous aimeriez apprendre d'avantage au sujet de l'étude, veuillez s'il vous plaît compléter l'information suivante. **VEUILLEZ S'IL VOUS PLAÎT ÉCRIRE EN LETTRE MOULÉES.**

Nom : \_\_\_\_\_

Numéro de téléphone:

Jour: (\_\_\_\_\_) \_\_\_\_\_

Soirée: (\_\_\_\_\_) \_\_\_\_\_

\_\_\_\_\_  
Signature

\_\_\_\_\_  
Nom (Lettres moulées)

\_\_\_\_\_  
Date

\*\*\*Veuillez s'il vous plaît envoyer ce formulaire dans l'enveloppe fournie lorsqu'il a été complété.

Merci beaucoup pour votre intérêt et votre considération.

Appendix C

Dear (patient name),

Approximately 2 months ago, you received an invitation to be contacted to hear more about a study entitled *The Lived Experience of Women Possessing a Genetic Predisposition to Developing Breast Cancer: A Grounded Theory Study*. This study is being conducted by Julia DiMillo as a prerequisite for her master's degree in Educational Counselling, at the University of Ottawa.

You were contacted as a woman who was eligible to participate in this study. We are sending this letter as a reminder for the study, and are once more inviting you to hear more about the study. You are not obligated to participate. Also, whether or not you choose to participate will not affect the care you usually receive at the WBHC.

If you might be interested, I invite you to read more about the study on the next page where you will find a short summary and a form that you can complete if you decide that you would like to be contacted by the researcher to hear more about the study.

Thank you very much for your time and consideration,

Dr. Audley Bodurtha  
MD, FRCPC  
Surgeon  
Medical Director, Women's Breast Health Centre

Dear (patient name),

Approximately 2 months ago, you received an invitation to be contacted to hear more about a study entitled *The Lived Experience of Women Possessing a Genetic Predisposition to Developing Breast Cancer: A Grounded Theory Study*. This study is being conducted by Julia DiMillo as a prerequisite for her master's degree in Educational Counselling, at the University of Ottawa.

You were contacted as a woman who was eligible to participate in this study. We are sending this letter as a reminder for the study, and are once more inviting you to hear more about the study. You are not obligated to participate. Also, whether or not you choose to participate will not affect the care you usually receive at the WBHC.

If you might be interested, I invite you to read more about the study on the next page where you will find a short summary and a form that you can complete if you decide that you would like to be contacted by the researcher to hear more about the study.

Thank you very much for your time and consideration,

Dr. Shailendra Verma  
MD, FRCPC  
Medical Oncologist

## Permission to Contact Patients Form

**Title:** The Lived Experience of Women Possessing a Genetic Predisposition to Developing Breast Cancer: A Grounded Theory Study

**Investigators:**

Julia DiMillo

(Principal Investigator)

Dr. Eva Tomiak, MD

(Responsible Principal Investigator)

Dr. André Samson, PhD.

(Co-Investigator)

**Description:**

We are asking for your permission to contact you to hear more about a research study being conducted by Julia DiMillo for her master's degree in Educational Counselling, at the University of Ottawa. The primary researcher is being supervised by Dr. André Samson (PhD), a professor at the University of Ottawa in the Faculty of Education.

The purpose of this study is to better understand the experiences of women who have tested positive for mutations of the BRCA1 or BRCA2 genes. More specifically, the researcher would like to examine whether these women experience stigma, and the effect this might have on their day-to-day life.

Women who have mutations of the BRCA1 or BRCA2 genes and who are between the ages of 20 and 40 years are eligible to participate.

As a participant in this study, you will be asked to participate in one interview lasting about 90 minutes and to complete a short demographic questionnaire.

Your decision to participate in this study is completely voluntary, and the decision not to participate in the study will not have any impact on the usual care you receive at the clinic.

We are currently only asking for your permission to be contacted to hear more about the study and ask that you return this form completed and signed if you are interested. If you indicate that you are interested to hear more about the study, a researcher will contact you by telephone and will describe the research in more detail. You will then have the chance to decide whether or not you wish to participate.

This research project has been approved by the Ottawa Hospital Research Ethics Board (OHREB). If you have any questions about your rights as a research participant, this research, you may contact the Chairman of the Ottawa Hospital Research Ethics Board

If you have any questions about this study, please contact:  
Julia DiMillo (Principal Investigator)

Thank you for your time and consideration.

Version date: July 20<sup>th</sup>, 2009

Chère (nom de la patiente),

Deux mois passés, vous avez reçu une invitation à apprendre davantage au sujet d'une étude ayant lieu au Centre de santé du sein de la femme de L'Hôpital d'Ottawa. Cette étude, intitulée *L'expérience des femmes qui présentent une mutation génétique les prédisposant au cancer du sein : Étude faisant appel à une théorie à base empirique*, est menée sur des femmes ayant reçu un diagnostic indiquant qu'elles présentent une mutation des gènes BRCA1 ou BRCA2. L'étude est menée par Julia DiMillo, une étudiante à la maîtrise en counselling éducationnel à l'université d'Ottawa.

Nous avons communiqué avec vous parce que vous étiez une femme admissible à participer à cette recherche. Nous vous envoyons ce rappel pour vous réinviter à apprendre davantage au sujet de l'étude. Cependant, vous n'êtes pas obligée de participer et peu importe votre décision, celle-ci n'aura aucune conséquence sur la qualité des soins que vous recevez habituellement au Centre.

Si vous croyez que vous pourriez vouloir prendre part à cette étude, je vous invite à prendre connaissance de l'étude sur la page qui suit, où vous y trouverez un résumé de l'étude ainsi qu'un formulaire que vous pourrez remplir advenant que vous décidiez que vous aimeriez que la chercheuse principale communique avec vous pour vous transmettre de plus amples renseignements au sujet de l'étude.

Nous vous remercions pour votre temps et votre intérêt,

D<sup>r</sup> Audley Bodurtha  
MD, FRCPC  
Chirurgien  
Directeur médical, Centre de santé du sein de la femme

Chère (nom de la patiente),

Deux mois passés, vous avez reçu une invitation à apprendre davantage au sujet d'une étude ayant lieu au Centre de santé du sein de la femme de L'Hôpital d'Ottawa. Cette étude, intitulée *L'expérience des femmes qui présentent une mutation génétique les prédisposant au cancer du sein : Étude faisant appel à une théorie à base empirique*, est menée sur des femmes ayant reçu un diagnostic indiquant qu'elles présentent une mutation des gènes BRCA1 ou BRCA2. L'étude est menée par Julia DiMillo, une étudiante à la maîtrise en counselling éducationnel à l'université d'Ottawa.

Nous avons communiqué avec vous parce que vous étiez une femme admissible à participer à cette recherche. Nous vous envoyons ce rappel pour vous réinviter à apprendre davantage au sujet de l'étude. Cependant, vous n'êtes pas obligée de participer et peu importe votre décision, celle-ci n'aura aucune conséquence sur la qualité des soins que vous recevez habituellement au Centre.

Si vous croyez que vous pourriez vouloir prendre part à cette étude, je vous invite à prendre connaissance de l'étude sur la page qui suit, où vous y trouverez un résumé de l'étude ainsi qu'un formulaire que vous pourrez remplir advenant que vous décidiez que vous aimeriez que la chercheuse principale communique avec vous pour vous transmettre de plus amples renseignements au sujet de l'étude.

Nous vous remercions pour votre temps et votre intérêt,

D<sup>r</sup> Shailendra Verma  
MD, FRCPC  
Oncologue médical

## Formulaire de demande à des fins de communication avec des patientes

**Titre :** *L'expérience des femmes qui présentent une mutation génétique les prédisposant au cancer du sein : Étude faisant appel à une théorie à base empirique*

**Investigateurs :**

Julia DiMillo (Chercheuse principale)	D <sup>r</sup> Eva Tomiak, MD (Co-chercheuse)	D <sup>r</sup> André Samson, PhD. (Co-chercheur)
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**Description :**

Nous souhaitons obtenir votre permission afin de communiquer avec vous afin de vous transmettre de plus amples renseignements au sujet d'une étude effectuée par Julia DiMillo en vue de satisfaire les exigences de sa maîtrise en counselling éducationnel. La chercheuse sera sous la supervision du docteur André Samson, professeur à la Faculté d'éducation de l'Université d'Ottawa.

L'objectif de cette recherche est de mieux comprendre les expériences vécues par les femmes qui ont reçu un résultat positif indiquant qu'elles ont une mutation des gènes BRCA1 ou BRCA2. Plus particulièrement, la chercheuse aimerait mieux comprendre l'expérience de ces femmes face au stigma, et l'impact que cela pourrait avoir dans leur quotidien.

Les femmes présentant une mutation des gènes BRCA1 ou BRCA2 et qui sont âgées de 20 à 40 ans, seront admissibles à cette étude.

À titre de participante à cette étude, vous serez invitée à participer à un entretien qui durera environ 90 minutes et à remplir ensuite un court questionnaire démographique.

Votre décision de faire partie ou non de cette recherche est complètement libre et volontaire. Si vous décidez de ne pas participer à cette recherche, cette décision n'aura aucun impact sur les soins qui vous sont habituellement dispensés à la clinique.

Pour l'instant, nous souhaitons simplement obtenir votre permission avec vous afin de vous transmettre de plus amples renseignements au sujet de l'étude. Nous vous prions de retourner ce formulaire signé si vous souhaitez effectivement y prendre part. Si vous indiquez que vous aimeriez recevoir de plus amples renseignements au sujet de l'étude, une chercheuse communiquera avec vous par téléphone et vous fournira de plus amples renseignements au sujet de l'étude. Vous aurez ensuite la chance de décider si vous aimeriez y participer ou non.

Ce projet a été approuvé par le Conseil d'éthique en recherches de L'Hôpital d'Ottawa. Si vous avez des questions concernant vos droits à titre de participante ou cette recherche, veuillez communiquer avec le président du Conseil d'éthique en recherches de L'Hôpital d'Ottawa,

Pour toute question concernant ce projet, veuillez communiquer avec :  
Julia DiMillo, Chercheuse principale

Nous vous remercions pour votre temps et votre intérêt.

## Formulaire de demande à des fins de communication avec des patientes

**Titre :** *L'expérience des femmes qui présentent une mutation génétique les prédisposant au cancer du sein : Étude faisant appel à une théorie à base empirique*

**Investigateurs :**

Julia DiMillo

(Chercheuse principale)

D<sup>r</sup> Eva Tomiak, MD

(Co-chercheuse)

D<sup>r</sup> André Samson, PhD.

(Co-chercheur)

Oui, j'aimerais en apprendre davantage au sujet de l'étude.

Si vous avez indiqué que vous aimeriez en apprendre davantage au sujet de l'étude, veuillez compléter l'information suivante. **VEUILLEZ ÉCRIRE EN CARACTÈRES D'IMPRIMERIE.**

Nom : \_\_\_\_\_

Numéro de téléphone :

Jour : (\_\_\_\_) \_\_\_\_\_

Soirée : (\_\_\_\_) \_\_\_\_\_

\_\_\_\_\_  
Signature

\_\_\_\_\_  
Nom (en caractères d'imprimerie)

\_\_\_\_\_  
Date

\*\*\* Une fois complété, veuillez nous faire parvenir ce formulaire dans l'enveloppe ci-jointe.

Nous vous remercions pour votre temps et votre intérêt.

Appendix D  
**Participant Information and Consent Form**

The Experiences of Women Possessing a Genetic Predisposition to Developing Breast  
 Cancer: A Grounded Theory Study

You are being asked to participate in a research study being conducted by Julia DiMillo as a requirement for her master's degree in Educational Counselling, at the University of Ottawa. The primary researcher is being supervised by Dr. André Samson, professor at the University of Ottawa in the Faculty of Education.

**Purpose of this research study**

The purpose of this study is to better understand the experiences of women who have tested positive for mutations of the BRCA1 or BRCA2 genes. More specifically, the researcher would like to examine whether these women experience stigma due to their positive test result, and the impact this might have on their day-to-day life.

In this study, stigma is being defined as experiencing injustice and being singled out by society because of a particular condition occurring in a person's life, or because of their social membership in a certain group.

Your decision to participate in this study is entirely voluntary, and the decision not to participate or to withdraw from the study will not have any impact on the care you receive at the clinic and the Ottawa Hospital.

**Procedures**

If you choose to participate in this study, you will be asked to participate in an interview which is anticipated to last approximately 1 and ½ hours. The primary researcher for this study (Julia DiMillo) will be conducting the interviews. During the interview, you will be asked a series of questions asking you to describe your experience when you received your positive test result, as well as the impact this experience has had on your life. The interview questions will also be provided to you in written format during the interview. You will be asked questions similar to: "Could you please describe your life since you first received your positive test result for the BRCA1 or BRCA2 mutation?". You are not obligated to answer any of the questions, and are free to decline answering them at any time.

Prior to participating in the interview, you will be asked to complete a short questionnaire asking you some basic information about yourself, your family, and your family history of breast cancer. This questionnaire should only take approximately 5-10 minutes to complete.

The interviews for this study will be tape-recorded in order to allow for immediate transcription. This will allow the researcher to better describe, understand and analyze your experiences alongside those of other women who have also received a positive test result. The tape-recorded interviews, as well as all written information, will be given a pseudonym to preserve your identity and will be kept in a secure location to which only the primary researcher will have access. At the end of the interview you will

be given the opportunity to discuss your feelings/concerns, and any questions that may have arisen over the course of the interview.

### **Are there any risks to participating in the research?**

There are no known risks, whether physical or emotional, to participating in this study. However, it is possible that during the completion of the questionnaire, or during the interview, certain questions could make you feel uncomfortable, or may bring up some negative feelings associated with the period surrounding when you received your test results. If you find that any time during this study you experience negative effects or distressing feelings due to the content of the study, please contact Linda Corsini, Social Worker for the Women's Breast Health Center of the Ottawa Hospital, at 613-798-5555, extension 16563.

### **Benefits**

You may not directly benefit from participating in this study. It is possible, although, that sharing your experiences of living with the positive results of your genetic test may be emotionally beneficial to you, allowing you to articulate your experience, and knowing that other women are also undergoing similar experiences. Through your participation in this study, it is hoped that researchers will better understand and conceptualize the experiences of women who have received a positive test result indicating that they possess a mutation of the BRCA1 or BRCA2 genes, and consequently provide them with better care in the future.

### **Withdrawing from the study**

Your participation in this study is entirely voluntary, and there will be no penalty if you choose to withdraw at any time. In the event that you choose to withdraw from the study, please notify the primary researcher. If throughout the course of the study, your welfare becomes a concern to the researcher, she may suggest that you discontinue your participation and withdraw from the study.

**Costs:** You will not be paid for participating in this study.

### **Limits of confidentiality**

All of the personal information entrusted to the researcher will be kept confidential, except as required or permitted by law. Your identity will not be revealed in any publication, presentation or submission of this study. All personal information that leaves the clinic will be given a unique study number in order to preserve your identity and make your name unidentifiable.

If any information that could influence whether you would like to pursue participating in this study comes to light, you will be informed. You will be given a copy of this Information and Consent form which you can keep.

If you have any concerns about your rights as a research participant, you may contact the Chair of the Ottawa Hospital Research Ethics Board (OHREB) at 613-798-5555, extension 14902. The OHREB and the Ottawa Hospital Research Institute (OHRI) may audit the records of this study.

### Questions

Please contact Julia DiMillo, the Principal Investigator for this study at 613-837-4712 or through email at [jdimi009@uottawa.ca](mailto:jdimi009@uottawa.ca)

### Consent

I have read the above statements and any questions I may have concerning the research have been answered. I am voluntarily agreeing to participate in this study. I will receive a copy of this three-page consent form.

- Yes, I agree to be contacted for further follow-up questions.
- No, I do not agree to be contacted for further follow-up questions.

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Name of Participant (Please print)

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Signature of Participant

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Date

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Name of Investigator/Delegate

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Signature of Investigator

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Date

Version date: April 14, 2009

## **Formulaire d'information et de consentement des participantes**

### L'expérience des femmes qui possèdent une mutation des gènes BRCA1 ou BRCA2 : Une recherche utilisant une théorie ancrée

Vous êtes invité à participer dans une étude conduite par Julia DiMillo pour satisfaire les exigences de sa maîtrise en counselling éducationnel. La chercheuse sera supervisée par le Dr. André Samson, professeur de l'Université d'Ottawa, faculté d'éducation.

#### **L'objectif de la recherche**

L'objectif de cette recherche est de mieux comprendre les expériences vécues par les femmes qui ont reçu un résultat positif indiquant qu'elles ont une mutation des gènes BRCA1 ou BRCA2. Plus particulièrement, la chercheuse aimerait mieux comprendre l'expérience de ces femmes face au stigma, et l'impact que cela pourrait avoir dans leur quotidien.

Au cours de cette étude, le concept du stigma sera défini comme ayant éprouvé une injustice et ayant été singularisé par la société due à une condition spécifique survenant dans la vie d'un individu, ou due à leur appartenance à un certain groupe.

Votre décision de faire partie de cette recherche est complètement libre et volontaire et votre décision de participer à cette recherche n'aura aucun impact sur les soins que vous recevez régulièrement à la clinique ou à l'Hôpital d'Ottawa.

#### **Les procédures de la recherche**

Si vous acceptez de participer à cette recherche, vous serez invité à participer à une session d'interview qui durera environ 1 heure et demie. Cette session sera menée par la chercheuse principale de cette étude (Julia DiMillo). Au cours de l'interview, on vous posera une série de questions au sujet de vos expériences depuis que vous avez reçu la nouvelle de votre résultat positif, ainsi que l'impact que ces expériences ont eu sur votre vécu. Les questions vous seront fournies en format écrit durant l'interview. On vous invitera à répondre à des questions semblable à : « Pourriez-vous s'il vous plaît décrire votre existence depuis que vous avez appris que vous aviez une mutation des gènes BRCA1 ou BRCA2 ? ». Vous n'êtes pas obligé de répondre à aucune de ces questions, et vous pouvez refuser de répondre à n'importe quel moment.

Avant que l'interview débute, vous serez invitée à compléter un court questionnaire démographique avec l'objectif de récolter de l'information générale, telle que l'historique du cancer du sein dans votre famille. Ce questionnaire devrait prendre environ 5-10 minutes à compléter.

Chaque interview faisant partie de cette recherche sera enregistrée pour faciliter la transcription des données. De plus, cela facilitera aussi l'analyse des données, ainsi que la compréhension de votre expérience. Pour conserver votre identité, chaque interview et toute information écrite sera donné un pseudonyme et gardé dans un endroit sécuritaire auquel seule la chercheuse principale aura accès. À la conclusion de chaque interview,

vous aurez la chance de discuter vos sentiments/inquiétudes, et poser les questions auxquelles vous avez songé au cours de l'interview.

### **Quels sont les risques potentiels entraînés par votre participation ?**

Votre participation à cette recherche n'entraîne aucun risque ou aucune conséquence négative auxquels nous ne sommes au courant. Cependant, il se pourrait que certaines questions du questionnaire ou de l'interview vous rendent mal à l'aise ou elles pourraient évoquer certaines émotions négatives. Par conséquent, si vous trouvez que votre participation à cette recherche vous cause une forme de détresse émotionnelle, contactez Mme. Linda Corsini, travailleuse sociale pour le Centre de santé du sein de la femme de l'Hôpital d'Ottawa, au (613) 798-5555, poste 16563.

### **L'apport de votre participation**

Il se pourrait que vous ne bénéficiiez pas personnellement en participant à cette recherche. Cependant, il est aussi possible qu'en partageant votre expérience que vous bénéficiiez émotionnellement grâce à avoir discuté de vos réussites et de vos difficultés. Votre participation à cette recherche risque d'aider aux chercheurs à mieux comprendre l'expérience des femmes qui vivent actuellement une situation semblable. Par conséquent, les soins offerts aux femmes possédant une mutation des gènes BRCA1 ou BRCA2 pourront potentiellement être améliorés dans le futur.

### **Le retrait de votre participation à cette recherche**

Votre participation à cette recherche est complètement volontaire, et il n'y aura aucune conséquence si vous décidez de vous retirer à n'importe quel moment. Si vous choisissez de vous retirer du projet, veuillez s'il vous plaît informer la chercheuse principale. Si au cours de l'étude la chercheuse s'inquiète de votre bien-être, il se pourrait qu'elle suggère que vous cessiez de participer, et que vous vous retirez de la recherche.

**Coûts :** Votre participation dans cette recherche ne sera pas rémunérée.

### **Les limites de la confidentialité**

Toute information divulguée à la chercheuse au cours de la recherche sera gardée strictement confidentielle à tout moment, à moins de stipulations contraires prévues à la loi. Votre identité ne sera jamais révélée, même si la recherche est présentée ou soumise pour publication. Toute information qui sort de la clinique sera assignée un code unique, pour préserver votre identité et pour assurer que vous n'êtes pas identifiée.

Si aucune information qui pourrait influencer votre motivation pour participer à cette recherche se présenterait, vous serez informé dans les plus brefs délais. Vous allez recevoir une copie de se formulaire d'information et de confidentialité que vous pourrez garder.

Si vous avez des soucis concernant vos droits comme participante, vous pouvez contacter le président du Conseil d'éthique en recherche de l'Hôpital d'Ottawa au  
Le Conseil d'éthique en recherche de l'Hôpital d'Ottawa et  
l'Institut de Recherche de l'Hôpital d'Ottawa pourra auditer les données de cette recherche.

### Questions

Contactez s'il vous plaît Julia DiMillo, la chercheuse principale de cette étude

### Consentement

J'ai lu le document, et toutes les questions que j'ai posées au sujet de la recherche ont été répondues. En signant ce document, j'accepte volontiers à participer à cette recherche. J'ai reçu une copie de ce formulaire de trois pages.

- Oui, je consente d'être contactée pour des questions supplémentaires.
  
- Non, je ne consente pas d'être contactée pour des questions supplémentaires.

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Nom de la participante (lettres moulées)

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Signature de la participante

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date

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Nom de la chercheuse

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Signature de la chercheuse

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date

Version : le 14 avril, 2009

## Appendix E

## Interview Protocol

The following questions are for a study attempting to better understand the experiences of women who possess mutations of the BRCA1 or BRCA2 genes.

Please describe your experiences to the best of your ability, and in as much detail as possible. If you feel that certain questions make you uncomfortable, please do not answer them. I would like to remind you that you are not obligated to answer any of these questions, and that you may remove yourself from the study at any time with no adverse consequences. Do you have any questions before we begin?

- 1) Could you please describe your life since you first received your positive test result for the BRCA1 or BRCA2 mutation?
- 2) Please describe how this news has impacted your life. How has this experience impacted:
  - You emotionally?
  - The way you perceive yourself?
  - Your decision-making for the future?
  - Those you care for such as your family and friends?
  - The way your family and friends treat you?
  - The level of support you receive from family and friends?
- 3) Since you received your positive test result, could you describe an incident when you felt that you have:
  - Experienced prejudice, or that others have judged you?
  - Have been treated differently by others?

- Have been placed in a different category than others?
  - Felt more comfortable amongst other women who have received the same test result?
- 4) Do you have any final comments or questions for me as we conclude the interview?

## Entrevue

Décrivez s'il vous plaît votre expérience avec le plus de détails possible, et du mieux que vous ne le pouvez. Si vous trouvez que certaines questions vous rendent mal à l'aise, vous n'êtes pas obligés de répondre. D'ailleurs, vous n'êtes pas obligée de répondre à aucune de ces questions, et vous pouvez vous retirer de cette étude à n'importe quel moment sans conséquences négatives. Avez-vous des questions avant que l'entrevue débute?

- 1) Pourriez-vous s'il vous plaît décrire votre existence depuis que vous avez appris que vous aviez une mutation des gènes BRCA1 ou BRCA2?
- 2) Décrivez s'il vous plaît l'impacte de cette nouvelle sur votre vie.
  - Au point de vue émotionnel.
  - La façon ce dont vous vous percevez?
  - La façon selon laquelle vous faites des décisions pour le futur?
  - Vos relations avec ceux que vous aimez, comme votre famille et vos amis?
  - La qualité du soutien que vous recevez de votre famille et/ou de vos amis?
- 3) Depuis que vous avez reçu votre résultat positif:
  - Vous sentez-vous différente des autres?
    - Si oui, comment, et pouvez-vous me décrire un incident en particulier?
  - Est-il plus facile de communiquer avec des femmes qui partagent la même expérience que vous?

- Si oui, pouvez-vous me donner un exemple particulier?

4) Avez-vous des commentaires ou des questions à ajouter?

## Appendix F

*Context Following the Receipt of Test Results. Major Theme of Self-Perception: I Want to Be Me Again. Description of the Categories and Sub-Categories*

Cognitions: Trying to Make Sense of Reality		Emotions: I Don't Know What to Do	Behaviours: A Quest for Control	
Adaptive: I Think I Can Do This	Protective: If Only I Could Turn Back Time		Adaptive: Making Changes	Protective: Seeking Control
<ul style="list-style-type: none"> <li>• Seeing the process as a whole</li> <li>• Looking at the process from a third perspective</li> <li>• Acceptance</li> <li>• Not dwelling</li> <li>• Feeling relaxed</li> <li>• Being thankful</li> <li>• Having a positive outlook</li> <li>• Sense of empowerment</li> <li>• Hoping to choose the right path</li> <li>• Wanting to help others</li> <li>• Wanting to become an advocate</li> <li>• Wanting to educate others</li> <li>• Wanting to connect with similar others</li> <li>• Convincing</li> </ul>	<ul style="list-style-type: none"> <li>• Ignorance is bliss</li> <li>• Denial</li> <li>• Minimizing</li> <li>• Avoidance</li> <li>• Justification</li> <li>• Wanting to be in control</li> <li>• Hoping to have less genetic impact</li> <li>• Contemplating if she hadn't been tested</li> <li>• Belief that one should have control over getting cancer</li> <li>• Internal pressure</li> <li>• Contemplating having breast cancer</li> </ul>	<ul style="list-style-type: none"> <li>• Guilt</li> <li>• Regret</li> <li>• Shock</li> <li>• Anger</li> <li>• Overwhelmed</li> <li>• Sadness</li> <li>• Frustration</li> <li>• Feeling upset</li> <li>• Feelings of depression</li> <li>• Devastation</li> <li>• Hopelessness</li> <li>• Worry</li> <li>• Confusion</li> <li>• Feeling strange</li> <li>• Feeling powerless</li> <li>• Fear of making changes</li> <li>• Fear of her options (losing her breasts/ovaries)</li> <li>• Fear for her children</li> <li>• Fear for family</li> <li>• Fear for her family's</li> </ul>	<ul style="list-style-type: none"> <li>• Proactive behaviour</li> <li>• Trying to make healthy changes</li> <li>• Seeking additional information</li> <li>• Keeping her options open</li> <li>• Planning for the future</li> <li>• Educating herself</li> <li>• Living in the moment</li> <li>• Making a conscious effort to take opportunities</li> <li>• Seeking others who share her viewpoint</li> <li>• Coping based on past experience(s)</li> <li>• Voicing that she is different</li> <li>• Choosing to know</li> </ul>	<ul style="list-style-type: none"> <li>• Attempting to control the future</li> <li>• Putting on a brave face</li> <li>• Focusing on the process/details</li> <li>• Using statistics</li> <li>• Giving up</li> <li>• Avoiding other for fear of loss</li> <li>• Need to keep the secret</li> </ul>

herself		health	
<i>Context Following the Receipt of Test Results: Self-Perception. Description of the Categories and Sub-Categories</i>			
Cognitions: Trying to Make Sense of Reality		Emotions: I Don't Know What to Do	Behaviours: A Quest for Control
Adaptive: I Think I Can Do This	Protective: If Only I Could Turn Back Time		Adaptive: Making Changes      Protective: Seeking Control
<ul style="list-style-type: none"> <li>• Hoping for support</li> <li>• Hoping for a positive outcome</li> <li>• Wanting to be treated like others</li> </ul>		<ul style="list-style-type: none"> <li>• Fear of other's reactions</li> <li>• Fear of prejudice</li> <li>• Fearing a child's resentment</li> <li>• Fear of spilling her secret</li> <li>• Feeling vulnerable</li> <li>• Feeling isolated</li> <li>• Uncertainty</li> <li>• Insecurity</li> <li>• Doubt</li> <li>• Blaming herself</li> <li>• Feeling less like a woman</li> </ul>	<ul style="list-style-type: none"> <li>• Realization that something must be done</li> <li>• Vigilance</li> <li>• Turning to technology for social support</li> </ul>

## Appendix G

*Context Following the Receipt of Test Results. Major Theme of Perception of Others:  
Can They Accept the New Me? Description of the Categories*

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Source of Help/Support: I Don't Know What I Would do Without Them	Source of Distress: I Wish They Would Treat Me Like Before
<ul style="list-style-type: none"> <li>• Social support</li> <li>• Friendship</li> <li>• Support from husband</li> <li>• Comfort in knowing others share her viewpoints</li> <li>• Support from similar others</li> <li>• Feeling accepted</li> <li>• Medical support</li> <li>• Solidarity with other BRCA gene mutation carriers</li> </ul>	<ul style="list-style-type: none"> <li>• Feeling misunderstood</li> <li>• Feeling judged</li> <li>• Rejection</li> <li>• Avoidance from family</li> <li>• Familial ignorance</li> <li>• Others' opinions</li> <li>• External pressure</li> <li>• Perceived pity</li> <li>• Perceived insensitivity</li> <li>• Public's ignorance</li> <li>• Others' disagreement with testing</li> </ul>

## Appendix H

*An Uncertain Conclusion to an Unending Process: Living with the BRCA Genetic Mutation. Description of the Categories*


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Unmet Needs: Unanswered Questions	Effects on her Womanhood: Should I Have More Children?	Effect on her Reality: I Wish I Could Change My Reality	Impact on Loved Ones: When Should I Tell My Children?
<ul style="list-style-type: none"> <li>• Inadequate support</li> <li>• Lack of guidance</li> </ul>	<ul style="list-style-type: none"> <li>• Feeling a desire to conceive after testing</li> <li>• The ethics of childbearing after testing</li> </ul>	<ul style="list-style-type: none"> <li>• Allowing herself to take a glimpse at reality</li> <li>• Choosing to face reality</li> <li>• Relief at knowing the truth/facing reality</li> <li>• Being forced to face reality</li> <li>• Wishing she could change her reality</li> <li>• Realizing the impact of the test result</li> <li>• Realizing the importance/ impact of support</li> </ul>	<ul style="list-style-type: none"> <li>• Familial discord</li> <li>• Impact of the test on family members</li> <li>• Testing children</li> <li>• Telling her children</li> <li>• Trying to protect her children from the future</li> <li>• Fear possessed by family members</li> <li>• Impact on raising her children</li> </ul>

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## Appendix I

*An Uncertain Conclusion to an Unending Process: Medical Consequences. Description of the Categories*


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Medical Procedures: I Hope This is Not Cancer	Relations with the Medical Staff: I Hope He/She Agrees With My Chosen Course of Action
<ul style="list-style-type: none"> <li>• Ovary removal</li> <li>• Medical complications</li> <li>• Potential scares</li> </ul>	<ul style="list-style-type: none"> <li>• Feeling as though she is treated with superior care</li> <li>• Encountering new professionals</li> <li>• Frustrations with the medical team</li> <li>• Choosing to go against Doctor recommendations</li> </ul>

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