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UNIVERSITY OF SAN DIEGO
Hahn School of Nursing and Health Science
DOCTOR OF PHILOSOPHY IN NURSING

Managing Susceptibility to Hereditary Breast and Ovarian Cancer

by

Cynthia E. Perry, RN, MSN

A dissertation presented to the

FACULTY OF THE HAHN SCHOOL OF NURSING AND HEALTH SCIENCE

UNIVERSITY OF SAN DIEGO

In partial fulfillment of the

requirements for the degree

DOCTOR OF PHILOSOPHY IN NURSING

Date: July 2005

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MANAGING SUSCEPTIBILITY TO HEREDITARY BREAST AND OVARIAN CANCER

Abstract

The recent identification of Breast Cancer 1 (BRCA1) and BRCA2 genes offers an opportunity for high-risk individuals to learn whether they may be genetically predisposed to develop breast and/or ovarian cancer. The purpose of this study was to examine how unaffected women, identified as BRCA positive and variant of uncertain significance (VUS) mutation carriers, managed their susceptibility to hereditary breast and ovarian cancer (HBOC). Thirty North American women ranging in age from 22 to 60 years responded to open-ended interviews. These interviews were analyzed using constant comparative method to generate a grounded theory.

Managing Susceptibility was identified as the basic social process, which characterized how these unaffected women responded to genetic testing and managed their risk of HBOC. Five categories were found that explain the actions, interactions, and consequences of managing susceptibility. These were: (a) gaining awareness, (b) confronting uncertainty and getting tested, (c) disclosing results, (d) deliberating and making risk management decisions, and (e) reflecting on actions.

These women regarded breast and/or ovarian cancer as a predictable outcome, given their family history, and felt they had a responsibility to their family to prevent this danger if possible. After gaining awareness of their increased risk, they sought genetic counseling to take responsibility for their perceived susceptibility and were influenced by feelings of obligation to their family. Participants disclosed their test results to seek support and because of a sense of duty to inform their family members of their risks, no

matter how difficult it was for them personally. They also felt they had a responsibility to persuade their family to act on the information. Past family and personal experiences, present view of themselves and their relationships, and aspirations for the future were all part of their complex risk management decision making. Engaging in risk management was seen as providing them with control over their susceptibility to HBOC. Those choosing prophylactic surgeries wanted to prevent cancer, as they were not satisfied with the limitations of vigilant surveillance which provided only early detection. By taking these measures they not only gained some control over their lives, but as importantly, could maintain their identity as mother and nurturer.

The study's findings support other research in genetic testing and risk management and have important implications for health policy, nursing practice, and future research.

DEDICATION

To Emma and Earl, my mother and father, who nurtured my love for learning.

To my husband Gene and daughter Tera: thanks for being my partners in this journey and especially for believing in me!

ACKNOWLEDGMENTS

It was only with the support and assistance of several wonderful people that this dissertation became a reality. I would like to express my sincerest thanks to:

The women who have shared their stories with me. Each one contributed in her own personal style, but each one with honesty, courage, and spirit. This dissertation would not have been possible without the voices of these women, moved by their desire to help others. I am so grateful to each person who shared how she courageously managed her susceptibility to hereditary breast and ovarian cancer.

A talented social scientist and nurse, Dr. Mary Rose Muller, my chairperson and cheerleader who gave me countless hours of her valuable time and expertise, her constant encouragement, endless patience, and accurate and sensitive feedback throughout this dissertation journey. Your careful attention to structure was the magic that transformed this into a finished dissertation. Thank you too for your guidance in helping obtain my Oncology Nursing Foundation grant.

A very generous professor and nurse, Dr. Diane Hatton, my committee member and member of my qualifying exam committee. I owe a debt of gratitude for her believing in the worth of this project from the start, for helping me understand grounded theory method, and for mentoring me through manuscript publication and another research process. Thank you for your constant encouragement, valuable insight, and good humor.

A gifted and generous physician, Dr. Linda Wasserman, my committee member and mentor from the University of California, San Diego. For allowing me the opportunity of access to her cancer genetics practice and BRCA support group, for

believing in my undertaking from the start, and providing me with a monthly forum for discussing ideas and issues over the last three years. Thank you for your vast experience, knowledge, support, and advice.

To Dr. Patricia Roth for chairing my qualifying exam committee and for her belief and support in this adventure into cancer genetics. For supporting my attending the Summer Genetics Institute and my nomination for the ARCS Foundation scholarship. Thanks for your constantly upbeat and encouraging words and wonderful example as a nurse educator and leader.

For three supportive and endlessly helpful genetic counselors, who have always been valuable peer reviewers of my ideas and openly shared their genetic counseling experiences: Dr. Eric Rosenthal, Kristin Kalla, and Amy Tranin. For giving their time, helping me make the connections within their healthcare systems, help with recruitment, and for encouragement all along the way, you are the best.

Mindy Tinkle and Francine Nichols, nurse researchers and educators at the National Institute of Nursing Research, Summer Genetics Institute, who generously shared their knowledge and gave me the confidence to begin my research with their thoughtful review of my first research proposal in genetics.

Nancy Gillespie and Nancy Bennett who transcribed hours of audiotapes. Their attention to detail, listening and replaying tapes to capture every word and inflection, demonstrated respect for the women who participated in this study.

Two dedicated executive directors of peer support groups for hereditary breast and ovarian cancer, Sue Friedman, FORCE and Joanne Riediger-Duebel, Hereditary

Breast and Ovarian Society of Canada for providing access to the FORCE research website and supporting my research.

To my friends and colleagues, Melissa Kaime, Judee Wood, Lori Judson, Karen Nielsen-Manicucci, Lisa Kaiser, Linda Hansen Kyle, Pat Bradley, Leonie Sutherland, Mary Ann Simanello and my other classmates. You have patiently supported me during the course of this study. Thank you for your encouragement to continue until completion.

To the Hahn School of Nursing, Oncology Nursing Foundation, ARCS Foundation – Achievement Rewards for College Scientists, and National Institute of Nursing Research, for their generous funding of education and research, which meant a great deal to me.

Finally, always there for support, advice, comfort, care, and feeding, my family who patiently endured my neglect as I worked on this dissertation. Thank you, Gene, Tera, and Emma – you waited with such good grace till all this dissertation business was finished and still remembered who I am.

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CHAPTER 1

Focus of the Inquiry

The revolution in genetics and molecular biology over the last ten years provides new opportunities for the prevention of hereditary breast and ovarian cancer. The recent identification of Breast Cancer 1 (BRCA1) and Breast Cancer 2 (BRCA2) susceptibility genes offers an opportunity for high risk individuals to learn whether they may be genetically predisposed to develop breast or ovarian cancer. Although these advances may represent major steps forward in the battle against cancer, they also raise complicated questions about the value and consequences of genetic testing. The impact of this new genetic knowledge on client experiences of genetic testing and risk management is not well understood. This dissertation sought to fill part of this gap by proposing a grounded theory of how unaffected BRCA positive and variant of uncertain significance (VUS) mutation carriers conceptualized their breast and/or ovarian cancer risk, interpreted and coped with BRCA test information, and managed their susceptibility to breast and ovarian cancer.

Background

During the average lifetime, one in eight women will be affected by breast cancer (Feuer & Wun, 1999). It is the second most common cause of cancer-related death in

women, with an estimated 211,240 new cases and 40,410 fatalities in 2005 (American Cancer Society, 2005). Ovarian cancer accounts for 4% of all cancers among women and causes more deaths than any other cancer of the female reproductive system. In 2005, it is estimated that ovarian cancer will be diagnosed in 22,220 women and almost 16,210 will die from it (ACS, 2005).

One of the major advances in the understanding of breast and ovarian cancer during the past 10 years is the recognition that some of these cancers have a genetic basis. Because genes predispose to cancer, the evolving technology of genetic testing allows presymptomatic testing of persons at high risk. This testing provides information about a person's susceptibility to a disease and is referred to as predictive or susceptibility testing. Approximately 5% to 10% of breast and ovarian cancers are hereditary (Krainer et al., 1997).

Identification of the Breast Cancer 1 (BRCA1) and Breast Cancer 2 (BRCA2) gene mutations in 1994 and 1995, respectively, has enabled women to obtain more precise estimates of their risk of developing breast and/or ovarian cancer (Miki et al., 1994; Wooster et al., 1994). Women with a mutation in the BRCA1 and BRCA2 genes are at an increased lifetime risk for developing breast and ovarian cancer. This risk varies according to age, ethnicity, and family history (Ford, Easton, Stratton, Narod, Goldgar, & Devilee, 1998; Struewing et al., 1997). Based on pooled data from 22 studies, among first degree relatives of 500 index patients with BRCA mutations, the average cumulative risks of breast and ovarian cancer by age 70 years in BRCA1 carriers were 65% and 39% respectively. The risk estimates for BRCA2 mutations were 45% and 11% respectively (Antoniou et al., 2003). This is the situation likely to be encountered in clinical genetics

situations. Higher estimates were reported in the New York Breast Cancer Study (King, Marks, & Mandell, 2003) of 104 mutation-positive Ashkenazi Jewish women with breast cancer. Estimates were based only on relatives whose mutation status were known. These breast cancer estimates were 69% and 74% by age 70 years, for BRCA1 and BRCA2 mutation carriers, respectively, and 46% and 12% for ovarian cancer for BRCA1 and BRCA2, respectively (King et al., 2003).

Family history characteristics that have been associated with an increased likelihood of carrying a BRCA mutation include the following (a) multiple cases of breast cancer in the family, (b) both breast and ovarian cancer in the family, (c) one or more family members with 2 primary cancers, and (d) Ashkenazi Jewish background (Couch, 1997; Frank et al., 1998; Shattuck-Eidens et al., 1997).

Hereditary breast and ovarian cancer (HBOC), like that caused by the BRCA mutations, is characterized by early onset of breast cancer (5-15 years earlier than non-hereditary cases), bilateral breast involvement, a history of both breast and ovarian cancer, breast cancer in male family member(s), vertical transmission through both maternal and paternal lines, and familial association with tumors of other organs, especially the ovary and prostate gland (Couch, 1997; Frank et al., 1998; Narod et al., 1991; Parmigiani, Berry, & Aguilar, 1998; Sellers et al., 1994; Shattuck-Eidens et al., 1997)

One of the major benefits of BRCA genetic testing is the possibility of more individually tailored interventions to reduce breast and ovarian cancer mortality. However, the optimal strategy for achieving this goal remains unresolved. Today, women with positive BRCA mutations must make a decision between risk reduction and early

detection. The risk reduction options are chemoprevention and surgical interventions with bilateral prophylactic mastectomy (BPM) and bilateral prophylactic oophorectomy (BPO). Early detection involves vigilant breast and ovarian screening, including self- and clinical breast exams, mammography, magnetic resonance imaging, transvaginal ultrasound, and cancer antigen 125 testing (Burke et al., 1997).

Although recent studies (Hartmann et al., 2001; Hartmann et al., 1999; Kauff et al., 2002; Meijers-Heijboer et al., 2001; Rebbeck et al., 2004; Rebbeck et al., 2002; Scheuer et al., 2002) indicate that surgical interventions may reduce the incidence of breast and ovarian cancer, the available screening and chemoprevention options have unproven effectiveness in women with BRCA mutations (Brekelmans et al., 2001; King et al., 2001; Komenaka et al., 2004; Kuhl et al., 2000; Meijers-Heijboer et al., 2001; Narod et al., 2000; Scheuer et al., 2002; Tilanus-Linthorst, Obdeijn, Bartels, de Koning, & Oudkerk, 2000; Warner et al., 2001). As a result, recommendations for management are generally based on expert opinion (Burke et al., 1997).

Decision Making in a BRCA Context

Predictive genetic testing informs us only about a future condition that may (or may not) develop. Although the lifetime risk is high, there is always a substantial component of uncertainty. There is uncertainty about whether the condition will develop, when, and how severe it will be. There is further uncertainty about the risk reduction and disease detection interventions available. Thus decision making has the potential to elicit a state of uncertainty or decisional conflict about which course of action to take (Guerriere & Llewellyn-Thomas, 2001; O'Connor, 1993).

In BRCA related decisions, options are risky, as any choice involves negative aspects which may contribute to avoidance-avoidance conflict. Outcomes for these

decisions are multidimensional, involving potential consequences at many levels (e.g. health, emotional well-being, or insurance loss). The decisions are linked sequentially: first there is the decision to get tested, and next the behavioral decision of what to do with the results.

Genetic counseling and testing rely heavily on risk communication to provide information about personal and familial cancer risk (Botkin et al., 1996; Peters & Stopfer, 1996). An accurate understanding of risk among participants may be critical to their decision making about whether to test, and for those who receive positive or variant of unknown clinical significance test results, to their decision making about risk management (Croyle & Lerman, 1999). The emerging literature on risk communication suggests that most individuals with some family history of cancer, including those at low to moderate risk, overestimate their personal cancer risk. This finding of exaggerated perceptions of personal risk has been documented in research on hereditary breast and ovarian cancer families (Berry, Parmigiani, Sanchez, Schildkraut, & Winer, 1997; Bluman et al., 1999; Hallowell, Statham, & Murton, 1998b; Schwartz et al., 2000; Struewing, Lerman, Kase, Giambarresi, & Tucker, 1995; Winer, Borstelmann, & Bluman, 1997). Participant decision making about genetic testing is influenced less by their actual risk than by their perceived risk and emotional factors (Lerman, Tercyak, Croyle, & Hamann, 2002). A meta-analysis of 12 studies of outcomes of genetic counseling for breast/ovarian cancer showed that counseling improved the accuracy of risk perception (Meiser & Halliday, 2002).

In addition, cancer worry and cancer specific distress have been shown to motivate use of BRCA genetic testing in high risk families (Durfy, Bowen, McTiernan,

Sporleder, & Burke, 1999; Lerman, Schwartz et al., 1997). Skirton (2001) in a grounded theory study of genetic counseling found the need for certainty emerged as a powerful factor that motivated clients to pursue genetic counseling. Webster and Kruglanski (1994) describe the need for cognitive closure as an individual drive for certainty and a discomfort with ambiguity. Skirton proposed that clients who request a referral to genetic services may have a greater need for closure than those at genetic risk who avoid or do not seek genetic counseling. This could account for the variability in approaches to genetic counseling between different members of the same family. Obtaining certainty may be a way of enabling a client to gain control of the situation. Berkenstadt, Shiloh, Barkai, Katznelson, and Goldman (1999) found that counselees who obtained a more certain diagnosis or specific recurrence risk felt a greater sense of control.

Research suggests that emotional factors can modify the cognitive processing of risk-related information when an individual is faced with a personally relevant health threat (Croyle, Yi Chun, & Hart, 1997; Leventhal, Safer, & Panagis, 1983). This is common in risky decisions made under emotional stress (Janis & Mann, 1977). Given the possibility of receiving a positive genetic test, one would expect genetic testing to be particularly stressful and effect cognitive processing.

Important to a client's decision making after BRCA mutation testing is to understand the impact of undergoing predictive testing upon emotional state. Knowledge of anxiety and distress are important because psychological distress has the potential to interfere with the clients' understanding and synthesis of genetic and medical information, and to impair adherence to recommendations for screening and possible prevention (Lerman & Schwartz, 1993; Lerman, Trock, & Rimer, 1991).

The few studies of psychological outcomes associated with genetic testing for BRCA mutations have shown no increase in distress among those who receive positive or negative test results (Croyle, Smith, Botkin, Baty, & Nash, 1997; Lerman & Croyle, 1996; Schwartz et al., 2002). This was also true in one long-term study (n= 65 female participants) that explored the psychosocial consequences of carrying a BRCA mutation. Five years after genetic testing, BRCA carriers did not differ from non-carriers on several distress measures (van Oostrom et al., 2003). This is consistent with the psychological consequences (including anxiety, depression, general distress, and situational distress) of predictive genetic testing in general. Test results did not predict emotional consequences in two systematic review of predictive genetic testing (Broadstock, Michie, & Marteau, 2000; Butow, Lobb, Meiser, Barratt, & Tucker, 2003). Despite findings of diminished distress in tested individuals, most studies also report increased distress among small subsets of tested individuals. However, most of these increases are within the normal range of distress (CancerNet, 2005; Smith, West, Croyle, & Botkin, 1999). In interpreting these studies, the authors caution that all are from programs in which results disclosure were preceded by extensive genetic counseling about risks and benefits of BRCA testing and psychological assessment.

These studies on psychological consequences indicate that obtaining genetic testing may be less stressful than living with the awareness of familial risk for cancer (Coyne, Kruus, Racioppo, Calzone, & Armstrong, 2003). Mediating factors include the test result status of other family members. Female BRCA carriers who were the first in their families tested or whose siblings were negative, had significantly higher distress than other female BRCA carriers (Smith et al., 1999). Wylie, Smith, and Botkin (2003)

reported significantly higher levels of distress in BRCA mutation carriers whose spouse was highly anxious and non-supportive. Thus it is important that research consider the context of the individual tested to determine which individuals requesting genetic testing may require additional emotional support.

While the motivation given for pursuing genetic susceptibility testing includes improving health behaviors (Isaacs et al., 2002; Jacobsen, Valdimarsdottir, Brown, & Offit, 1997; Lerman, Seay, Balshem, & Audrain, 1995; Struwing et al., 1995), it remains unclear to what degree BRCA mutations carriers will alter their breast and ovarian cancer risk management behaviors. Three of four studies have reported increased screening behavior from baseline in BRCA mutation carriers. Mammography screening uptake ranged from 59% to 93.4%, with lower uptake rates in younger carriers. These results are far from ideal.

Breast Cancer Screening

Lerman et al. (2000) reported that disclosure of positive BRCA mutation test results did not lead to increased use of annual mammograms or ovarian cancer screening tests in women (n=29) in a hereditary cancer registry. Sixty eight percent complied with mammography recommendations before BRCA testing and 68% reported adherence one year after receiving positive test results. Use of CA-125 testing and transvaginal ultrasound (TVUS) were 21% and 15%, respectively, one year post testing.

Peshkin et al. (2002) determined from a prospective observational study of 41 BRCA carriers, overall, the use of breast cancer screening was good (clinical breast exam uptake for carriers: 95%; noncarriers: 77%), including mammography uptake (in carriers: 59%, in noncarriers: 47%). However, there was a relatively low uptake rate of

mammography in younger carriers (ages 25-39: 39% versus age \geq 40: 74%). Schwartz et al. (2003) reported that CA-125 and TVUS screening was 43% and 40% respectively, both reflecting an increased use compared with the year prior to testing.

Botkin et al. (2003) studied women for two years following BRCA 1 testing. Both carriers and non-carriers significantly increased their use of mammography and breast self-exam from baseline. For women 40 years and older, 82% of mutation carriers obtained a mammogram in each year following testing. Younger carrier women also significantly increased their mammography utilization from baseline. However, overall, 29% of the carrier women did not obtain a single mammogram by 2 years post-testing. At one and two years post testing, they reported TVUS use of 26% and 11% respectively, and CA-125 use of 32% and 37% respectively.

Scheurer (2002) also presented prospective evidence that BRCA testing and genetic counseling increased screening in 251 BRCA mutation carriers followed over a mean of 24.8 months. There was an overall significant increase in mean number of mammograms, clinical breast exams (CBE), ovarian ultrasonograms, and CA-125 determinations after genetic testing. On average after 15 months, 93.4% had a screening mammogram, 83.3% were performing breast self exam (BSE), 97.4% had a screening CBE, 67.6% had CA-125 testing and 72.9% had a TVUS.

One concern about genetic testing for cancer risk is the possibility that testing-related distress would reduce adherence to cancer screening. However, this has not been demonstrated among participants who have been tested in either Botkin et al. (2000) or Lerman et al. (2000) studies. It may be that genetic information leads to risks being

perceived as unmodifiable and to less adherence to behaviors that would lower health risks (Senior, Marteau, & Peters, 1999).

Although these are the first studies to characterize screening behaviors following BRCA mutation testing, they shed little light on the factors that influenced these surveillance decisions. Limitations of these studies are the relatively short timeframe for follow-up and that most data were obtained from clinical research programs involving very high-risk families. Results from clinical settings remain to be reported.

Prophylactic Surgeries

A few studies have recently been published indicating the extent to which prophylactic surgeries are chosen as HBOC prevention options in BRCA mutation carriers. These studies also included factors that influenced the decisions for prophylactic surgeries and the psychosocial implications of these surgeries for women with BRCA mutations.

Studies from the Netherlands demonstrate different results in the selection of prophylactic surgeries by women with BRCA mutations than the United States. In the Netherlands, Meijers-Heijboer et al. (2000) reported that 51% of unaffected (cancer free) BRCA mutation carriers chose BPM over screening and 64% chose BPO within 2 years after testing. Parenthood was found as a predictor for BPM and age was associated with BPO.

Studies from the United States have reported fewer women choosing prophylactic surgery following BRCA mutation testing. Scheuer et al. (2002) studied 233 affected and unaffected women with BRCA mutations over a mean period of 24.8 months. They reported that 14.9% underwent BPM at a median of 5.3 months after test results and

50.3% underwent salpingo-oophorectomy at a median of 3.4 months after receiving genetic test results. Women electing BPM were younger and had a stronger family history of breast and ovarian cancer than those opting for screening. Those electing BPO were older (64% >40years) and more likely to have had a prior breast cancer diagnosis than those not opting for surgery. Lerman et al. (2000) reported only 3% of unaffected carriers had undergone BPM within a year of learning their BRCA mutation status and 13% obtained BPO. In a Utah kindred of BRCA1 mutation carriers followed for two years, Botkin et al. (2003) found that oophorectomy was obtained by 46% (12/26) of carriers, including 78% (7/9) of women 40 years of age and older. It was also noted that an additional 30% (11/37) of the women in this study had obtained BPO before testing. In contrast, mastectomy was not utilized within the first 2 years following testing, although 11% were considering this procedure.

Lodder et al. (2002), from the Netherlands group, indicated that BPM decisions were not only related to higher general and cancer-related distress but women opting for this choice were more often in their thirties, had young children, and had a longer awareness of the genetic nature of cancer in the family, than those opting for screening. This finding, that the level of general and cancer related distress influences the degree to which BPM is chosen as a risk reduction intervention in women at risk for HBOC, has been reported in other studies (Lodder et al., 2002; Meiser, Butow, Freidlander et al., 2000; Scheuer et al., 2002; Stefanek, Hartmann, & Nelson, 2001; Stefanek, Helzlsouer, Wilcox, & Houn, 1995; Wagner et al., 2000).

The psychological sequelae of BPM in high and moderate risk women, including those with BRCA mutations, is starting to be reported in the literature. Collectively data

suggest that women report satisfaction with their decision to undergo BPM and adjust well emotionally after surgery, although satisfaction with reconstruction may be less optimal (Borgen et al., 1998; Frost et al., 2000; Stefanek et al., 1995). These outcomes must be weighed against the irreversibility of the decision, potential problems with implants and reconstructive surgery, and the occurrence of adverse psychological and social outcomes in some women (Frost et al., 2000).

In Lodder et al.'s (2002) study above it is unclear why most women who underwent BPM (54%) were satisfied with the decision, despite a negative impact on body image, intimate relationship with their partner, and physical well being. Other reports suggest that although BPM may have psychological benefits for some (Hatcher, Fallowfield, & A'Hern, 2001) between 10% and 25% of women who choose BPM report dissatisfaction or an adverse psychological impact (Frost et al., 2000; Payne, Biggs, Tran, Borgen, & Massie, 2000). Although the clinical significance of elevated distress in choices for prophylactic surgery is unclear, it is likely that those who receive positive and VUS BRCA test results could benefit from decision making support.

Socio-economic Factors

In addition to the geographic variation in the United States and Netherlands studies, prophylactic surgery may vary according to culture, healthcare system, insurance coverage, provider attitudes, and other socio-economic factors (Eisinger, Geller, Burke, & Holtzman, 1999). The nature of these socio-economic factors have received little attention in the literature.

An international research study in three genetic clinics located in Montreal (Quebec, Canada), Marseilles (France) and Manchester (Great Britain) by Julian-Reynier

et al. (2001), demonstrated the existence of variations in the acceptability of the preventive strategies in English, French, and Canadian women at risk for HBOC, attending a cancer genetics clinics for the first time. British women were more in favor of BPO and chemoprevention than the French and Canadian women. The British and Canadian women were more in favor of BPM than the French. It was hypothesized that differences in prophylactic surgery by the French women was that they attach more importance to their breasts and ovaries than the British or Canadian women, who have a greater tendency to value life itself most of all (Julian-Reynier et al., 2001).

Bouchard et al. (2004) reported substantial differences in the way cancer geneticists deal with environmental risk factors, breast and ovarian cancer testing, chemoprevention, and prophylactic surgery. This team suggested that attitudes towards preventive measures may be partially explained by cultural factors. Cultural differences included the social representation of risk and health. Physicians from Canada and Anglo Saxon countries promote a model of health that suggests individual responsibility for risk management. In France, more emphasis seems to be put on medical authority in the relationship between providers and patients. In addition, body integrity and the symbolic value of the breasts may carry a different value for French physicians, which may explain differences in acceptability of BPM. Also, interpretations of scientific evidence and medical uncertainties and the impact of leadership, with respect to new ideas and technology innovation, could contribute to the context in which BRCA testing is disseminated in the different countries (Bouchard et al., 2004).

Cost may also be a factor in the higher use of prophylactic surgery in the Netherlands, where universal health coverage is available to women with BRCA

mutations. As health insurance coverage may affect patient decision-making, current information regarding reimbursement practices of third-party payers is needed. A retrospective study by Kauff et al. (2001) of hospital billing records of 38 women with BRCA mutations who underwent either a risk-reducing BPM or BPO revealed that 38 of 39 (97%) prophylactic surgeries were covered in full, minus applicable deductibles or coinsurance. The rate of insurance reimbursement did not vary with type of insurance, personal history of cancer, or type of procedure. Updated studies on the influence of health insurance on BRCA testing and risk management options selected, need to be reported.

Although these first studies to characterize screening and prophylactic surgery behaviors following BRCA testing have been reported, they shed little light on how women interpret and respond to genetic risk information and the factors that influenced their risk management decisions. The reason for the different behavioral responses between the women followed at different research hereditary cancer clinics is unclear. How they arrived at their choices is even less clear. Certainly socio-cultural differences are a factor as suggested above. Furthermore, most studies only provide evidence of the emotional impact of BRCA mutation testing in the short term, which as studies of predictive testing indicate may change over time (Butow et al., 2003).

An understudied area of BRCA research is the family and contextual factors involved in genetic testing and risk management of women with positive and variant of uncertain significance test results. Initial data suggest that genetic testing for breast cancer susceptibility is motivated partly by the desire to help other family members (Geller, Doksum, Bernhardt, & Metz, 1999). Although the majority of genetic testing

participants communicate their risk to other family members, only recently has data been reported on BRCA mutation carriers about the content, process, and impact of these communications (Bonadona et al., 2002; d'Agincourt-Canning, 2001; Hamilton, Bowers, & Williams, 2005; Hughes et al., 2002; Segal et al., 2004; Smith et al., 1999; Tercyak, Peshkin, DeMarco, Brogan, & Lerman, 2002; Wagner Costalas et al., 2003; Wylie et al., 2003). These studies reinforce that the familial context in which genetic testing is conducted may be important for understanding how individuals react to their genetic test results.

Studies on unaffected women who receive BRCA variant of uncertain significance results could not be found, however, two qualitative studies were found in affected women. A qualitative pilot study of 6 affected women with variants of uncertain significance described how these women were unaware their test results could be indeterminant, thus they were unable to process the information in terms of health care decision making (Frost, Venne, Cunningham, & Gerritsen-McKane, 2004). Another study included 12 affected women with “inconclusive” test results, but whether these were noninformative results or inconclusive results from a variant of uncertain significance is not clear. The definition given for the inconclusive result was “a known BRCA1/2 mutation was not identified” (Hallowell et al., 2002, p.79). In the case of these affected women, a noninformative result is one in which no mutation is identified when they are from a family that fits a familial cancer syndrome (Sadler, Wasserman, Fullerton, & Romero, 2004). It is noninformative in that it is not helpful in defining the genetic risk, for the rest of the family. This lends credence to the fact that possible outcomes of genetic testing can be confusing, for all of us, including the patient. There is

a need for more in-depth research that explores unaffected women with BRCA VUSs' experiences of managing their susceptibility to hereditary breast and ovarian cancer, particularly those who have been in receipt of their test result for some time.

Rees, Fry, and Cull (2001) identified that the influence of personal experience of cancer, through involvement with affected relatives, has been neglected in the literature and found strong theoretical grounds for the hypothesis that dimensions of personal experiences may influence response to cancer risk. "Perceptions of breast cancer and beliefs about the disease are likely to influence how an individual reacts (in terms of thoughts, feelings, and decisions) to their own risk status" (Rees, Fry, & Cull, 2001).

As a nurse practitioner with a focus on health promotion entering the field of cancer genetics, what intrigued me from the beginning was an awareness that the knowledge gained by BRCA genetic diagnosis was breaking down the traditional boundaries between prevention and early detection. In cancer susceptibility testing, in order to target disease prevention or health promotion interventions, individuals are being categorized according to genetic risk. These are not like the traditional environmental risks that are external to the individual and over which an individual often has little control, or lifestyle risk factors over which one has some control. This genetic risk resides within one's physical being, putting them in a state between health and illness, their body existing as a constant source of danger. One of the questions that concerned me was what this new technology would do to one's sense of self and social relationships.

Although genetic professionals are concerned that clients accurately evaluate their risk of breast and/or ovarian cancer, from a practical standpoint an individual is likely to evaluate their risk within the context of a broad range of other commitments and concerns

that relate to values, employment, and relations with family and friends. Considering that a woman's view of her future life may be altered by the risk knowledge of a BRCA mutation, the scores on traditional psychometric instruments, as scores on depression scales used in previous research, seemed a minor reflection of the impact of BRCA testing. Thus the influence of family and social contextual factors on women's decision making became the focus of this research. Although my initial plan was to explore unaffected women's risk management decision making after testing with a positive or VUS BRCA mutation, it became obvious after a few interviews that some women came to BRCA testing with plans for prophylactic surgery. To better understand these a priori decisions, the focus changed to explore the broader context of genetic testing in which unaffected women make risk management decisions.

Much of the current research is based on a static view of genetic testing use, largely from the perspective of health care providers and researchers, using standardized instruments. What is lacking are examinations of the total genetic testing experience, including risk management, that focus on the clients' perspective, as well as an understanding of the evolving process as clients move through a trajectory of managing their susceptibility to hereditary breast and ovarian cancer.

Lines of Inquiry

As a result of broadening the study's scope, the purpose of the study was expanded to explore the influence of family and social contextual factors in how unaffected BRCA positive and VUS carriers conceptualized their cancer risk, interpreted and coped with BRCA test information, and made subsequent risk management decisions; and to develop a grounded theory based on the perceptions, beliefs, and actions

of these women. The questions which were addressed included: (a) Given the benefit of hindsight, how would unaffected BRCA positive and VUS carriers describe their experience of discovering their BRCA status? (b) What is the influence of womens' experiences of breast cancer in their family in how they conceptualized their breast and ovarian cancer risk? (c) How do women interpret, assign meaning, and act on the complex information about positive and VUS test results and risk management? (d) What is the decision making process through which they arrive at these actions? and (e) What are the roles their family and others played in their decision making?

As this study was primarily concerned with obtaining the client's perspective, it was important to use a method that enabled clients to express themselves freely and to raise issues of relevance to them. Use of the grounded theory approach allowed the researcher to be guided and directed during the research process by the data, and was suitable for use in this area in which there has been little previous research. It also offered an opportunity to develop a theory for managing susceptibility to hereditary breast and ovarian cancer. This methodological approach has its theoretical underpinnings in symbolic interactionism which brings meaning, identity, and experience of everyday life to explain the social world (Blumer, 1969).

Theoretical Underpinnings of Methodology

Grounded theory method was introduced by Glaser and Strauss in 1967 as a process that provides the discovery of theory from qualitative data (Glaser & Strauss, 1967). Pragmatism and symbolic interactionism are the theoretical bases of grounded theory, and the phenomena of concern are patterns and processes of social units (Strauss & Corbin, 1994).

Symbolic interactionism is an approach to the study of human conduct and human group life, focusing on the meaning of events to people in natural settings. It is concerned with the study of the inner or “experiential” aspects of human behavior, that is, how people define events or reality and how they act in relation to their beliefs (Chenitz & Swanson, 1986).

Symbolic interactionism is strongly influenced by the work of George Herbert Mead (1934), a philosopher and psychologist at the University of Chicago during the first third of the twentieth century. Symbolic interactionism is a perspective in social psychology that is relevant to the concerns of nursing. Charon (1998) identifies five central ideas of symbolic interactionism:

1. Symbolic interactionism focuses on social interaction in its human understanding, rather than on personality characteristics, or how society or social situations cause human behavior. People constantly undergo change in their interactions, and society arises and changes through social interaction. Interaction means that individuals are not only influenced by others, but individuals constantly influence one another. A more active and dynamic human emerges, rather than an individual simply responding to others in the environment.
2. Human action is caused not only by social interaction but also from interaction within oneself. We act based on how we are thinking in the present situation. Although our thinking may be influenced by others with whom we interact, our own thinking always matters.

3. Humans define their situation as they go along rather than sensing the environment directly. As we interact with others and ourselves, we develop our definitions of what is taking place and decide how to act in that situation. We do not respond to reality as it is, but to reality as we define it.
4. Our actions are always caused by what happens in the present, specifically, how we are defining what is happening in the present situation. We are not simply products of our past. It is what is happening right now that matters in what we end up doing. Like everything in the environment, our past is defined by us and it is applied to the situation at hand.
5. Humans are seen as always active and dynamic rather than passive and static, able to take an active part in their own action. The human is 'emergent', always changing as he or she deals with situations encountered.

Mead's work was heavily influenced by pragmatism. Pragmatism is important to symbolic interactionism in its approach to how humans relate to their environment. Pragmatism teaches that we always intervene in determining what 'real' is, that knowledge is believed and remembered because of its usefulness to us, that objects are defined according to their use to us, and humans must be understood primarily by focusing on what they do in real situations (Armour, 1969; Charon, 1998; Kaplan, 1964).

According to Blumer (1969), symbolic interactionism views human behavior as "the result of a vast interpretive process in which people, singly and collectively, guide themselves by defining the objects, events, and situations they encounter" (Blumer, 1969, p. 132). The human being interacts, uses perspectives, defines situations, acts according to the present, and is agentic. The symbolic interactionism perspective conceptualizes the

individual as more complex, contradictory, situational, dynamic, and less predictable and passive than other social-scientific perspectives (Blumer, 1969).

Using a symbolic interactionist perspective, grounded theory provides a way to study human behavior and interaction. It describes an approach to study basic social and psychosocial processes which account for variation in interaction around a phenomenon or problem. Chenitz and Swanson (1986) indicate grounded theory is particularly useful to conceptualize behavior in complex situations, to understand unresolved or emerging social problems, and to understand the impact of new ideologies. It can also form the basis for interventions and social policy related to phenomena studied.

Grounded theory, like other forms of qualitative research, makes its greatest contribution in areas in which little research has been done. In these areas, theory testing cannot be done since the variables relevant to the concepts have not yet been identified (Chenitz & Swanson, 1986). The specific focus of grounded theory on theory generation adds an important dimension to data analysis. This method stresses that theory must come from data, not prior knowledge, and that the operations leading to theoretical concept formation must be revealed (Robrecht, 1995). It does this by interpreting and explaining the perspectives and actions of multiple subjects, by clarifying “patterns of action and interaction between and among various types of social units” (Strauss & Corbin, 1994), p. 278).

In summary, grounded theory offers a systematic method to collect, organize, and analyze data derived from women with positive and VUS BRCA mutation test results. Grounded theory can provide an approach to theory development based on this study of BRCA genetic testing and risk management and the contexts and social forces that

impinge on this process. Such theory can provide a means of conceptualizing the interacting influences of personal characteristics and social processes, as they bear on the decisions of women with this new information.

Significance of the Study

Until recently, genetic testing for BRCA mutations was conducted only in medical research settings. Because of the commercialization of these genetic tests, testing for BRCA mutations are available to most clinicians. Findings from this community study are timely and of importance in the current clinical environment where clients are asking informed questions about HBOC genetic testing.

Beginning knowledge about cancer genetic testing exists in these areas (a) emotional factors which predict use of BRCA genetic testing, (b) psychological outcomes of BRCA genetic testing, (c) family communication about results of BRCA testing, (d) emotional factors which predict use of prophylactic surgery, (e) risk reduction effectiveness of prophylactic surgery, and (f) behavioral outcomes of BRCA testing.

Individuals at increased risk of breast and/or ovarian cancer face uncertainty about if and when cancer will develop and decisions about how to manage their risk. Personal cancer risk estimates are imprecise. We are unable to predict which women receiving genetic test results will have difficulties adjusting to their genetic risk. There are concerns that inaccurate risk perceptions and distress may interfere with recommended risk management for women at increased risk. For those receiving uncertain test results, like a variant of uncertain clinical significance, the way these uncertain results are interpreted and used by clients in their decision making process is unknown. It is imperative that women who have had genetic testing and are currently

managing their susceptibility to HBOC be given a voice. Their experiences have relevance for others seeking predictive cancer testing. No specific study has examined the overall genetic testing experience, including risk management, from the perspective of unaffected positive and VUS mutation carriers.

This study provides insight into the lives of two groups of women and how their personal and family past and present experiences with breast and/or ovarian cancer influenced how they conceptualized their cancer risk, interpreted and coped with BRCA test information, and made subsequent decisions to manage their susceptibility to HBOC. Recent studies have begun to examine the impact of BRCA genetic testing.

This study has added to the increasing body of literature addressing cancer genetic susceptibility testing. Grounding the theory of managing susceptibility to hereditary breast and ovarian cancer in unaffected women with positive and VUS BRCA mutations permits a better understanding of this experience from the perspective of these high risk women. This understanding enables nurses in genetics to plan, implement, and evaluate strategies for nursing interventions, as well as influence social and political policies which affect women seeking to manage their susceptibility to hereditary breast and ovarian cancer. Improved knowledge about risk management decision making should lead to development of decision aids, as well as other informational and emotional support therapies.

Nurses are instrumental in the assessment and identification of women at high risk for breast and ovarian cancer, for whom genetic testing is an option to be considered. They can help individuals make decisions about initiating genetic testing, interpreting the genetic information received, disclosing this information to family, and risk management

options. Nurses have an important role in helping clients by providing informational and emotional support while the client is considering risk management strategies. The nurse is also instrumental in helping clients cope with the consequences of their decisions and ensuring coordinated follow-up. Through nursing advocacy, social policy can be generated for ethical practices to prevent insurance and employment discrimination and for the just distribution of health care dollars so all high risk women have access to this new technology.

To broaden the focus of this study a comprehensive review of the literature on breast and ovarian cancer genetic testing, risk management options and their effectiveness, decision making, and risk management behaviors was undertaken from a variety of perspectives. These perspectives are discussed and synthesized in the following chapter.

CHAPTER II

CONTEXT OF THE INQUIRY

The purpose of this chapter is to provide a background for this study derived from four bodies of literature. First, the current literature on genetic testing for hereditary breast and ovarian cancer (HBOC) with an emphasis on the BRCA1 and BRCA2 mutations are explored. Second, the risk reduction and early detection management options and the effectiveness of these interventions are examined. Third, literature related to decision theory with a review of decision making in a genetics context is analyzed. Fourth, a look at how predictive genetic testing is affecting health behaviors to reduce risk of HBOC is reviewed. The resulting analysis and critique of this literature provides the background and justification for this study.

Genetic Testing for Breast and Ovarian Cancer

One of the major advances in the understanding of breast and ovarian cancer during the last ten years is the recognition that these cancers have a genetic basis. Because genes predispose to cancer, the evolving technology of genetic testing allows presymptomatic testing of persons at high risk of developing cancer. This testing provides information about a person's susceptibility to breast and ovarian cancer.

Breast and Ovarian Cancer Susceptibility Genes

Breast cancer is considered a multifactorial disease caused by genetic and non-genetic factors. Clinicians and clients have long recognized familial patterns of breast cancer. Since December 1990, genetic epidemiology has improved our understanding and ability to assess the risk of women with a family history of breast cancer (Gauthier-Villars et al., 1999). Seven genes that predispose women to breast cancer have been identified: tumor suppressor protein (p53), BRCA 1, BRCA2, and PhosphataseTENsin (PTEN), MSH2, STK11/LKB1 and ATM. Of these, BRCA1 and BRCA2 mutations are the most common of the dominantly inherited genes (Domchek & Garber, 2001). Current research also suggests a possible risk association between breast cancer and a number of common genetic VUSs, which are likely to vary with environmental exposures and other non-genetic risk factors. The contribution of these genetic VUSs to multifactorial cancer risk is yet to be determined (Culver, Hull, Levy-Lahad, Daly, & Burke, 2000).

Localization of the BRCA1 gene on chromosome 17q12-21 in 1990 provided the evidence for transmission of breast and ovarian cancer susceptibility (Narod et al., 1991). BRCA1 is associated with disease in 45% of families with multiple cases of breast cancer and up to 90% of families with both breast and ovarian cancer (Easton, Ford, & Bishop, 1995). BRCA2 was identified in 1994 and localized on the long arm of chromosome 13q12-13. Mutations in BRCA2 account for approximately 35% of multiple-case breast cancer families. These mutations are also associated with male breast cancer, ovarian cancer, prostate cancer, and pancreatic cancer (Gayther et al., 1997; Wooster et al., 1994). These susceptibility genes demonstrate a pattern of autosomal dominant inheritance, with

approximately 50% of susceptible individuals inheriting the predisposing mutation (Noorani & McGahan, 1999).

The discovery of BRCA1 and BRCA2 affords an opportunity for identification of a subset of women at very high risk of developing breast or ovarian cancer (Easton et al., 1995). Five to ten percent of women with breast cancer in the United States have BRCA1 or BRCA2 mutations. This translates to a prevalence of about 1 in 800 among the general population (Amlung, Huelman, & Skinn, 1998; Peto, Easton, Matthews, Ford, & Swerdlow, 1996). Based on pooled data from 22 studies, among first degree relatives of 500 index patients with BRCA mutations, the average cumulative risks of breast and ovarian cancer by age 70 years in BRCA1 carriers were 65% and 39% respectively. The estimates for BRCA2 mutations were 45% and 11% respectively (Antoniou et al., 2003). This is the situation likely to be encountered in clinical genetics situations. BRCA 2 mutations are also associated with a 10% increased lifetime risk of breast cancer in male carriers (Begg, 2002). Higher estimates were reported in the New York Breast Cancer Study of 104 mutation-positive Ashkenazi Jewish women with breast cancer (King et al., 2003). Estimates were based only on relatives whose mutation results were known. These estimates were 69% and 74% for breast cancer by age 70 years for BRCA1 and BRCA2 mutation carriers, respectively, and 46% and 12% for ovarian cancer for BRCA1 and BRCA2, respectively (King et al., 2003).

Family history characteristics associated with an increased likelihood of carrying a BRCA mutation include (a) multiple cases of breast cancer in the family, (b) both breast and ovarian cancer in the family, (c) one or more family members with 2 primary

cancers, and (d) Ashkenazi Jewish background (Couch, 1997; Frank et al., 1998; Shattuck-Eidens et al., 1997).

Hereditary breast and ovarian cancer (HBOC), like that caused by the BRCA mutations, is characterized by early onset of breast cancer (5-15 years earlier than non-hereditary cases), bilateral breast involvement, a history of both breast and ovarian cancer, breast cancer in male family member(s), vertical transmission through both maternal and paternal lines, and familial association with tumors of other organs, especially the ovary and prostate gland (Couch, 1997; Frank et al., 1998; Narod et al., 1991; Parmigiani et al., 1998; Sellers et al., 1994; Shattuck-Eidens et al., 1997).

These characteristics are predicted by the Knudson model, in which individuals with an inherited predisposition carry one mutated allele in the germline and therefore all of their cells. Acquisition of a mutation that inactivates the second copy of the susceptibility gene in a single cell, usually with other genetic changes, results ultimately in cancer. Most cancer susceptibility genes (including BRCA1 and BRCA2) behave as tumor suppressor genes and fit this model (Knudson, 1989).

Hereditary Breast and Ovarian Cancer Risk Assessment

Individuals seeking hereditary breast and ovarian cancer (HBOC) risk assessment are not all looking for the same information. Some are seeking specific information about cancer risk to make decisions about prophylactic surgery and to improve their health behaviors. Others want to learn about risk for their children, while others are looking for reassurance that they have overestimated their risk (Garber, 2000; Jacobsen et al., 1997; Lerman, Lustbader et al., 1995; Lerman, Seay et al., 1995; Struewing et al., 1995). Some women consider the availability of genetic testing an opportunity while others view it as a

threat to self-concept, family relationships, or insurability. Once people understand and assimilate the potential outcomes of testing, fewer undergo testing (Ajzen & Fishbein, 1980; Biesecker et al., 2000). Age (over 40 years) and strong family cohesion have been shown as predictors of BRCA1 and BRCA 2 testing (Biesecker et al., 2000).

Genetic Testing

The sensitivity of tests for detecting BRCA1 and BRCA2 mutations is dependent on the method used for genetic analysis and the prior risk of the person tested (based on the person's cancer history, family history, and ethnic background). Testing an individual affected with breast or ovarian cancer is the most effective way of determining if a BRCA1 or BRCA 2 mutation is the cause of breast and/or ovarian cancer within the family (Culver et al., 2000).

A mutation has been identified in the family. Once a mutation is identified, other family members are tested for the same mutation. In most populations, mutations are rare so that only one mutation will be present in the family. A negative test in this situation is definitive. A family member with a negative result does not share the predisposing family mutation and does not have increased risk of breast or ovarian cancer. Their risk is the same as the general population (Singletary & Robb, 2000).

A mutation has not been identified in the family. A person tested in this situation will have one of three possible results:

1. A genetic mutation is present. A positive result means that a deleterious mutation in the BRCA1 or BRCA2 gene was found. A deleterious mutation is one that causes loss of the gene's tumor-suppressor function and therefore increases the risk of breast and ovarian cancer.

2. A genetic mutation is not present. When a cancer-predisposing BRCA1 or BRCA2 mutation is not identified in a cancer-affected individual in a family with an increased risk of HBOC, negative results are uninformative (Culver et al., 2000). Current BRCA analysis may not detect some types of abnormalities in the BRCA1 and BRCA2 genes. Also, some women who have negative test results may still be at high risk for HBOC by mutations in other genes. This result provides limited information because the family mutation is unknown. This individual could be truly negative, or there could be a mutation in a different gene, or the cancer could have a nonhereditary origin. Thus, when a negative result is obtained, it could be good news fraught with survivor guilt (as is the case of some true negatives for a familial mutation) or a negative result could be ambiguous and raise more questions than answers (Peshkin, DeMarco, Brogan, Lerman, & Isaacs, 2001). Studies have demonstrated that 16% to 66% of high-risk families do not carry detectable mutations in BRCA1 or BRCA 2 (Ford, Easton, Stratton, Narod, Goldgar, Devilee et al., 1998; Frank et al., 1998). It is important that these individuals understand that a negative test is not reassurance that breast or ovarian cancer will never develop.

3. A genetic test result is inconclusive. Since BRCA1 and BRCA 2 are large genes, the laboratory may uncover a new sequence alteration, generally involving a single nucleotide change in the gene, which may or may not disrupt the function of the protein. There are no validated functional assays to evaluate this further. The individual then must be told that the result could be either a functional mutation or a polymorphism of no clinical significance. About 13% of BRCA analysis results are reported as genetic variants of uncertain significance (Frank et al., 2002). With additional research, it may be

possible to define the majority of these VUSs, but this may require years (Singletary & Robb, 2000).

When a genetic mutation cannot be ruled out, an individual may still be at increased risk for cancer and therefore need to obtain appropriate surveillance (Peshkin et al., 2001). Although case studies have been reported, studies on unaffected women with a BRCA VUS and their genetic testing and risk management experiences could not be found. However, two studies were found in women affected with breast cancer. A qualitative pilot study of 6 affected women with VUSs described how these women were unaware their test results could be indeterminant, thus they were unable to process the information in terms of health care decision making (Frost et al., 2004). Another study included 12 affected women with “inconclusive” test results, but whether these were uninformative results or inconclusive results from a VUS is not clear. The definition given for the inconclusive result was “a known BRCA1/2 mutation was not identified” (Hallowell et al., 2002, p.79). In the case of these affected women, a uninformative result is one in which no mutation is identified when they have a family history that fits a familial cancer syndrome (Sadler et al., 2004). It is uninformative in that it is not helpful in defining the genetic risk, for the rest of the family. This lends credence to the fact that possible outcomes of genetic testing can be confusing, for all of us, including the patient. There is a need for more in-depth research that explores unaffected women with BRCA VUSs’ experiences of managing their susceptibility to hereditary breast and ovarian cancer, particularly those who have been in receipt of their test result for some time.

Options for Risk Reduction and Early Detection

The available management options for women with BRCA1 or BRCA2 mutations are limited. These women must make a decision with a primary focus on either HBOC risk reduction (prophylactic surgeries or chemoprevention) or early detection (intensive surveillance) (Burke et al., 1997). Minimal data exist on the outcomes of interventions to reduce risk in clients with a genetic susceptibility to breast or ovarian cancer. However, prospective research results for 1-8.8 years post BRCA testing, in affected and unaffected BRCA mutation carriers, are starting to be reported. Retrospective and prospective evidence for BPM in women with BRCA mutations has been reported for 13.4 years and 5.5 years respectively. Due to short term and limited evidence, the clients' preferences, therefore, are important factors in their risk management decisions.

In 1997 the Cancer Genetics Studies Consortium, a task force organized by the National Human Genome Research Institute, made recommendations for cancer surveillance and risk reduction for individuals carrying mutations in BRCA1 or BRCA2 genes. Based on the group's expert opinion and observational studies, early breast and ovarian cancer screening are recommended for individuals with BRCA1 mutations and early breast cancer screening for those with BRCA2 mutations (Burke et al., 1997). These recommendations are in need of updating (Domchek & Garber, 2001).

Screening

Current screening recommendations for HBOC include monthly breast self examination by 18-21 years of age, semi-annual clinical breast examination, and annual mammography beginning between the age of 25 and 35 years for carriers (Burke et al., 1997). For ovarian cancer screening, transvaginal ultrasound and CA 125 are

recommended annually or semi-annually, beginning at age 25-35 years (Burke et al., 1997). The following review explores the effectiveness of these modalities for women with HBOC.

Breast Cancer Surveillance

Brekelmans et al. (2001) reported the results from a HBOC surveillance program that included monthly breast self-examination (BSE), semi-annual clinical breast examination (CBE), and yearly mammography for a median follow-up of three years. In 118 BRCA1 and BRCA2 carriers, 9 cases of breast cancer were diagnosed, 5 of which were diagnosed in the screening program (this is a 56% sensitivity). However, 4 of the 9 tumors in mutation carriers were interval cancers, not detected in the course of screening. An interval malignancy is one that becomes evident between annual screening mammography, which indicates the malignancy either went undetected by the last mammogram or developed during the interval since the last screen (Komenaka et al., 2004). In addition, there were more lymph node positive tumors in the group of known mutation carriers than in the high-risk or moderate-risk groups (56% versus 33% versus 25%). Therefore, in BRCA1 and BRCA2 mutation carriers, this type of surveillance may have a lower sensitivity and also might result in detection of later-stage tumors. Brekelman et al. concluded that to reduce breast cancer mortality a substantial proportion of small cancers have to be detected in the BRCA1 and BRCA2 group. He indicated that a more intensive screening program might be warranted. Two early-detection options are in the process of being evaluated, digital mammography and magnetic resonance imaging (Lewin et al., 2001; Tilanus-Linthorst et al., 2000).

A similar surveillance sensitivity of 50% was reported by Scheuer et al. (2002), who followed 165 male and female BRCA mutation carriers in a surveillance program that included annual mammography, monthly BSE, and CBE 2 to 4 times annually. After 24.8 months of follow-up, breast cancer was diagnosed in 12 women: 6 tumors were diagnosed by mammography and 6 were interval cancers.

These studies indicate that cancers in BRCA mutation carriers grow rapidly; half appear in the interval between annual mammograms, and half have spread to axillary lymph nodes by the time they are detected. Komenaka et al. (2004) suggest that strong consideration should be given to screening BRCA positive women at more frequent intervals and to using additional imaging techniques, such as breast ultrasonography and/or breast magnetic resonance imaging (MRI), as a part of this screening.

Ziv, Shepherd, Smith-Bindman, & Kerlikowske (2003) postulated that the lower sensitivity of mammography in BRCA mutation carriers may result from higher breast density in women with a family history of breast cancer. Tilanus-Linthorst et al. (2002) suggested that a decreased probability of detection by mammography was due to the morphological features of BRCA related tumors (less spiculated masses due to lack of tumor surrounding fibrosis).

Recent evidence indicates that magnetic resonance imaging (MRI) offers better sensitivity than mammography for the early detection of breast cancer in BRCA mutation carriers, however specificity is generally lower (Kriege et al., 2004; Kuhl et al., 2000; Stoutjesdijk et al., 2001; Tilanus-Linthorst et al., 2002; Warner et al., 2001). Magnetic resonance imaging of the breast provides information about breast tissue vascularity that is not available from mammography (Lieberman, 2004).

In the largest prospective study comparing mammography, CBE, and MRI, Kriege et al. (2004) examined 1909 women who had a genetic or familial predisposition to breast cancer. Of these women, 358 had BRCA mutations, twice as many as in all previously published evaluations of MRI in high risk women. In this analysis, MRI, compared to mammography, had higher sensitivity (71% versus 40%) but lower specificity (90% versus 95%). Of the 45 breast cancers found, 22 (49%) were identified by MRI but not mammography, 10 (22%) were identified by both modalities, and 8 (18%) were identified by mammography, but not MRI. In other studies, MRI sensitivity was reported at 86% to 100%, and specificity ranged from 91% to 99% (Kuhl et al., 2000; Podo et al., 2002; Warner et al., 2001). In studies by Kuhl et al., Podo et al., Warner et al., and Stoutjesdijk et al. (2001) mammography was shown to have a sensitivity in the range of 13% to 43% and a specificity range of 93% to 100%. Although breast MRI is highly sensitive, its disadvantages include cost (\$700 to \$1000, about 10 times the cost of a mammogram), variation in enhancement during the menstrual cycle (midcycle is optimal), and imperfect specificity (Lieberman, 2004).

None of the surveillance studies among BRCA mutation carriers have addressed the effectiveness of screening methods in terms of outcomes, such as breast cancer mortality or quality of life. In addition, most studies did not delineate whether tests were done in healthy (unaffected) women or were part of a work up for breast cancer (Lieberman, 2004).

Ovarian Cancer Surveillance

Screening and early detection of ovarian cancer are more difficult than for breast cancer. The majority of ovarian tumors are diagnosed at stage III and IV disease (about

70%), and mortality for these high stage cancers is high (Greenlee, Hill-Harmon, Murray, & Thun, 2001; Rebbeck et al., 2002). Clinical recommendations are even more limited in ovarian cancer screening. Neither transvaginal ultrasound (TVUS) nor serum tumor marker, cancer antigen 125 (CA-125) measurement, have been shown to reduce morbidity or mortality from ovarian cancer, and the efficacy of these approaches have not been reported in women with BRCA 1 and BRCA2 mutations (Cherry & Vacchiano, 2002; Offit, 1998).

CA-125 testing. The value and limitations of CA-125 were reported in a study of 5550 women by Einhorn et al. (1992) who found a false positive rate of 96.6%. Also concerning, was the fact that three women who tested negative subsequently proved to have ovarian cancer. A similar lack of specificity was reported by Troiano, Quedans-Case, and Taylor (1997) and DePriest, Gallion, Pavlik, Kryscio, and Van Nagell (1997). DePriest et al. made screening available to 6470 women who were either postmenopausal or greater than 30 years old with a family history of ovarian cancer. A total of 90 participants underwent surgery which showed 37 serous cystadenomas and 6 primary ovarian cancers, 5 which were stage 1A. Only one cancer was detected by pelvic examination, and none had an elevated serum CA 125, further demonstrating the limitation of these methods. In contrast, Jacobs et al. (1996) measured CA 125 levels annually in 22,000 women over 45 years old. They reported the serum CA 125 level correlated well with the cumulative risk of developing ovarian cancer.

Zurawski, Orjaseter, Andersen, and Jellum (1988) found overall that the CA 125 antigen test lacks sensitivity in stage 1 ovarian cancers where it is elevated in only 50% of cases. It also lacks specificity because several benign conditions elevate CA-125

levels, such as fibroids, endometriosis, pregnancy, and liver disease (Cherry & Vacchiano, 2002; Zurawski, Knapp et al., 1988; Zurawski, Orjaseter, Andersen, & Jellum, 1988)

Transvaginal ultrasound (TVUS). There is limited data regarding the potential benefit of transvaginal ultrasound in screening women at inherited risk of ovarian cancer. Limitations of TVUS include a lack of specificity and inability to detect primary peritoneal cancer or cancer in normal-size ovaries (Tailor et al., 2003; van Nagell et al., 2000). Peters and Stopfer (1996) reported a study in which 1061 women with a family history of ovarian cancer were screened with pelvic ultrasound. Only 3 of 61 women with abnormal results had ovarian cancer, 2 with stage I and 1 with stage III. Tailor et al. (2003) reported a 10 year observational study of 2500 asymptomatic women, with at least one affected relative with ovarian cancer, in which ultrasound test sensitivity was 92% and specificity 97.8%. There were 11 screening detected cancers, 1 false negative, and 93 false positives.

Currently underway in the United Kingdom is a phase II trial evaluating the effectiveness of ovarian cancer screening using annual TVUS and serum CA 125 levels. Also in the United States, the National Cancer Institute is conducting a controlled clinical trial in which 74,000 women are randomized to regular medical care or research-based screening for ovarian and other cancers. Because the efficacy of the ovarian cancer screening approaches are not known, high risk women are often advised to undergo bilateral prophylactic oophorectomy when childbearing is complete (Rebbeck, 2000).

Chemoprevention

The use of chemoprevention in BRCA mutation carriers is in its infancy. To reduce morbidity and mortality, a chemopreventive agent must arrest carcinogenesis before the emergence of an invasive or clinically detectable cancer (Sporn, 1993). The only Food and Drug Administration (FDA) approved risk reduction agent for women at high risk of breast cancer is tamoxifen (Stefanek et al., 2001). Tamoxifen is an estrogen receptor (ER) antagonist. It is thought to be effective because the risk of breast cancer is related to levels of endogenous and exogenous hormones (Cauley et al., 1999; Henderson, Ross, & Bernstein, 1988; Schairer et al., 2000).

In the Breast Cancer Prevention Trial (BCPT), tamoxifen administered to healthy high risk women for 5 years was shown to reduce the risk of invasive breast cancer by 49% and non-invasive breast cancer by 50% (Fisher et al., 1998). Reduction in breast cancer risk was noted among women with a family history of breast cancer, as well as those without a family history. In this randomized controlled trial of 288 incident breast cancer cases, only 19 were BRCA mutation cases (8 BRCA1 and 11 BRCA2). A higher proportion of BRCA 2 patients were ER positive compared to BRCA1 patients, which may explain the benefit from tamoxifen. Although there was a reduced incidence of 69% in estrogen receptor-positive tumors, there was no difference between the treatment and placebo group in the occurrence of ER negative tumors. The benefits of tamoxifen were countered with an increased incidence of adverse events of endometrial cancer and vascular events (stroke, pulmonary embolus, and deep venous thrombosis) and cataracts among women over 50 years. Due to the small sample size, the study did not reach

statistical significance and thus only suggests the preventive role of tamoxifen in BRCA2 carriers (Fisher et al., 1998).

In a matched case-control study, Narod et al. (2000) investigated the preventive effect of tamoxifen in BRCA carriers, comparing 209 participants with bilateral breast cancer with 384 mutation carriers with unilateral disease. Tamoxifen was associated with a 50% reduction in contralateral breast cancer, with greatest reduction following 2-4 years of use. BRCA1 mutation carriers had greater protective effects of tamoxifen in this study. Two biases may have occurred in these results. Because this study was restricted to living women, selection bias may have occurred if those who died were different from the study population in terms of tamoxifen use and contralateral breast cancer outcome. Also information bias may have occurred, as questionnaires were completed on average 11.8 years after diagnosis (Calderon-Margalit & Paltiel, 2004).

These contradictory results do not provide conclusive evidence of efficacy of tamoxifen. Other small studies have suggested tamoxifen may have some efficacy in BRCA1 carriers, despite estrogen receptor status (Daidone et al., 2002; Eisinger et al., 2001; Foulkes et al., 2002). Because tamoxifen appears to be most effective at preventing breast cancers with estrogen receptors, and because 70% to 80% of BRCA1 associated breast cancer lack estrogen receptors, its effectiveness is unknown (Johannsson, Idvall, & Anderson, 1997; Lippman & Brown, 1999). Chemoprevention in BRCA2 carriers seems more plausible than in BRCA1 carriers, since BRCA1 carriers are more likely to be estrogen receptor negative. In fact, in a pilot survey of physicians Peshkin, Isaacs, Finch, Kent and Schwartz (2003), found they were more likely to recommend tamoxifen to a BRCA2 carrier than to a BRCA1 carrier (73% versus 57%).

Another estrogen-receptor modulator that acts as both an agonist and antagonist to estrogen, raloxifene, holds promise as an agent for breast cancer risk reduction. The Study of Tamoxifen and Raloxifene for the Prevention of Breast Cancer (STAR), a randomized double-blind trial, will help determine whether raloxifene is more or less effective than tamoxifen in reducing the incidence of invasive breast cancer in postmenopausal women (Brown & Lippman, 2000).

Prophylactic Surgery

Several studies have evaluated bilateral prophylactic mastectomy (BPM) in women with BRCA mutations. Retrospective and prospective studies show a high degree of risk reduction with this procedure.

In a subset analysis of Hartmann et al. (1999) retrospective cohort study of 214 high risk women who underwent BPM, BRCA mutations were found in 26 women (18 BRCA positive and 8 variants of uncertain significance). None of these 26 women had developed breast cancer at median follow-up of 13.4 years (Hartmann et al., 2001).

Hartmann's results have been supported by Meijers-Heijber et al. (2001) in a prospective study from the Netherlands. Seventy-six unaffected BRCA1 and BRCA2 mutation carriers were followed for three years after BPM and 63 mutation carriers had regular surveillance follow-up. Although six cases of breast cancers would have been expected in the BPM group had the procedure not been done, none were reported in the BRCA carriers. However, 8 breast cancers were identified in women using regular surveillance. Fifty eight percent of the BPM group had also undergone bilateral prophylactic oophorectomy (BPO).

In a recent study in medical centers in the Netherlands, North America, and the United Kingdom, the efficacy of BPM for risk reduction in women with BRCA mutations was analyzed (Rebbeck et al., 2004). This Prevention and Observation of Surgical End Points (PROSE) study compared breast cancer in 105 unaffected BRCA mutation carriers, who underwent BPM, with 378 unaffected mutation carriers who did not choose BPM. Breast cancer was diagnosed in two (1.9%) women who had BPM and in 184 (48.7%) of matched controls who did not have BPM, during a mean follow-up of 6.4 years. The two breast cancers in women with BPM occurred in women with subcutaneous mastectomies. Subcutaneous BPM leaves substantial breast tissue intact, including the nipple-areolar complex. Total BPM removes substantially more breast tissue and thus requires more extensive reconstruction. Bilateral prophylactic mastectomy reduced the risk of breast cancer in BRCA mutation carriers by approximately 90% in women with intact ovaries and by approximately 95% in women with prior or concurrent BPO. This data suggests that BPM may significantly reduce the risk of breast cancer for women with BRCA1 and BRCA2 mutations.

Bilateral Prophylactic Oophorectomy (BPO)

Although the Cancer Genetics Studies Consortium concluded there was insufficient evidence to recommend BPO for reducing ovarian cancer risk, the NIH Consensus statement on Ovarian Cancer recommended that women at inherited risk for ovarian cancer undergo BPO after child-bearing or age 35 years (Burke et al., 1997; NIH, 1995). Since publishing of these statements, two recent studies among BRCA carriers have demonstrated its effectiveness.

Rebbeck and colleagues (2002) reported a multicenter, case control study (n=259 with BPO and n=292 controls without BPO) in which BPO reduced the risk of ovarian cancer or papillary serous peritoneal cancer associated with BRCA1 or BRCA2 mutations by 96%, and the risk of breast cancer by 53%. The average length of follow-up was 8.2 years for those undergoing surgery and mean age at surgery was 40.9 years. However, papillary serous peritoneal cancer did occur in eight (3.1%) women who had undergone BPO. Six of these cancers were stage I and diagnosed at the time of BPO. After a mean follow-up of 8.8 years, 58 ovarian cancers (19.9%) were found among the BRCA controls. None of 124 BRCA carriers who had BPO by age 35 years had developed cancer, suggesting that timing of BPO may be important. This study provides support for significant ovarian cancer risk reduction with BPO.

Kauff et al. (2002) in a prospective study, compared the effect of risk-reducing salpingo-oophorectomy (RRSO) with that of surveillance for ovarian cancer, on the incidence of subsequent breast and BRCA-related gynecologic cancers, in BRCA mutation carriers 35 years of age or older. During a mean follow-up of 24.2 months, breast cancer was diagnosed in 3 of 98 (3.1%) women who chose bilateral RRSO and peritoneal cancer was diagnosed in 1 (1%). In the 72 women that chose surveillance for ovarian cancer, breast cancer was diagnosed in 8 (11%) women, ovarian cancer in 4 (5.5%), and peritoneal cancer in 1 (1.4%). The 5 year cancer free survival estimates for unaffected BRCA mutation carriers who had RRSO was 94% compared to 69% for mutation carriers that chose surveillance. The complication rate was minimal, as only 4 of 98 (4.1%) women had any surgical complications. Thus, BPO can reduce the risk of breast and ovarian cancer, but cannot confer complete ovarian cancer prevention.

Although BPO is an option offered to women at high risk for ovarian cancer, it is unclear to what extent this surgery is discussed with women in the context of breast cancer risk reduction. A BRCA1 and BRCA2 mutation carrier may choose this surgery alone because BPO may substantially reduce breast cancer risk, without incurring the possible impact of BPM on body image. However, this surgery has to be weighed against the consequences of surgical menopause after BPO, with all its associated sequelae. Unresolved issues include: (a) the best type of oophorectomy; salpingo-oophorectomy, or BPO, or BPO with removal of the uterus, and (b) use of hormone replacement therapy after BPO, to ameliorate the symptoms of menopause.

The above studies showing the efficacy of prophylactic surgeries lends evidence of the need for revision of the Cancer Genetics Studies Consortium recommendations for women who are BRCA mutation carriers. In light of the unfolding evidence on efficacy of prophylactic surgical treatment options, how do women with BRCA1 and BRCA2 mutations sort out the information and make decisions about risk management? The next section will review the literature on decision theory and decision making in a genetic context.

Decision Theory

Both methods and theories in modern studies of decision making are highly diverse. Philosophers, psychologists, economists, and mathematicians all have different ideas about what decision and choice are, and about how they are to be understood and incorporated into a larger theoretical context (Rachlin, 1989). Decision research has traditionally explored two questions. The first is normative: how can decisions best be made? The second is descriptive: how are decisions actually made (Baron, 1994;

Chapman & Sonnenberg, 2000)? These broad types of decision research provide the framework for this decision theory literature review.

Normative Theories

Normative models of decision making describe what people ought to do if they wish to be rational decision makers (von Neumann & Morgenstern, 1947). These rational theories of choice (e.g., expected utility theory, multiattribute utility theory, probability theory) are based on mathematical and statistical proofs and propose that decision makers follow a highly rational procedure for making decisions (Bekker et al., 1999). They assume decision processes that are consequential and preference-based. They are consequential in that alternatives are interpreted in terms of their expected consequences. They are preference-based in that consequences are evaluated in terms of personal preferences. March (1994) described rational choice as based on the answers to four basic questions: (a) Alternatives: what actions are possible? (b) Expectations: what future consequences might follow from each alternative? How likely is each possible consequence, assuming that alternative is chosen? (c) Preferences: how valuable (to the decision maker) are the consequences associated with each alternative? and (d) Decision rule: how is a choice to be made among the alternatives in terms of the value of their consequences?

Classical decision theory assumes that people have consistent preferences, know their preferences, are familiar with all the alternatives that are available to them, have information about the consequences of the alternatives, and combine the information according to the expected utility rule, which weights outcomes by their probability of occurrence (Carroll & Johnson, 1990). The rational or best course of action is the one that

has the highest expected utility (personal value) (Bekker et al., 1999; Hastie & Dawes, 2001).

Although these models are logical and appealing, research data show that actual decisions do not always follow the rational model (Kahneman, Slovic, & Tversky, 1982). Rational choice theories have adapted to such observations by introducing the idea that rationality is limited. Although decision makers try to be rational, they are constrained by limited cognitive capability and incomplete information, and thus their actions may be less than rational despite their best intentions (Hastie & Dawes, 2001; Kahneman et al., 1982).

Descriptive Theories

Descriptive theories, in contrast to normative theory, describe how people actually make decisions, not how they ought to decide. In cases of uncertain information, both rational and irrational mechanisms influence judgments and behavior (Kahneman et al., 1982). Cognitive psychology research over the last forty years has shown that decision makers have biological limitations on information processing, in their attention, memory, comprehension, and communication (Carroll & Johnson, 1990; Newell & Simon, 1972; Simon, 1955). Instead of considering all alternatives, decision makers appear to consider only a few and to look at them sequentially rather than simultaneously (Simon, 1982). They do not consider all consequences of their alternatives. They focus on some and ignore others. Instead of a complete, consistent set of preferences, decision makers seem to have incomplete and inconsistent goals, not all of which are considered at the same time (March, 1994). The first alternative that meets or exceeds a specific aspiration level

is selected. This “satisficing” helps to terminate the search for alternatives and speeds the decision making process (Simon, 1982).

The effects of these limitations on judgment and decision making are important. Since we cannot process large amounts of information at one time, we tend to simplify situations, to formulate decisions through limited viewpoints that highlight some aspects of the situation but ignore others. We have developed a variety of logical shortcuts, rules of thumb, or heuristics for making good decisions with our limitations (Kahneman et al., 1982; Simon, 1955). These procedures form the core of theories of limited rationality.

Limited Rationality

Decision makers use various information and decision strategies to cope with their limitations in information and information-handling capabilities. Psychological studies of individual decision making have identified numerous ways in which they react to cognitive constraints (Kahneman et al., 1982). The present intention is to characterize only a few of these principal speculations developed as a result of psychological studies of individual information processing. Two of the fundamental simplification processes are framing and heuristics.

Framing. Framing involves how you see the problem. People respond to situations as they interpret them, not as they exist in some objective reality. Tversky and Kahneman (1981) discuss the ‘frame’ that people use to identify decision problems and their components. Decision makers adopt paradigms to tell themselves what perspective to take on a problem, what questions should be asked, and what technologies should be used to ask the questions. These frames focus attention, simplify analysis, and direct attention to different options and preferences. A decision will be made in one way if it is

framed as a gain (e.g., survival rates) and another if as a loss (e.g., mortality rates) (Hastie & Dawes, 2001). Decision makers typically frame problems narrowly, rather than broadly. Many times they are content to find a set of sufficient conditions for solving a problem, not the most efficient set of conditions (March, 1994).

Heuristics. Decision makers recognize patterns in the situations or problems they face and apply rules-of-thumb to solve the problem or make the decision. These heuristics reduce complex mental processes to simpler cognitive tasks saving time and cognitive resources (Kahneman et al., 1982). An example in consumer purchases might be ‘judge quality by price’. Tversky and Kahneman’s research in heuristics showed that people’s reasoning is influenced by prior experiences with similar events. This ‘representativeness’ heuristic involves the recognition of patterns in the situation and application of rules of appropriate behavior to those situations. Characteristics of the representativeness heuristic have been strongly reflected in descriptions and discussion of intuitive judgments in nursing practice (Cioffi, 1997). Benner and Tanner’s (1987) descriptive study of the use of intuition by expert nurses discussed ‘similarity recognition’ as recognition of ‘fuzzy’ resemblances despite marked differences in the objective features of past and current situations. Studies of expertise generally reveal that experts substitute recognition of familiar situations and rule-following for calculation (March, 1994).

A classic form of heuristic is ‘availability’, which involves the assessment of the probability of an event based on the ease with which instances come to mind (Friedlander & Stockman, 1983). This retrieval and construction of similar instances has been shown

to be affected by recency (Slovic, Fischhoff, & Lichtenstein, 1982), vividness (Nisbett & Ross, 1980), and salience (Arkin & Duval, 1975).

Affective reactions are also a means to facilitate information processing and decision making. The 'affect heuristic' represents the contribution of feelings in decision making and refers to the "goodness" or "badness" of feelings experienced (Slovic, Finucane, Peters, & MacGregor, 2004, p.312). It is a conscious or unconscious feeling state (e.g. fear, relief) that the person experiences while making a decision. Zajonc (1980) argued that affective reactions to stimuli occur automatically and subsequently guide information processing and judgment. Thus they may serve as orienting mechanisms, helping to quickly and efficiently evaluate complex and uncertain situations.

A key feature of heuristics is they usually do a good job, but not necessarily the best job given the information at hand (and they sometimes do poorly). Katapodi, Facione, Humphreys, and Dodd (2005) have identified heuristics that create biases in perceived breast cancer risk. Heuristics are also easier to use than sophisticated decision rules, such as those proposed by economists and management scientists (Kahneman et al., 1982).

These existing decision theories indicate that decision making is a highly complex concept. This body of knowledge and related analytical techniques of different degrees of formality are designed to help a decision maker choose among a set of alternatives in light of their possible consequences (Web Dictionary of Cybernetics and Systems, 2002). Most of the research in nursing science is grounded in either normative (analytical) decision making theory or in information processing theory.

Most nursing research has been done on professional decision making in clinical settings (Benner, 1984; Benner, Tanner, & Chesla, 1996; Hannah, Reimer, Mill, & Letourneau, 1987; Lauri & Salantera, 1998; Luker, Hogg, Austin, Ferguson, & Smith, 1998; Narayan & Corcoran-Perry, 1997; Tanner, 1986; Watkins, 1998). In this research, expert practitioners are able to make accurate decisions about clinical problems based on accumulated knowledge of similar cases, and patients and novice practitioners do not have the same knowledge and experience to readily determine what is happening and action required.

Although researchers in other disciplines have made significant contributions to building decision science over the last 40 years, only a few nurse researchers have focused their research in patient decision making (Degner et al., 1997; Degner, Davison, Sloan, & Mueller, 1998; Hollen, 1994; Kelly-Powell, 1997; Llewellyn-Thomas et al., 1991; O'Connor et al., 2002; Pierce, 1993; Pierce & Hicks, 2001; Pierce, 1996). This research has not been focused on women with the BRCA susceptibility genes. The degree to which findings associated with these patient and professional clinical reasoning are applicable to the context of patient genetic decision making is unknown. A better understanding of the family and social context in which patient genetic testing and risk management decision making takes place can add to this body of knowledge.

Decision Making in a Genetics Context

Difference in the Nature of Genetic Disease

Genetic disease does not fit the traditional medical model of disease. In this traditional model, disease was an 'individualized' phenomenon, the patient was an individual with a disease who was receiving treatment. Genetic problems may be shared

with family members and future offspring and the genetic treatment(s) may have benefits or liabilities for others (Hamilton et al., 2005; Kegley, 2000).

Further, genetic testing may reveal the gene carrier, which may lead to some guilt and negative feelings. In fact, various studies have revealed feelings of guilt and a sense of defectiveness among people tested with genetic disease (Dorval, Paternade et al., 2000). Mutation carriers who passed on the genetic susceptibility to their child may suffer from transmitter guilt, and those who have not inherited the genetic mutation may experience survivor guilt (Offit, 1998; Terdiman, Conrad, & Sleisenger, 1999). This guilt phenomenon is less true in the traditional biomedical model, as disease is seen as something which happens to someone, and for which there is usually no personal responsibility. Genetic disease, unlike other disease, is seen as closely tied to personal identity and personal destiny. This identification of genes with self, not only promotes the guilt phenomenon, but also brings forth concerns about autonomy and decisional privacy.

In addition, most genetic diseases have an open-ended quality. That is, genetic factors interact among themselves in complex ways and these genetic factors interact in multiple ways with environmental factors. This makes the prognosis of genetic disease problematic. Lappe (1987) states this poetically, "Genes are like the dots on a pointillist painting: without their context and interaction with other genes, the picture remains an abstraction" (p. 5). Holtzman (1989) argues that in dealing with genetic diseases the doctrine of specific etiologies needs to be replaced with three categories of etiologies: genetic, environmental, and modulating (i.e. age, family, race, climate, stress).

A final difference with genetic disease, such as with BRCA mutations, is the fact that the person involved may be asymptomatic, they have a genetic defect, but may not

now nor may ever have, the disease in the future. Disease is not only a pathologic entity, it is a social construct, almost always accompanied by a change in the ontologic status of the ill person (Rollin, 1979). The change which occurs when the symptom experienced is labeled as a disease, is likely to happen now on the basis of genetic information. An asymptomatic person may be given the social status of 'sick', which may lead to self-image or identity problems, loss of employment, and insurance. Thus social contexts and actions may impact heavily on this person, their autonomy, and privacy (Surbone, 2001, 2004).

Decision Making In A BRCA Context

BRCA1 and BRCA2 mutation testing is considered predictive testing. Specifically, genetic testing is obtained by affected and unaffected individuals to predict future risk of breast and ovarian cancer. The hope with this type of testing is that early identification of clients at risk for breast and ovarian cancer will lead to reduced morbidity and mortality through risk identification, targeted surveillance, and risk reduction (Evans, Skrzynia, & Burke, 2001). It is hoped that by informing people of their genetic susceptibility to disease, they will be motivated to reduce their risks.

What is decided? BRCA1 and BRCA 2 mutation testing and presymptomatic diagnosis have brought such questions as (a) should I do the test? (b) at what age? and (c) what protective or preparative measures should I take if I am at increased risk (Shiloh, 1996)? Although the cumulative lifetime risk is high (45%-65% for breast cancer and 11% to 39% risk for ovarian cancer with BRCA1 and BRCA2 mutations, respectively), there is always a substantial component of uncertainty (Antoniou et al., 2003; Easton et al., 1995; Ford, Easton, Stratton, Narod, Goldgar, & Devilee, 1998; Struewing et al.,

1997). Not only is there uncertainty about whether a breast and/or ovarian cancer will develop, but also when it may appear and how severe it will be. As noted previously, there is further uncertainty with the risk management options available.

In BRCA related decisions, options are risky, as any choice involves negative aspects and may result in avoidance-avoidance conflict. Some decisions are of the risk-versus-risk type in which one decides between two risks: risk of breast cancer versus risk of the side effects of a chemoprevention agent (tamoxifen) which increases the risk of endometrial cancer. Other decisions belong to the difficult category of risk-versus-cost, where the client chooses between the risk of ovarian cancer and not having children, as the result of a BPO (Lave, 1987).

Outcomes from these decisions are frequently multidimensional, involving potential consequences at many levels (e.g. health, emotional well-being, or insurance loss). Many times the decisions are linked sequentially, like deciding to get BRCA genetic testing and then deciding what to do with the results. Decisions involve other members of the family and future generations and are related to personal values about self and family. These factors have the potential to create decisional conflict (Guerriere & Llewellyn-Thomas, 2001; Janis & Mann, 1977; O'Connor, 1995; Shiloh, 1996).

Who makes the decision? In the context of genetic risk, who makes the decision is not as straightforward as it may seem. Traditionally, genetic counseling has been non-directive, with the provider refraining from recommending which option should be chosen, and leaving the responsibility with the client. In part, this is a result of specific sociological movements, like individualism and feminism that have influenced client-provider relationships (Gortner, 1990; Zussman, 1997). This approach, based on values

of autonomy and the legal imperative of informed consent, is not always welcomed. Lippman-Hand and Fraser (1979) described frequent requests for guidance by clients. Similarly Karp (1983) described the frequently asked question, "What would you do in my place?" Denying clients' request for advice may impede the counseling relationship or be interpreted as lack of care. Clients may also interpret this as indicating the counselor's judgment about the severity of the information provided (Shiloh & Saxe, 1989). Lippman and Wilfond (1992) wrote that "no single story, however balanced, can ever be neutral or value free" (p. 936-937). Additional studies showed that different ways of presenting genetic risks (e.g. percentages-odds, words-numbers, positive-negative presentations) result in differing perceptions and choice of options by clients (Kessler & Levine, 1987; Marteau, 1989; Robinson, Bender, & Linden, 1989; Shiloh & Sagi, 1989). Robinson, Bender, and Linden (1989) showed that in prenatal genetic counseling, clients' decisions depended on who provided the counseling.

Patients' involvement in healthcare decisions appears to be related to the way patients approach the decision problem and the amount of control they prefer in making decisions. In addition, individuals seem to have certain styles for approaching decision problems. These styles influence how they structure the decision problem, gather information, and determine their level of involvement (Pierce, 1993; Pierce & Hicks, 2001). Decision styles range from avoidance to engagement in the decision problem. These styles are dynamically influenced by: "deferring responsibility, avoidance, information seeking, and deliberation" (Pierce, 1993; Pierce & Hicks, 2001 p. 270; Pierce, 1996).

Degner (1998) indicates that the degree of control desired by an individual in a decision can be classified as passive, collaborative, or active. Level of control desired appears to vary according to several factors. Passivity is encouraged by the novelty of the disease and its severity (Davidson, Degner, & Morgan, 1995; Degner & Sloan, 1992). Younger, well-educated women tend to prefer a more active role in decision making (Benbassat, Pilpel, & Tidhar, 1998).

An important aspect of the decision making process is the healthcare provider and client interaction. The exchange of information between client and provider in the decision context has been well studied, including the client preferences for type and amount of information (Benbassat et al., 1998). Clients preferred information on the course of their disease, available treatment options, and likelihood of cure (Davison & Degner, 1998; Degner et al., 1997). Although it seems the amount of information provided does not necessarily affect preferences, the manner of presentation and level of explanation does (Llewellyn-Thomas et al., 1991; Mazur & Hickman, 1994).

Miller, Brody and Summerton (1988) postulated a minimax hypothesis, where individuals are motivated by a desire to minimize the maximum danger to themselves. When faced with a medical decision, a client may prefer to relinquish control over the decisional process to an identified expert, whose decision is perceived to be a more reliable guarantee of minimizing aversiveness than one's own (Carver, Scheier, & Weintraub, 1989; Miller et al., 1988).

How are decisions made? There are few studies of information processing leading to decisions involving genetic risks. A systematic review of 547 randomized studies of informed decision-making, concluded there was a "paucity of well-designed,

theoretically driven, and adequately operationalized research assessing informed client decision making” (Bekker et al., 1999).

There is little guidance in the literature to explain how a client goes about evaluating alternatives, though it is suboptimal by decision analysts standards if all alternatives are not given adequate consideration. The way a client understands the alternatives and their implications is even more unclear (Pierce & Hicks, 2001).

Individuals often make decisions based on what they believe is important for themselves and their families (Baron, 1994). Their satisfaction with the decision process is often determined by the degree to which the decision made is consistent with their values (O'Connor & O'Brien-Pallas, 1989).

An understanding of how decisions are made regarding BRCA susceptibility testing also requires an understanding of how the client understands risk information.

Risk and Decisions

Protocols for genetic counseling and testing rely heavily on risk communication to provide information about personal and familial cancer risk (Botkin et al., 1996; Peters & Stopfer, 1996). In pretest counseling, sessions focus on the client’s family history of cancer and their risk of having inherited a cancer mutation. Post test counseling focuses on interpreting the test results, of individual and family risks for developing cancer, and on options for risk management. An accurate understanding of risk among participants may be critical to their decision making about whether to test, and for those who test positive, to their decision making about risk management (Croyle & Lerman, 1999).

To the lay person, the calculations of risks are averages that have limited value. Risk is experienced as a symptom of a hidden or future disease, as a subjective

personalized experience (Gifford, 1986). According to Gifford (1986), risk for breast cancer becomes internalized and is experienced as a state of being, which leads to an ambiguous relationship between health and ill-health. Personal experiences, as well as social and cultural background, mediate an understanding of clinical risk. 'Unmeasured ambiguity and uncertainty' surround the meaning of risk to the lay public (Gifford, 1986). Cultural systems create collective notions of risk and help in evaluating which risks are worth taking, who should take them, who is accountable for them, and whether a danger is possible to control (Douglas & Wildavsky, 1982; Jacobs, 2000).

Yates and Stone (1992) analyzed risk in various situations. They defined a risk-taking problem as a special kind of decision problem, because the relevant options include other considerations besides risk. That is, the worth of an alternative is a function of the risk and of other considerations with positive benefits, as well as possible negative features. Yates and Stone proposed that risk is characterized by three critical elements that interact to reduce an option's worth (a) potential losses, (b) the significance of the losses, and (c) the uncertainty of those losses. Risk is an inherently subjective construct: what is considered a loss is specific to the individual, as are the significance of the loss and its chance of occurring. Although Yates and Stone's work is based on experimental data, other studies support their ideas on genetic risk. It is perceived by clients as: (a) a global concept, sometimes interpreted as severity (Lippman-Hand & Fraser, 1979; Teigen, 1988); (b) as one of many considerations that complicates the decision (Frets, Duivenvoorden, Verhage, Ketzer, & Neirmeijer, 1990); and (c) as subjective by nature and relevant in decision making only in so far as it is subjective (Sagi, Shiloh, & Cohen, 1992; Shiloh & Saxe, 1989). Yates and Stone also emphasized the importance of the

multidimensionality of the risk concept. A single number ignores the multi-dimensional nature of the significance of the negative outcome and uses probabilities for specific outcomes as representations of uncertainty, whereas uncertainty is a larger concept (McCormick, 2002; Mishel, 1990; Morse & Penrod, 1999; Penrod, 2001).

The emerging literature on risk communication suggests that most individuals with some family history of cancer, including those at low to moderate risk, overestimate their personal cancer risk (Martin & Lobchuk, 2003). This finding of exaggerated perceptions of personal risk has been documented in studies about genetic testing (Andrykowski, Munn, & Studts, 1996), breast cancer (Black, Nease, & Tosteson, 1995; Lerman, Kash, & Stefanek, 1994; Smith et al., 1996), and in research on hereditary breast and ovarian cancer families (Berry et al., 1997; Bluman et al., 1999; Hallowell, Statham, & Murton, 1998a; Schwartz et al., 2000; Struewing et al., 1995; Winer et al., 1997). Black et al. (1995) found in women between 40 and 50 years of age, without a family history of breast cancer, respondents overestimated their probability of dying of breast cancer within the next 10 years by more than 20 fold.

Participants' decisions about genetic testing are influenced less by their actual risk than by their perceived risk and emotional factors (Lerman et al., 2002). Studies of breast cancer risk counseling programs are mixed on their reported effectiveness in changing perceptions of personal risk of cancer through standard education and counseling approaches (Lerman, Biesecker et al., 1997; Lloyd et al., 1996; Meiser & Halliday, 2002; Morris, Johnson, Krasikov, Allen, & Dorsey, 2001; Watson et al., 1999). However, a meta-analysis of 12 studies of outcomes of genetic counseling for women at increased

risk of developing breast cancer showed that counseling improved the accuracy of risk perception (Meiser & Halliday, 2002).

Sociocultural Context of Breast Cancer Risk

A look at the sociocultural context of breast cancer offers some insight into women's exaggerated perceptions of personal risk of breast cancer.

Exaggerated perception of risk has been mediated by a discourse of fear of breast cancer. Breast cancer is a symbolic issue in women's health and many emotionally charged political battles center around prevention, early diagnosis and treatment, and access to research (Surbone, 2001). In addition, breast cancer screening has developed in the context of risk and fear of breast cancer.

Through the success of American women activists, large sums of money have been earmarked for breast cancer research and have moved breast cancer to center stage as a public health concern in the United States. Yet through these successes, through the adoption of the term 'epidemic' and the 'risk' paradigm of biomedicine, these same actions have unintentionally reinforced exaggerated fears of breast cancer as an epidemic that threatens all women, especially younger women (Lerner, 1999).

Despite tremendous research efforts, science has not cured or prevented breast cancer. Instead, it has offered an elaboration of breast cancer risk factors and programs of vigilant surveillance via mammography, breast examination, and genetic testing. Attempts to reduce women's fear of breast cancer, through an elaboration of risk statistics, may exacerbate this fear through the increasing screening and surveillance recommendations that accompany designations of risk. This increasing fear in turn precipitates a greater desire for certainty. Ironically, increasing certainty and control can

be achieved by providing probabilistic risk information, including predictive genetic testing (Press, Fishman, & Koenig, 2000).

Lastly, the suffering and death from metastatic breast cancer are devastating and its image is imposed on many women with BRCA mutations. Current studies which attempt to address some of the psychosocial and interpersonal aspects of genetic testing, do not consider the sociocultural climate within which BRCA genetic decisions are made. The social and cultural context of genetic testing for breast cancer, specifically women's fear and use of individualized risk statistics, have implications for women making decisions about BRCA mutations.

Most research on how genetic-related decisions are made attempt to relate antecedent factors influencing the decisions. This previous research examines the correlation between specific choices and relevant psychosocial variables. Important to decision making is to understand the impact of undergoing predictive genetic tests upon emotional state and thus upon decisions leading to risk reduction and detection of disease.

Psychological Factors Influencing BRCA Decisions

Emotional Factors Motivating Genetic Testing

Research suggests that emotional factors can modify the cognitive processing of risk-related information when an individual is faced with a personally relevant health threat (Croyle, Yi Chun et al., 1997; Leventhal et al., 1983). This is common in risky decisions that are made under emotional stress (Janis & Mann, 1977). Given the possibility of receiving a positive or VUS BRCA mutation test result, one would expect genetic testing to be particularly stressful.

Janis and Mann (1977), in their conflict theory, used experiments to test the deleterious effects of high levels of stress on decision-making and risk-taking. Stress interferes with one's ability to consider the most relevant features of the situation and to carefully consider the pros and cons of alternate options. The conflict model maintains that extremely low stress and extremely intense stress lead to defective decision patterns, whereas moderate levels of stress are more adaptive and enhance vigilant decision making patterns.

Cancer worry, cancer specific distress, and frequent intrusive thoughts have been shown to motivate use of BRCA genetic testing in high-risk families (Durfy et al., 1999; Lerman, Schwartz et al., 1997). The specific effects of distress and emotional factors on genetic testing decisions and outcomes may depend on the level and type of distress, and on beliefs about being able to control the risk (Croyle & Lerman, 1999).

When outcomes are perceived as controllable, cancer-specific distress may motivate coping strategies, including genetic testing. McCaul, Branstetter, Schroeder, and Glasgow (1996) reported this in a meta analysis on mammography screening in women at risk. However, global distress, a perceived lack of control, may promote feelings of fatalism that interfere with health protective behaviors and thus avoidance of genetic testing. Lerman et al. (1998) found more frequent depression symptoms at six month follow-up in individuals who had high levels of cancer-related distress at baseline, but declined testing, than those who received positive test results. Their analysis suggested that those who declined testing were motivated to be tested to reduce distress, however, because of fears of discrimination and other adverse outcomes, they avoided testing. Thus, their cancer risk status remained uncertain and depression levels increased over

time. This suggests that genetic testing is a coping response that may be facilitated by disease-specific distress, if this action is perceived as leading to increased control over disease outcomes (Lerman et al., 2002).

Distress, Anxiety, and Depression After BRCA Testing

When clients were asked to speculate about what their reactions would be to disclosure of positive BRCA mutation carrier status, they expected depression (80%), anxiety (83%), and impairment in their quality of life (46%) (Lerman, Seay et al., 1995). In contrast to their expectations, the few studies of psychological outcomes associated with genetic testing for BRCA mutations, have shown low levels of distress among those found to be carriers and noncarriers (Croyle, Smith et al., 1997; Lerman et al., 1996; Schwartz et al., 2002).

Broadstock, Michie, Marteau (2000) in a systematic review of psychological consequences of predictive genetic testing found that none of the 15 studies reported increased distress (including anxiety, depression, general distress, and situational distress) in carriers or non-carriers at any point during the 12 months post testing. No differences were found at 12 months (three analyses) or at 3 years (three analyses) in any of the psychological outcomes measured. In all studies, emotional states remained within normal ranges. Both carriers and non-carriers showed decreased distress after testing, with this decrease being greater and more rapid among non-carriers. Test results did not predict emotional consequences in the majority of studies. Only three studies in this predictive genetic testing review were of HBOC, but the results are consistent with genetic testing results from Huntington's disease, spinocerebellar ataxia, and familial adenomatous polyposis. In the first long term study, Van Oostrom et al. (2003) reported

the psychosocial consequences of carrying a BRCA mutation 5 years after genetic testing. On several distress measures, there was not a difference between carriers and non-carriers.

Despite positive outcomes regarding distress in most studies, there were small subgroups of those tested with increased distress (Lerman et al., 2002). However, most increases were within the normal range of distress. Coyne, Kruus, Racioppo, Calzone, & Armstrong (2003) surveyed women in a high risk clinic and found that obtaining genetic testing may be less stressful than living with the awareness of a familial risk for cancer. The test result status of other family members is highly influential on the psychological impact of an individual's test result (Smith et al., 1999). Female BRCA carriers, who were the first in their families tested or whose siblings were negative, had significantly higher distress than other female BRCA carriers. Also, in those who were the first in the family tested, some distress related to the burden of conveying genetic information to relatives has been noted (Bish et al., 2002). Wylie, Smith, and Botkin (2003) reported significantly higher levels of distress in BRCA mutation carriers whose spouse was highly anxious and non-supportive. Thus it is important that research consider the family context of the individual tested to determine which individuals requesting genetic testing may require additional emotional support.

Broadstock et al. (2000) proposed several factors which may influence emotional consequences of genetic testing (a) awareness of pre-test risk status, (b) psychological coping mechanisms, (c) sample selection, and (d) counseling. Another explanation for test results not predicting emotional outcomes is the coping mechanisms that many individuals use in the face of a threat. One such mechanism is threat minimization,

whereby those facing a positive threat perceive it to be less serious than those not facing the threat (Croyle, Yi Chun et al., 1997).

Most of the studies reviewed on psychological factors were of self-selected populations, many in research registries, who had agreed to participate in psychological studies and have been followed up for no more than 5 years. Most of the studies had optimal models of genetic counseling that may have more beneficial outcomes than those in a clinic setting. Also, it may be that genetic mutation carriers experience more distress as they approach the likely onset of their condition (or the age of an affected relative).

Although research on behavior change after genetic testing is limited, initial results do not support substantial effects (Marteau & Lerman, 2001). In the next section, risk management behavior after genetic testing will be considered in greater detail.

Risk Management Decisions in BRCA Mutation Carriers

Screening Behavior

While motivation given for pursuing BRCA genetic testing included increased motivation to do self breast exams, and get regular clinical breast examinations and mammograms (Durfy et al., 1999; Jacobsen et al., 1997; Lerman, Seay et al., 1995; Struewing et al., 1995), limited data exists as to what degree clients participating in BRCA genetic testing will alter their breast and ovarian screening behavior over time.

Although cancer screening behaviors have been examined in women with a family history of breast and ovarian cancer, studies of breast and ovarian cancer screening in BRCA mutation carriers are few. Five such studies have been reported with varying results.

Breast Cancer Screening

Lerman et al. (2000) reported breast and ovarian cancer surveillance practices one year after BRCA mutation testing. They found that in 29 mutation carriers, disclosure of positive BRCA mutation test results did not lead to increased use of annual mammograms or ovarian cancer screening tests (68% complied with mammography recommendations before BRCA testing and 68% reported adherence one year after receiving positive test results).

Peshkin et al. (2002) determined from a prospective observational study of 41 BRCA1/2 carriers, overall the use of breast cancer screening was good (CBE uptake for carriers: 95%; noncarriers: 77%; and mammography uptake in carriers: 59%, in noncarriers: 47%). However, there was a relatively low uptake rate of mammography in younger carriers (ages 25-39 years: 39% versus age \geq 40 years: 74%).

Botkin et al. (2003) studied a kindred of women in Utah for two years following BRCA 1 testing. Both carriers and non-carriers significantly increased their use of mammography and breast self-exam from baseline. For women 40 years and older, 82% of mutation carriers obtained a mammogram in each year following testing. Younger carrier women also significantly increased their mammography utilization from baseline. Overall, 29% of the BRCA carrier women did not obtain a single mammogram by 2 years post-testing. At 2 years, 83% of the carrier women reported adherence to recommendations for breast self-exam and over 80% had obtained a clinical breast examination each year following testing.

Scheuer et al. (2002) also presented prospective evidence that BRCA testing and genetic counseling increased screening in 251 BRCA mutation carriers followed over a

mean of 24.8 months. There was an overall significant increase in mean number of mammograms, clinical breast exams, ovarian ultrasonograms, and CA-125 determinations after genetic testing. Breast self exam (BSE) was practiced by greater than 75% of the women at the time of genetic testing. The importance of BSE is supported in this group by the fact that it led to the diagnosis of five interval cancers, with four of the five tumors lymph node negative.

Ovarian Cancer Screening

In reports of ovarian screening behavior in BRCA carriers, compliance has been relatively low. At one year follow up, Lerman et al. (2000) reported BRCA carriers use of CA-125 testing and TVUS at only 21% and 15% respectively. Botkin et al. (2003) found that 19 BRCA carriers, at one and two years post testing, reported TVUS use of 26% and 11% respectively and CA-125 use of 32% and 37% respectively. In a clinical sample (n=79) of affected and unaffected BRCA participants, followed for 12 months after receipt of positive test results, Schwartz et al. (2003) found that CA-125 and TVUS screening was reported at 43% and 40% respectively, both reflecting an increased use compared with the year prior to testing. Scheuer et al. (2002) reported an overall increase in mean number of CA-125 and TVUS screening performed after genetic testing. On average after 15 months, 67.6% of participants were performing CA-125 testing and 72.9% were performing TVUS.

In the Botkin et al. (2003) and Lerman et al. (2000) studies above, they examined whether testing-related distress reduced adherence to cancer screening. This was not demonstrated among participants tested in either study. It may be that genetic information

leads to risks being perceived as unmodifiable and to less adherence to behaviors that would lower health risks (Senior et al., 1999).

Although these are the first studies to characterize screening behaviors following BRCA mutation testing, they shed little light on the factors that influenced these surveillance decisions. Limitations of these studies are: the relatively short timeframe for follow-up, most data were obtained from clinical research programs involving very high-risk families, and some studies mixed affected and unaffected BRCA carriers together in the reports.

Prophylactic Surgery

A few studies have recently been published about the extent to which prophylactic surgeries are chosen as HBOC prevention options in BRCA mutation carriers. Also included are factors that influence the decisions for prophylactic surgeries and the psychosocial implications of these surgeries for women with BRCA1 and BRCA2 mutations. Two recent studies indicate that the degree to which BPM is chosen as a risk reduction intervention may vary according to culture, healthcare system, insurance coverage, provider attitudes, and other social factors (Bouchard et al., 2004; Julian-Reynier et al., 2001).

Studies from the United States have reported fewer women choosing BPM than BPO following BRCA1 and BRCA2 mutation testing. Scheuer et al. (2002) studied 233 affected and unaffected women with BRCA mutations over a mean period of 24.8 months. These researchers reported that 14.9% underwent BPM at a median of 5.3 months after test results (prior to testing: 8.6% had BPM and 8% had undergone BPM for breast cancer) and 50.3% underwent risk reducing salpingo-oophorectomy (RRSO) at a

median of 3.4 months after receiving genetic test results. Women electing BPM were younger and had a stronger family history of breast and ovarian cancer than those opting for screening. Those electing RRSO were older (64% >40years) and more likely to have had a prior breast cancer diagnosis than those not opting for surgery. They did not have more family members affected with breast or ovarian cancer than those not opting for RRSO.

In a prospective observational study, Lerman et al. (2000) found only 3% of unaffected BRCA mutation carriers (n=29) had undergone BPM within a year of learning their BRCA mutation carrier status. A small subset (n=8) of these women had received BPM prior to genetic testing. Thirteen percent of carriers obtained BPO within a year following BRCA testing.

In a Utah kindred, Botkin et al. (2003) found in BRCA1 mutation carriers that BPM was not utilized within the first 2 years following testing, although 11% were considering this procedure. These researchers also reported that 46% (12/26) chose BPO, including 78% of women 40 years of age or older. In this study an additional 30% (11/37) had obtained BPO prior to testing. Income, education, family cancer history, personal cancer history, general distress, and test specific distress were not predictive of a decision to obtain BPO.

In the year following BRCA testing, Schwartz et al. (2003) reported that 27% of affected and unaffected mutation carriers and 5% of uninformative patients received BPO. Perceived risk for ovarian cancer, family history of ovarian cancer, ovarian cancer worries, age, and test results predicted undergoing BPO.

Studies from the Netherlands demonstrate higher selection rates of prophylactic surgeries in women with BRCA mutations than the United States. In the Netherlands, (Meijers-Heijboer et al., 2000) reported that 51% of unaffected (cancer free) BRCA1 and BRCA2 mutation carriers chose BPM over screening and 64% chose BPO within 2 years after testing. Parenthood was found as a predictor for BPM and age was associated with BPO.

Lodder et al. (2002), also from the Netherlands group, described follow-up of 26 BRCA carriers (some were the same as above study), assessing the influence of psychological distress on risk management options chosen. The 51% who chose BPM had significantly higher general and cancer-related distress levels than mutation carriers who opted for surveillance. A higher distress level in women opting for BPM was also observed in other studies (Meiser, Butow, Freidlander et al., 2000; Scheuer et al., 2002; Stefanek et al., 2001; Wagner et al., 2000). This difference in level of distress was highest at pre- and post-test and had almost disappeared at one year follow-up. Also, mutation carriers opting for BPM were more often in their thirties, had young children, and had a longer awareness of the genetic nature of cancer in the family than those opting for regular screening. Fear of leaving young children was an important independent factor in deciding for surgery (Lodder et al., 1999).

Psychosocial Outcomes

The psychosocial sequelae of BPM in high and moderate risk women, including those with BRCA mutations, have been reported in a few studies. Collectively data suggests that high risk women report satisfaction with their decision to undergo BPM and adjust well emotionally after surgery, although satisfaction with reconstruction may be

less optimal (Borgen et al., 1998; Frost et al., 2000). These outcomes must be weighted against the irreversibility of the decision, potential problems with implants and reconstructive surgery, and the occurrence of adverse psychological and social outcomes in some women (Frost et al., 2000). Moreover, two decades of research have shown that most women with breast cancer can safely be treated with breast-conserving surgery instead of mastectomy (Eisen & Weber, 2001). Thus it is difficult to accept that prevention of HBOC should be more extreme than the cure.

In Lodder et al. (2002) study above, about half the BRCA carriers who underwent BPM (n=14) reported a negative influence on body image, intimate relationship, and physical well-being. For spouses, surgery did seem to have a negative effect on the frequency of intimate contact with their spouses up to eight months after surgery. From interviews, the impression was that this reduction in intimate relationship was due more to the woman feeling inhibited to have intimate contact, than due to the partner's. However, all but one did not regret their decision at 1 year after testing. The major reason for the Dutch women's overall satisfaction may be due to a sense of relief as a result of significant risk reduction of developing breast cancer. Research by van Oostrom et al. (2003) corroborated these findings. These researchers reported a follow up study of women who had undergone BPM. At 5 years they reported a significant reduction in fear of cancer, but had a less favorable body image and changes in their sexual relationship.

One explanation for the few regrets reported after BPM might be that regretting one's autonomous decision for an irreversible surgical intervention may lead to 'cognitive dissonance' (Croyle, Smith et al., 1997). People want consistency between their cognitions (i.e. attitude, emotion, and behavior) as inconsistencies create

dissonance, which leads to uncomfortable and tense experiences (Festinger, 1957).

Cognitive dissonance is assumed to be smaller if one feels less autonomy in making a decision (e.g. because of external pressure). This could be seen in a large retrospective study of 370 high risk women who underwent BPM over a mean of 14.8 years (Stefanek et al., 1995). Of 21 (5.6%) women who regretted having undergone BPM, the subject of surgery had been initiated by their physicians (instead of by themselves) and they had insufficient information about surgery. Stefanek et al. (1995) also reported these women's reasons for regretting BPM as (a) severe emotional trauma and/or lack of psychological support after surgery, (b) complications of surgery and reconstruction, (c) dissatisfaction with cosmetic effect, (d) residual or phantom pain, (e) fears that implants would impede the adequacy of detecting cancer in residual breast tissue, and (f) diminished self-image or sexual satisfaction.

Cultural and Socio-Economic Differences

In addition to the geographic differences noted between Dutch and United States BRCA mutation carriers, prophylactic surgery may also vary according to culture, healthcare system, insurance coverage, provider attitudes, and other socio-economic factors (Eisinger et al., 1999). Minimal research has been done related to these factors.

In an analysis of the differences between United States (US) and French consensus statements (Burke et al., 1997; Eisinger et al., 1998) about clinical management of women with BRCA mutations, Eisinger, Geller, Burke, and Holtzman (1999) speculated that the difference in regard to prophylactic surgery partly reflected the cultural context in which physician and patients make decisions and health policies are formed. Although both consensus statements conclude that BPM and BPO are an option

for women despite incomplete evidence, the approach to decision making differs significantly. The French document describes each procedure as “a mutilation... (which) should be envisaged for medical reasons only” (Eisinger et al., 1998). It further indicates that doctors should “oppose” BPM under age 30 and BPO under age 35 years, and should consider them only when the risk of breast cancer is more than 60% and the risk of ovarian cancer is more than 20%. French law requires clear therapeutic justification for physicians to invade a patient’s body, even if the patient has given permission for the procedure. The French document also recommends that women wait several months before considering either procedure. In the US, only informed consent is required for these surgeries and does not speak to the possibility of active opposition to a woman’s intention or suggest a delay. Eisinger et al. (1998) suggest cultural differences related to the symbolic value of the breasts, attitudes toward fertility (French conservative attitude due to concern about low birth rate), and more general cultural norms of paternalism (French physicians) versus autonomy (US physicians) in decision making, resulting from values of communitarianism (French) versus individualism (US).

In the first international comparison of preventive strategies, Julian-Reynier et al. (2001) demonstrated the existence of variations in acceptability in English, French, and Canadian women at risk for HBOC attending three genetic clinics for the first time. These clinics were located in Montreal (Quebec, Canada), Marseilles (France) and Manchester (Great Britain). French women were the most reluctant about prophylactic surgeries, moderately favored chemoprevention, and highly favored mammography. British women were more in favor of prophylactic surgery and chemoprevention, but were least positive about mammography. Women from Quebec, Canada resembled the

French attitudes toward mammography and chemoprevention, but agreed more with the British about early BPM. Quebec women's attitudes toward BPO resembled the British for early age indications and with the French for indications for women over age 35 years. It was hypothesized that differences in prophylactic surgery by the French women was that they attach more importance to their breasts and ovaries than the British or Quebec women, who may have a greater tendency to value life itself over breasts and ovaries (Julian-Reynier et al., 2001).

The second part of this international research study was reported by Bouchard et al. (2004). Substantial differences in the way cancer geneticists deal with environmental risk factors, breast and ovarian cancer testing, chemoprevention, and prophylactic surgery were found. Cultural differences included the social representation of risk and health. Physicians from Canada and Anglo Saxon countries promote a model of health that suggests individual responsibility for risk management, where the physician instructs the patient about his/her health status and instructs him/her to take the necessary measures to avoid or restrict the effects of illness. In France, more emphasis seems to be put on medical authority in the relationship between providers and patients. In addition, body integrity and the symbolic value of breasts may carry a different value for French physicians, which may explain differences in acceptability of BPM. Also, interpretations of scientific evidence and medical uncertainties and the impact of leadership with respect to new ideas and technology innovation could contribute to the context in which BRCA testing is disseminated in the different countries, resulting in geographic variability (Bouchard et al., 2004).

Cost of genetic testing and prophylactic surgeries may also be a factor in the differences noted between countries. In the Netherlands, costs for genetic testing, surveillance, and prophylactic surgery are covered by both public and private health insurances. Kuerer et al. (2000) evaluated health insurance coverage policies in the US for BPM and BPO. This cross-sectional nationwide survey of 481 medical directors from the American Association of Health Plans, Medicare, and Medicaid showed significant variation for health insurance coverage for prophylactic surgery. Only 44% of private plans had specific policies for coverage of BPM for clients with a strong family history of breast cancer and 38% of plans had coverage for a BRCA mutation. Only 20% of total responding plans had a policy for coverage of BPO under any clinical circumstances. A more recent retrospective study by Kauff et al. (2001) at Memorial Sloan Kettering Hospital in New York reported that 38 of 39 risk reducing surgeries in women with BRCA mutations were covered in full, less deductibles and copayments. Rates of insurance reimbursement did not vary by type of insurance, personal history of cancer, or type of procedure. Information on insurance coverage needs to be updated, as both of these studies are dated and may not reflect current insurance policies.

Analysis and Critique of the Literature

Genetic testing is being applied to detect individual susceptibility to breast and ovarian cancer with a focus on individual risk management. The hope is that awareness of genetic risk will enhance informed risk management by clients. Researchers have recognized the movement of predictive genetic testing from the research to the clinical environment. There is an emerging body of literature which addresses data derived from clinical research programs involving very high risk families, some of whom were

members of cancer genetic registries. These research programs have included protocols for extensive pre-and post-test counseling about risks and benefits of testing, protections of ethical review, psychological assessment, and follow-up, staged over months. Few studies have presented data from clinic-based genetic testing programs, where the lack of protective factors associated with the research environment may have higher rates of adverse consequences for clients tested (Dorval, Paternade et al., 2000; Schwartz et al., 2002).

Additionally, literature exists which indicates that overestimated perceived risk, cancer worry, and cancer-specific distress motivate use of BRCA genetic testing in high-risk families (Durfy et al., 1999; Lerman, Schwartz et al., 1997; Lerman et al., 2002). This research further suggests that genetic testing is a coping response that can be facilitated by disease-specific distress, if this action is perceived as leading to increased control over disease outcomes. Although these studies shed some light on predictors of the use of BRCA genetic testing, little is known about the mechanisms by which risk perceptions, cognitive factors, and the influence of family and others affect a woman's management of her susceptibility to HBOC. No studies to date have addressed the genetic testing and risk management decision making experiences of unaffected women who receive variant of uncertain clinical significance BRCA test results.

A few studies have addressed the short term psychological impact of receiving BRCA mutation test results among research and clinic based families (Croyle, Achilles, & Lerman, 1997; Dorval, Paternade et al., 2000; Lerman & Croyle, 1996; Marteau & Croyle, 1998; Schwartz et al., 2002). This literature suggests that the severity of psychological risks posed by genetic testing is not great. Although some studies report an

initial increase in anxiety following predictive testing, this tends to be transient and not clinically significant. However, subgroups of individuals with certain psychological traits may be more vulnerable to adverse effects. Standardized measures of distress may not be sensitive enough to determine more subtle changes in functioning. It is unknown how individual differences in tolerance for uncertainty or need for information may moderate of the impact of genetic test results on psychological functioning (Croyle, Dutson, Tran, & Sun, 1995).

In addition, emerging data is mixed on providing evidence that genetic testing promotes changes in risk management (Botkin et al., 2003; Lerman et al., 2002; Meijers-Heijboer et al., 2000; Peshkin et al., 2002; Scheuer et al., 2002; Schwartz et al., 2003). Although the first studies to characterize screening and prophylactic surgery behaviors following BRCA testing have been reported, they shed little light on the factors that influenced these risk management decisions. Furthermore, the studies are complicated by mixing unaffected and affected women in the same report. Hallowell, Foster, Eeles, Ardern-Jones, and Watson (2004) in a qualitative study of affected womens' responses to BRCA genetic testing, indicated that a majority of women adopted a fatalistic approach with regard to their future health and did not regard their genetic risks as a threat to self. The data suggested that affected women understand genetic risks of HBOC within the context of their previous disease experience.

An understudied area of BRCA research is the family and social contextual factors involved in genetic testing and risk management by women with positive and VUS BRCA test results. Initial data suggest that genetic testing for breast cancer susceptibility is motivated partly by the desire to help other family members (Geller et

al., 1999). Although the majority of genetic testing participants communicate their risk to other family members, only recently has data been reported on BRCA mutation carriers about the content, process, and impact of these communications (Bonadona et al., 2002; Croyle & Lerman, 1999; d'Agincourt-Canning, 2001; Hamilton et al., 2005; Hughes et al., 1999).

Rees, Fry, and Cull (2001) identified that the influence of personal experience of cancer, through involvement with affected relatives, has been neglected in the literature and found strong theoretical grounds for the hypothesis that dimensions of personal experiences may influence response to cancer risk. "Perceptions of breast cancer and beliefs about the disease are likely to influence how an individual reacts (in terms of thoughts, feelings, and decisions) to their own risk status" (Rees et al., 2001).

Much of the current research is based on a static view of genetic testing use, largely from the perspective of health care providers and researchers, using standardized instruments. Current research describes pieces of the process, but does not attempt a view of the total experience of genetic testing. What is lacking is a focus on the clients' perspective, as well as an understanding of the evolving process as clients move through a trajectory of managing their susceptibility to hereditary breast and ovarian cancer.

Although this researcher's initial plan was to explore unaffected women's risk management decision making after testing with a positive or VUS result, it became obvious after a few interviews that some women came to BRCA testing with plans for prophylactic surgery. To better understand these apriori decisions, the focus changed to explore the broader context of genetic testing in which unaffected positive and VUS carriers came to manage their susceptibility of hereditary breast and ovarian cancer.

The purpose of the study then became to explore the influence of family and social contextual factors in how unaffected BRCA positive and VUS carriers conceptualized their cancer risk, interpreted and coped with BRCA test information, and made subsequent risk management decisions; and to develop a grounded theory based on the perceptions, beliefs, and actions of these women. With a broader scope, questions were added and included: (a) Given the benefit of hindsight, how would unaffected BRCA positive and VUS carriers describe their experience of discovering their BRCA status? (b) What is the influence of womens' experiences of breast cancer in their family in how they conceptualized their breast and ovarian cancer risk? (c) How do women interpret, assign meaning, and act on the complex information about positive and VUS test results and risk management? (d) What is the decision making process through which they arrive at these actions? and (e) What are the roles their family and others played in their decision making? The following chapter will explain the methodology used to explore these questions.

Chapter III

METHODOLOGY

This qualitative study was guided by the theoretical perspective of symbolic interactionism (Blumer, 1969) and the research approach of grounded theory (Glaser & Strauss, 1967; Strauss & Corbin, 1998). Symbolic interactionism is a useful perspective for the study of the meaning and consequences of BRCA mutation testing in unaffected women. By emphasizing personal interactions, this perspective encourages the researcher to study how people think, communicate, and interact and what effect these processes have on how they perceive their BRCA genetic testing and risk management experiences. Blumer (1969) explained about symbolic interactionism, “(it)...lodges its problems in this natural world, conducts its studies in it, and derives its interpretations from such naturalistic studies.” The method for this study is based on this philosophical perspective. Specifically, grounded theory approach was used in collecting data from unaffected BRCA positive and VUS mutation carriers in their own environments and in analyzing these data using constant comparative analytic techniques (Glaser, 1995; Strauss & Corbin, 1998).

This chapter describes the grounded theory method, discusses participant inclusion criterion, and entrée to the specific population addressed. Protection of the

participants is addressed and the progression of data collection and analysis is described. Finally the methods which provided methodological rigor are explained.

Grounded Theory

Interpretive inquiry, using grounded theory, is particularly suited to the study of BRCA genetic testing and risk management in unaffected women susceptible to breast and ovarian cancer, for three reasons. First, because of its focus on meaning defined through interaction, and its sensitivity to the evolving and unfolding nature of events, and to the interrelationships among conditions, actions, and consequences (Strauss & Corbin, 1998). Second, it is useful in conceptualizing behavior in complex situations and in understanding the impact of new technology on the health care system and the client (Swanson, 1986). Lastly, this method allows the researcher to obtain the intricate details about phenomena such as feelings, thought processes, and emotions that are difficult to learn about through more conventional research methods (Strauss & Corbin, 1998). It can also form the basis for interventions and social policy.

Strauss and Corbin (1998) use the term “grounded theory” to mean theory inductively derived from data (“grounded” in the data collected), systematically gathered, and analyzed through the research process. A researcher does not begin with a preconceived theory in mind, as in deductive reasoning. Instead, the research begins with an area of study and allows the theory to emerge from the data. It results in the development of middle-range theories to explain behavior and processes (Charmaz, 2000). Grounded theory, because it is drawn from data, offers insight, enhances understanding, and provides a meaningful guide to action for unaffected women undergoing genetic testing and managing their susceptibility to breast and ovarian cancer.

Nursing theory focuses on meaning and is developed for describing, explaining, predicting, or prescribing nursing care (Meleis, 1997; Parker, 2001). The primary purpose of this study was to enhance understanding of the social processes involved in BRCA genetic testing and managing susceptibility to hereditary breast and ovarian cancer. Since there is minimal knowledge about how BRCA genetic testing affects unaffected women's lives, this methodological approach is appropriate, as it helps to shed light on the basic social processes involved in BRCA genetic testing and managing susceptibility, including interactions with family, friends, and health care professionals.

Research Strategies

Sample Selection

As is consistent with grounded theory methodology, study participants were selected based on their ability and interest in explaining and articulating their experiences of BRCA genetic testing and how they came to manage their risk of hereditary breast and ovarian cancer. Therefore, the initial sample was a convenience sample. Subsequent sampling was based on the grounded theory process of theoretical sampling (Glaser & Strauss, 1967). Theoretical sampling is the process of deciding on who and what to sample, based on previously collected and analyzed data. This sampling was used to collect more data to test and develop categories and their relationships and to assure that the full range and variation in the categories existed. Theoretical sampling, intensive interviewing, and data analysis were concurrent, repeating processes and driven by the theoretical coding scheme that emerged. As the theoretical scheme emerged from the data analysis, through the process of constant comparison, more focused interview questions and specific types of participants were selected to broaden an understanding of women's

experiences of BRCA genetic testing and risk management of HBOC. Sample size was determined when no new conceptual data (information redundancy) was reached, and the requirement of data saturation was met.

Participant inclusion criteria for this study included (a) female age 18 and older, (b) carrier of a BRCA1 or BRCA2 mutation or a variant of uncertain significance, (c) not affected with breast or ovarian cancer prior to genetic testing, and (d) no psychiatric or cognitive disorder which would preclude informed consent.

Protection of Participants

After obtaining approval of the Committee on the Protection of Human Subjects at the University of San Diego and the Human Research Protection Program's Institutional Review Board at a large metropolitan medical center with a genetic screening program (see Appendix A), data were collected using semi-structured in-depth interviews of the participants (Appendix B). Prior to interviewing, the purpose of the study and assurance of confidentiality were explained to each participant. Also, their rights to refuse to answer any question or decide to terminate the interview at any point, if desired, were explained. Any questions were answered and each participant signed a consent form (see Appendix C). Out-of-state/country participants either faxed or mailed their signed consent forms to the researcher. A copy of the researcher signed consent form was then mailed to each participant, along with a check for \$50, in appreciation for their participation. Coded numbers provided confidentiality and anonymity of all participants so that no names or other identifying descriptions were present on the data. Only the researcher knew the list of names with corresponding codes. Identifying information was kept in a locked cabinet in the researcher's office.

Recruitment

Participants were recruited over a thirteen month period (May 2003 through June 2004) from multiple sources. Initially to gain entrée to a cancer genetics setting the principal investigator (PI) set up an independent study with the Director of the Clinical Cancer Genetics Specialized Clinical Unit and the cancer genetic counselor at a local university comprehensive cancer center. The PI continued monthly meetings with the director and cancer genetic counselor throughout the research data collection and analysis and attended all BRCA support group meetings sponsored by these professionals. Clients from this testing and counseling program were the first to be interviewed. However, only 5 clients were recruited from this program, so the PI continued recruitment using the following additional strategies: (a) posting on a national BRCA peer support internet website research page, (b) sending letters, flyers, and subsequent follow-up letters from the director of a cancer genetics program in a large tertiary medical center in the southwest (see Appendix D), (c) obtaining referrals from genetic counselors from a posting on the National Society of Genetic Counselors (NSGC) Cancer Genetics Research Directory, (d) sending letters and flyers from a nurse genetic counselor in the Midwest; and (e) writing an article in the online journal NurseZone which recruited for participants. Three participants were recruited by snowball sampling. The last three strategies were added to meet the needs of theoretical sampling. That is sampling was continued to explore and compare the dimensions in unaffected womens' interpretation of their VUS mutations and to achieve a broader age range of participants. This was important to give variation and density to the categories, because information about unaffected VUS carriers had not been previously reported in the literature.

To recruit from the online website, the executive director of the national support organization for women with hereditary breast and ovarian cancer gave approval for posting a flyer on their research page. An announcement describing the study was also sent by electronic mail from the executive director to members of the organization. An application was made and approval was also obtained for posting on the National Society of Genetic Counselors' (NSGC) Cancer Genetics Research Directory. This announcement described the study for referral by genetic counselors and solicited participants for the study.

Particularly difficult to recruit were unaffected women with a genetic variant of uncertain significance. Correspondence with Myriad Genetics laboratories indicated that 12% of patients who have BRCA1/2 testing, receive the variant of uncertain significance result. Of these patients who specify their personal history of cancer, 32% do not have a diagnosis of breast or ovarian cancer and 68% do (A. Deffenbaugh, personal communication, 21 October 2004). Based on these figures, only about 570 women tested through Myriad Genetics Laboratories (who hold the patent worldwide for BRCA genetic testing) would have met the criterion for this study in May 2002, when the study began.

Data Collection Procedure

Data were collected from individual interviews and observational memos. Unaffected women, who were BRCA or VUS carriers and chose to participate, contacted the interviewer by phone or e-mail. For local participants (n=8), a convenient time was determined for a 60 to 90 minute audiotaped, face-to-face interview conducted in their home. Informed consent was obtained at the time of the interview. For out-of-state and out-of-country participants (n=22), a convenient time was determined on intake for a

telephone interview from their home. Also requested at intake was a copy of the participant's BRCA test result, permission to audiotape the telephone interview, and their consent to participate. A copy of the consent form was sent to the participant by facsimile, postal mail, or e-mail and returned to the PI by facsimile or postal mail. During the telephone interview, informed consent was obtained and any questions the participant had were answered. The telephone interviews lasted from 60 to 160 minutes. The interviews were audiotaped and the researcher recorded socio-demographic and family history data, as well as observational and methodological notes.

Responding to open-ended, semi-structured interview questions (Appendix B), participants provided verbal descriptions of how they came to have BRCA testing, how they learned their test results and from whom, with whom they shared their results with, what measures, if any, they took to reduce their risk for breast and ovarian cancer, how they considered the advantages and disadvantages of the risk management options, what role their family and others played in their decision making, and what was needed to manage their health in the future. A short second interview was conducted with three participants to clarify if they had carried out their intended surgical procedures after the initial interview. Participants' not recruited through the two medical centers provided copies of their BRCA genetic test results for validation.

Data Management

Socio-demographic Data

Participants provided socio-demographic and family cancer history information (Appendix B). Variables included age, marital status, age and gender of children, occupation, education, race, ethnicity, family income level, history of cancer in self and

family, initial family member tested, BRCA results and date of testing, surgical history, current use of estrogen, and menopausal status. A family cancer history was obtained of first, second, and third degree relatives.

Qualitative Interview

Interviews evoked ideas, thoughts, and memories in the participants own words. The interviews were guided by open-ended questions designed to elude the meaning of the participant's genetic testing and subsequent risk management decision making experiences. The researcher endeavored during the interview to maintain enough flexibility to elicit individual stories while gathering information consistently to allow for comparison between and among participants. A copy of the interview guide is at Appendix B. Thirty interviews were audiotaped and later transcribed by a professional transcriptionist. Observational and methodological notes were recorded and kept with the transcribed notes of participants.

Participant Characteristics

A total sample of 30 participants was the data source. The sample consisted of two groups (a) unaffected females (no history of breast or ovarian cancer prior to genetic testing) who tested positive for a BRCA1 or BRCA2 mutation (N=21); and (b) unaffected females who tested with a genetic variant of uncertain significance (N=9). These VUS test results are those in which the lab identifies a new sequence alteration, which may or may not disrupt the function of the protein, and whose clinical significance has not yet been determined. Test results indicate a "genetic variant of uncertain significance" (Myriad Genetics, 2004). The women who participated in this study had genetic testing between 1994 and 2003, with the majority tested in 2002 and 2003.

The women ranged in age from 22 to 60 years, with a mean age of 40 years. The mean age of women in the BRCA positive group was younger (39 years) than the VUS group (43 years). Twenty (66%) were married, 3 were single, 3 were divorced, 3 were partnered same sex, and 1 was widowed. The median number of children was 2, with a range of 0-6. The mean age of their children was 17 years, with the mean age of children of women in the BRCA positive group younger than the VUS group (11.1 years versus 20.2 years, respectively). The mean years of education was 15.8, with a range of 11-21 years. The majority of the group were Caucasian (90%), 2 were Black, and 1 was Hispanic. Eight (27%) were of Ashkenazi Jewish descent, all in the BRCA positive group. The median family income range was \$50,000 to less than \$75,000. The majority of women had professional careers (e.g. teacher, speech pathologist, registered nurse, molecular biologist). The mean number of first degree relatives with breast or ovarian cancer was 1.5 (range 0-4) for BRCA positive carriers and 1.1 for BRCA VUS carriers. Twenty one participants had mothers with breast cancer, 1 with ovarian cancer, 1 had both ovarian and breast cancer, and five had neither parents with cancer. Table 1 presents participants' socio-demographic characteristics. Table 2 provides participants' family cancer histories.

Data Analysis

Analysis of the data was accomplished using grounded theory method. Transcriptions of the interviews and observational memos were analyzed using constant comparative analysis (Glaser & Strauss, 1967; Strauss & Corbin, 1998). Data were analyzed as they were collected, through the process of coding, for the purpose of generating conceptual categories. Common themes of the BRCA testing and risk

Table 1
Participants' Socio-demographic Characteristics

	Total Sample n=30	BRCA Positive n= 21 BRCA 1= 12; BRCA 2= 9	Variants of Uncertain Significance n= 9
Age (mean) (range)	40 years 22-60 years	39 years 22-54 years	43 years 29-60 years
Marital Status	Single n=3 Married n=20 Divorced n=3 Partnered same sex n=3 Widowed n=1	Single n=3 Married n=14 Divorced n=1 Partnered same sex n=3	Married n=6 Divorced n=2 Widowed n=1
Number of children (median) (range)	2 0-6	1 0-6	2 2-4
Children age (mean)	17 years	11.1 years	20.2 years
Years of education (mean) (range)	15.8 years 11-21 years	16.1 years 12-21 years	15 years 11-19 years
Race	Caucasian n= 27 Black n= 2 Hispanic n=1	Caucasian n= 21	Caucasian n= 6 Black n= 2 Hispanic n=1
Ashkenazi Jew	n= 8	n=8	n= 0
Family income (median range)	\$50,000-<\$75,000	\$75,000-<\$100,000	\$50,000-<\$75,000

Table 2
Participants' Family Cancer Histories

	Total Sample n= 30	BRCA Positive n= 21 BRCA1= 12; BRCA2= 9	Variants of uncertain significance n= 9
Parents with cancer	Mother cancer n= 17 Father cancer n= 1 Both parents cancer n= 7 Neither parents cancer n= 5	Mother cancer n= 11 Father cancer n= 1 Both parents cancer n= 5 Neither parents cancer n= 4	Mother cancer n= 6 Both parents cancer n= 2 Neither parents cancer n= 1
Mother with breast or ovarian cancer	Both n= 1 Breast n= 21 Ovarian n= 1 Neither n = 7	Both n= 1 Breast n= 14 Ovarian n= 1 Neither n = 5	Breast n= 7 Neither n = 2
Father's cancer type	Breast n= 1 Lung n= 1 Prostate n=2 Colon n=1 Skin n= 1 Precancer skin n=1 Kidney n=1	Breast n= 1 Prostate n=1 Colon n=1 Skin n= 1 Precancer skin n=1 Kidney n=1	Lung n= 1 Prostate n= 1
Mother's age at cancer diagnosis: mean (range)	48.5 years (33-77 years)	46 years (34-66 years)	53.4 years 33-77 years
Number of first degree relatives with breast or ovarian cancer: mean (range)	1.4 (0-4)	1.5 (0-4)	1.1 (1-2)
Total of first and second degree relatives with breast or ovarian cancer: (mean range)	2.6 (0-5)	2.7 (0-5)	2.3 (1-5)

management experience were identified that provided the context to meanings, circumstances, and conditions of what it was like to obtain a positive or VUS test result and make decisions about managing hereditary risk. As the data were collected through open-ended questions, interviews were coded using open, axial, and selective coding (Strauss, 1987). For purposes of clarity, these types of coding are presented as unique steps of the process, however these steps are not distinct or linear. Qualitative Solutions and Research's (QSR) software, NUD•IST VIVO 2.0 was used for data management.

Initially data (transcripts and observational memos) were examined line-by-line for any and all categories, and the categories were assigned names or codes through 'open coding'. This helped to break down, examine, and compare events, activities, and interactions for their similarities and differences. Conceptually similar events, activities, and interactions were then grouped together to form categories and subcategories (Strauss & Corbin, 1998). Samples of concepts with their properties and variations were drawn and theoretical ideas, questions, and codes were recorded and tracked using theoretical memoing. Once the initial set of categories were developed, using axial coding, the researcher searched for verification, saturation, and relevance of the categories. Eventually certain categories were found from various interviews (verification), were observed to be supported over and over again by data with no new information emerging (saturation), and were found to be related to other categories of data (relevance). Thirty interviews (21 positive and 9 variants) were reviewed and coded before saturation was reached. Interviews were reviewed, comparing and contrasting women in various age categories (20-29, 30-39, 40-49, 50-59; and one age 60 years) and by positive and VUS mutation status. The categories that were verified, reached saturation, and seemed to have

particular importance in relation to the phenomenon of BRCA genetic testing and risk management were tentatively labeled “central” or “core” categories. This then led to selective coding, a process of searching for the linkages among the various sub-categories and sub-processes in relation to the core category (Strauss & Corbin, 1998). The result of selective coding was theoretical integration and formation of a substantive grounded theory. A core variable was identified and theoretical scheme of categories and subcategories were organized around the core variable. Identifying a core variable in grounded theory consists of coding the data according to how the participants define or describe their central phenomenon of concern and asking the data a series of questions: “What is going on with the participants? What are the main problems? Why do these women need to be studied and interventions developed?” This category best explained how BRCA genetic testing, including risk management decision making was experienced and acted upon in women with positive and inconclusive BRCA test results (Corbin & Strauss, 1990; Swanson, 1986).

Methodological Rigor

By design, qualitative research involves interpretive description which requires attention to rigor. Thus for grounded theory to be accepted as methodologically sound, trustworthiness must be established. Lincoln and Guba (1995) identify four factors to ensure trustworthiness: credibility, transferability, dependability, and confirmability. Krefting (1991) indicates that credibility may be established by spending time with the informants, using reflexive analysis or continual examination of the researcher’s knowledge influence, writing memos, using peer examination, and employing consistent interviewing techniques. This research employed these techniques.

Guba and Lincoln (1989) propose that incorporating reflexivity will enhance the rigor associated with grounded theory studies. They maintain that objectivity can be addressed by coming clean about predispositions so readers can adjust offered interpretations in suitable ways.

The researcher is not a genetic oncology nurse. However, as an experienced medical surgical clinical nurse specialist, family nurse practitioner, educator, and administrator, with a career-long interest in genetics and primary and secondary disease prevention, the researcher has certain beliefs and preconceptions (feelings, theories, and inclinations) which were brought to this study. In the various nursing roles, the researcher has observed client decision making. Therefore, the following biases are disclosed to increase researcher sensitivity to beliefs and preconceptions.

Five researcher beliefs are identified. First, decision-making experiences vary among clients. Those experiences are influenced by a variety of personal, as well as professional experiences. Second, regardless of decision making ability, external forces such as physician, nurse, and genetic counselor style or insurance protocol can impact the process by which clients make decisions. Third, clients integrate many human experiences into the meaning of decision-making. Fourth, cognitive skills vary among clients and over time. These skills are influenced by their knowledge and experiences, their awareness of those experiences, and their ability to integrate knowledge and experiences into their decision making. Fifth, the psychological state of the client influences the context of decision making. Competing alternatives with uncertain risks and outcomes may affect the psychological equilibrium of clients facing stressful decisions.

Transferability or applicability to other women with a similar diagnosis is not a specific goal of this research. No attempt was made to generalize findings. Rich thick description of BRCA mutation carriers' experiences will enable readers to determine whether the findings can be transferred because of shared characteristics (Creswell, 1998; Erlandson, Harris, Skipper, & Allen, 1993). Study findings are presented as one possible account of BRCA carriers experiences and should be considered emergent and tentative (Glaser & Strauss, 1967; Strauss & Corbin, 1998). Since minimal research currently exists on how women subjectively experience BRCA genetic testing and risk management, coming to understand their perspective may help healthcare providers meet their concerns. Becoming sensitized to experiences about genetic susceptibility deepens understanding and allows for broadening perspectives, and this was the reason qualitative design was used in this study.

The techniques of discriminant sampling, supplemental validation, and peer review were used to enhance dependability and credibility (Creswell, 1998). Discriminant sampling involved posing questions that relate categories developed through axial coding and then returning to the data for evidence, incidents, and events that supported or refuted the questions, thereby verifying the data. Supplemental validation was used after writing the theory. The literature was referenced to give validation for the accuracy of the findings or how the findings differ from the published literature. Peer debriefing was accomplished by presenting analyses and conceptual abstractions of the data to two expert qualitative researchers so that inquirer biases were probed, meanings explored, and the basis for interpretations clarified (Lincoln & Guba, 1995). Two committee members were asked to consider the researcher's analysis because they were in the unique position

to be able to move between a narrowed and broadened view of the researcher's work. They were familiar with both the study's background and the researcher's biases. Discussion of the findings also occurred during the study. As categories and subcategories emerged, these committee members could confirm or challenge the researcher's analysis. During challenging, the researcher was asked to explain the rationale for decisions and synthesize all data which supported various categories and subcategories. Reflexive analysis was employed also to establish confirmability or neutrality as described by Sandelowski (1989). This served to root the findings in the informants' responses or data collected rather than the researcher's bias.

Another indicator of credibility of the findings comes from the informed clinical community, those clinicians who understand the phenomena because of their experience with it. This grounded theory account of BRCA mutation carriers and their testing and risk management experiences was presented to clinical experts and to other professional audiences.

Dependability and confirmability were also established by leaving an audit trail. Detailed recording of all research activities were accomplished. Guba and Lincoln (1995) recommend that both data and interpretations be traceable to their sources. Written records of observations, directions, and analysis were kept by memoing, using code notes, and observational, methodological, and theoretical notes. These written records were stored and managed with the NVIVO software program.

Summary

This chapter has reviewed the research method and specific strategies which were used to examine the BRCA genetic testing and risk management experiences of

unaffected BRCA carriers. To summarize, grounded theory was chosen because of its focus on meaning defined through interaction, its sensitivity to the unfolding nature of events (processes), and its ability to generate middle range nursing theory. By studying the complexities and variations of human social interaction, a better understanding of the social processes surrounding the phenomenon of BRCA genetic testing and risk management can be attained. The next chapters present the substantive theory derived in this study through the use of the analytic techniques described.

CHAPTER IV

FINDINGS – PART 1

The next two chapters present the grounded theory from analyzing interviews of 30 women who participated in this study. The substantive theory that emerged centers on BRCA testing and risk management by unaffected women with positive and VUS BRCA mutations. After careful questioning of these women and analysis of their responses, managing susceptibility to hereditary breast and ovarian cancer emerged as the basic social process of genetic testing and risk management for these women. Managing susceptibility is embodied in the proactive stance these women assumed in obtaining BRCA genetic testing and responding to prevent or mitigate the impact of breast and/or ovarian cancer on their lives. Susceptibility is viewed as emic (internal) and defined by the participants' belief that they are highly likely to develop breast and/or ovarian cancer in the future. They also perceive they have some control to prevent the devastating effects of these cancers. Figure 1 illustrates the categories and their relationship to Managing Susceptibility to Hereditary Breast and Ovarian Cancer. The components of the process, managing susceptibility, were fivefold: (a) gaining awareness, (b) confronting uncertainty

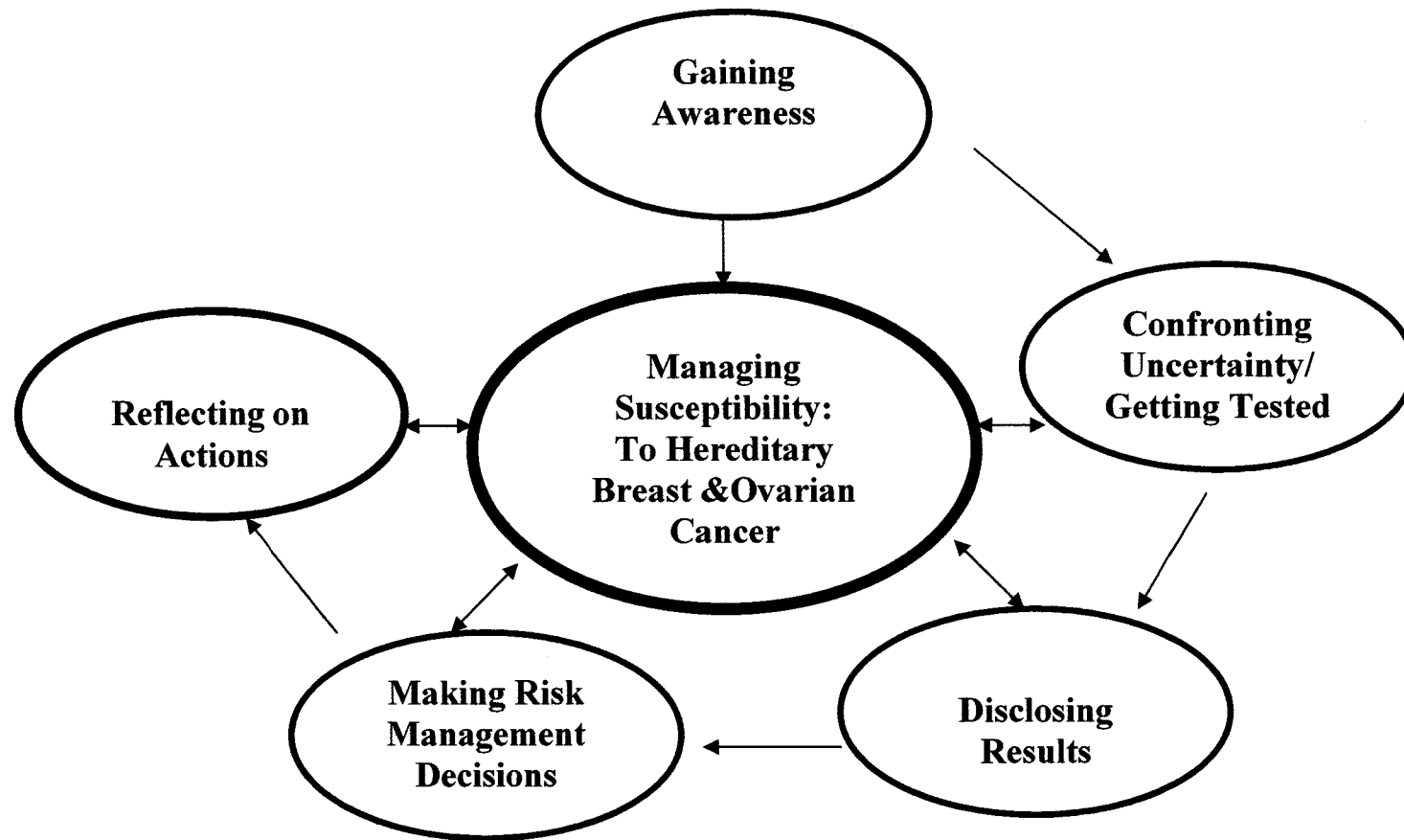


Figure 1. Managing Susceptibility to Hereditary Breast and Ovarian Cancer: A theoretical model of the process of BRCA genetic testing and risk management

and getting tested, (c) disclosing results, (d) making risk management decisions, and (e) reflecting on actions. Although the categories overlap and intersect, for descriptive purposes they are delineated separately. Each category explains the actions/interactions and consequences inherent in the process of managing susceptibility by women who are unaffected BRCA positive and VUS mutation carriers. Hereafter the two groups are referred to collectively as mutation carriers. Figure 2 delineates the categories, actions/interactions, and consequences of Managing Susceptibility to Hereditary Breast and Ovarian Cancer. This figure is presented on two pages only for readability purposes. No explicit comparison is made between the positive and VUS BRCA groups except where there are obvious differences.

These categories will be described in more detail, looking at the subcategories in each to formulate a grounded theory.

Overview

Gaining awareness was the antecedent to all subsequent action in the process of managing susceptibility for unaffected women mutation carriers. Whether these women became aware of their risk for hereditary breast and ovarian cancer (HBOC) as a child or adult, this awareness left them fearful, feeling vulnerable, and with a sense they were “playing a waiting game” for breast or ovarian cancer, which would eventually be their fate. After obtaining additional information from a family event, a health care provider, or the media, they decided to get BRCA testing to further clarify their risk and reduce the uncertainty.

Confronting uncertainty and getting tested was characterized by seeking further clarification of their family cancer history and overcoming professional, insurance, and

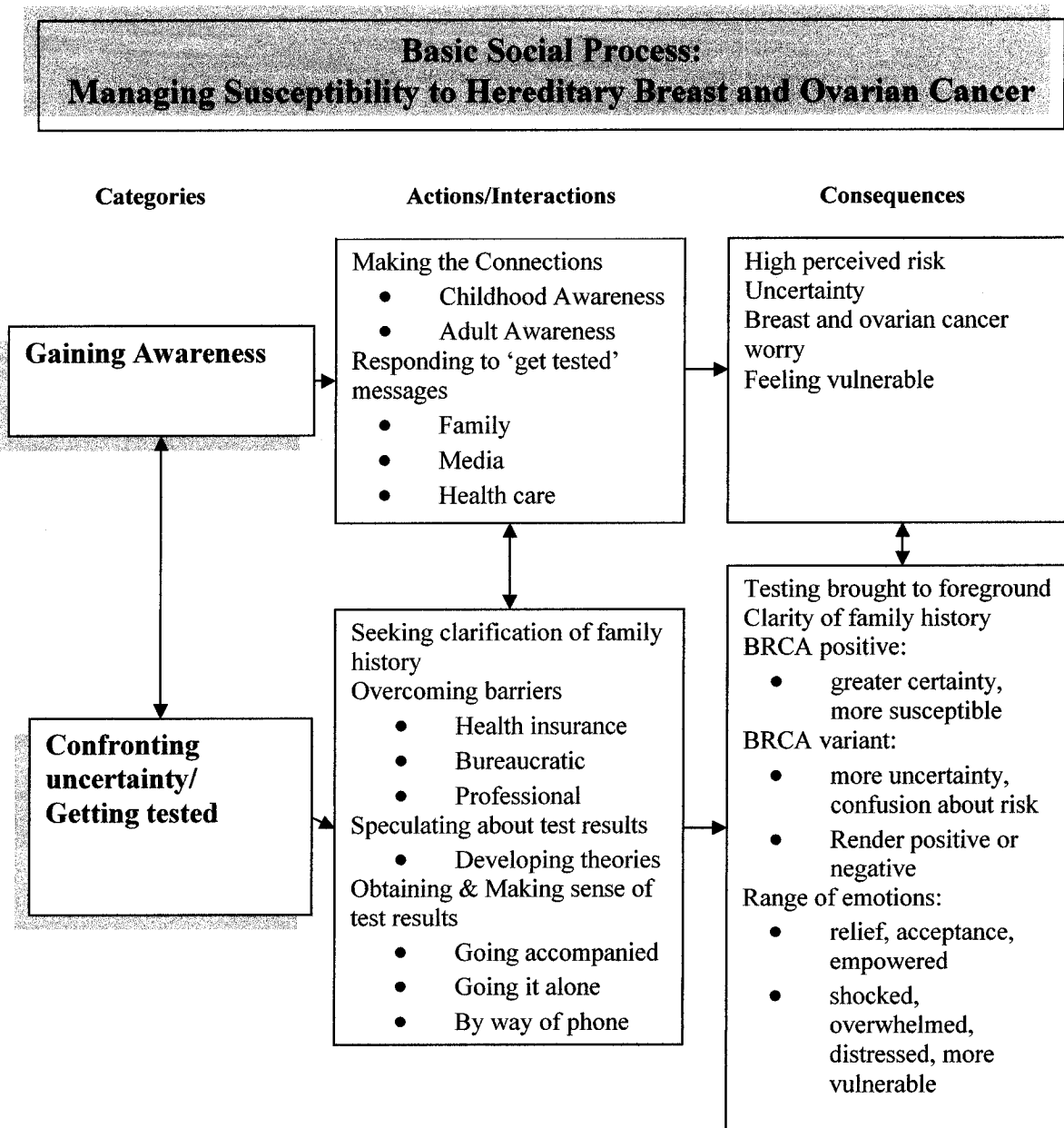


Figure 2. Representation of the first two steps in the process of Managing Susceptibility to Hereditary Breast and Ovarian Cancer. The process is continued on the next page.

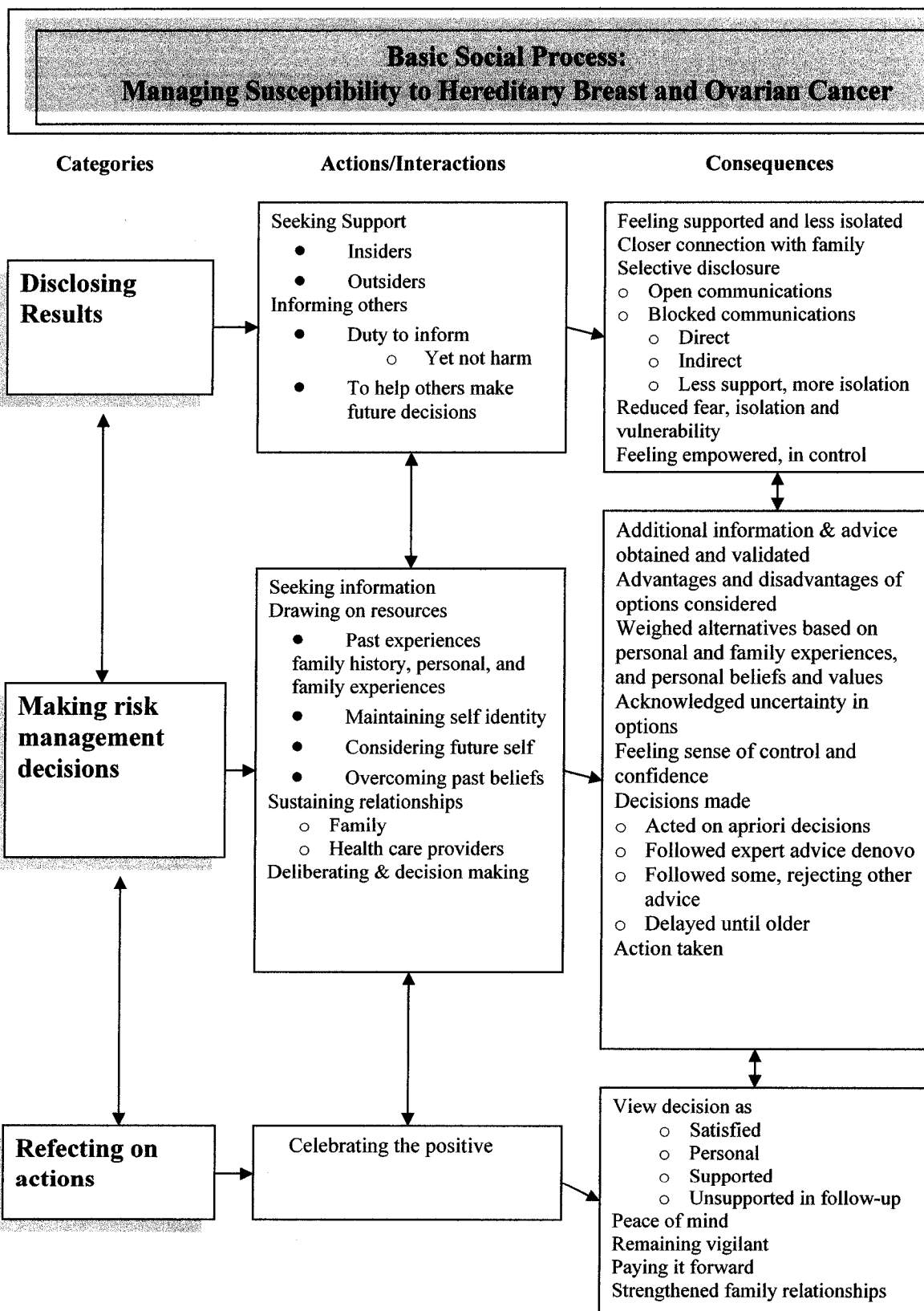


Figure 2. (Continued) Representation of the last three steps in the process of Managing Susceptibility to Hereditary Breast and Ovarian Cancer. The process is continued from the previous page.

bureaucratic barriers in the health care system. While waiting for their test results they developed theories about why they would or would not be positive. Influenced by this anticipation of results, when they received their results, they described a range of emotional reactions; from relief, acceptance, and feeling empowered to shocked, overwhelmed, distressed due to confusion and the frustration of uncertainty, and feeling vulnerable. The finality of their result, questions about management, questions from family and friends, and the prospect of lifelong surveillance were overwhelming. In dealing with these feelings, participants disclosed their genetic information to others for support and to fulfill their social obligations.

Using the words of these women to portray this integrated theory, the following sections elucidate the first parts of the trajectory of managing susceptibility in women BRCA mutation carriers. Although these categories and subcategories are presented in this order, some may be overlapping or intersecting, and should not be construed as mutually exclusive. Their experience begins with gaining awareness.

Gaining Awareness

Gaining awareness describes the process of these women as they discovered BRCA genetic testing. This category consists of two subcategories (a) making the connections and (b) responding to “get tested” messages.

Making the Connections

Childhood awareness. Most participants grew up knowing about the breast or ovarian cancer in their family, which left them feeling “cursed,” “fearful”, or “at a higher risk” than many of their friends. This awareness and the resulting sense of “fate”, waiting

for the breast or ovarian cancer to occur, were the driving forces that brought them to BRCA testing, as these participants recounted:

Feeling cursed. 32 years, BRCA 2. I grew up afraid of getting breast cancer because of my mother and her mother.... I sort of had it in the back of my mind and I was probably somewhat in denial about it and didn't really know what to do because it seemed like fate. Like one day I was going to get cancer and there wasn't a whole lot I could do about it.... And I decided that I needed to do something. I couldn't just live with this thing hanging over my head and feeling cursed.

Fear –a waiting game. A 32 year old mother with a genetic VUS. ...I've got so many horror stories with my mother (breast cancer), and ...I just knew especially since my cousin got it (breast cancer), it's kind of like a waiting game... When is coming? So ... the fear that you live with every day unfortunately.

Feeling at higher risk. 22 year old, BRCA 2. I think it goes back to... right after my mother died, I started hearing more about a lot of her first cousins being diagnosed or dying of breast cancer. And at that time I kind of thought ...it's in the family. I didn't really understand what the connection was, but I knew I was at a higher risk than many of my friends.

Adult awareness. For the seven participants without an affected mother, who did not grow up with the same fear and sense of vulnerability, a daughter's or sister's cancer, or father's test results brought them as an adult to awareness of their risk. Most of the time this lack of awareness of their breast or ovarian cancer risk occurred when the cancer came from the paternal side of the family or there was a death or divorce which caused the family to lose touch with the affected family members.

One 27 years old with a positive BRCA mutation had testing shortly after her sister was diagnosed with breast cancer. Neither of her parents had cancer. They subsequently found that her father carried the BRCA mutation and all five of her siblings had the mutation. "So when this (breast cancer) happened with (my sister), it was kind of an awakening." Another, a 30 years old whose father tested positive but did not share the results openly with his children, recounted:

It was really my mom that pushed me to do it (BRCA testing), and I started to learn a little bit more about the family history, about my dad's half sibling that had died of ovarian cancer. So that started to get me a little more paranoid about wanting to get tested.

Another BRCA positive participant, a 51 years old mother, was shocked when her daughter was diagnosed with breast cancer at 28 years and tested positive for both BRCA1 and BRCA2 mutations. This mother never knew her father, as he had left when she was a year old, and later died of alcoholism. She explained that she was not aware that breast cancer could be transmitted by her father :

And you know, it was kind of a shock because before (my daughter) was tested, I really never thought there was family history of breast cancer, because I knew no family history on my side. And I guess I had that fallacy that breast cancer only came from the mom's side of the family. And I knew there was no cancer on my mom's side. So there was kind of disbelief when she (daughter) came back carrying both of the mutations.

Responding to "Get Tested" Messages

With an awareness of their increased risk of breast or ovarian cancer, participants received messages from various sources, including (a) family, (b) media, and (c) health care providers, which stimulated their interest in BRCA testing. Reasons given for seeking BRCA testing included clarifying their susceptibility to breast and/or ovarian cancer, to learn about offsprings' and other family members' risks, and to do something which would reduce their risk for breast and/or ovarian cancer.

Family event. Sixty two percent (13) of the BRCA positive participants sought BRCA testing after notification of a mutation in the family. Two with variants of uncertain significances tested because a variant was identified in the family. These participants were encouraged by their family: mother, father, daughter, or siblings to have BRCA testing. A motivation for the testing was to receive information that they could

use, but also, could be shared with other family members. As this 25 year old participant described, her mother's testing had been done for her aunts:

After chemo and everything, her sisters ... because they're both younger and they have all girls they were really pushing her to get tested They both were of the age when my mother was diagnosed with breast cancer, so they wanted to know for themselves.

For a 22 year old whose mother died of breast cancer, it was a cousin's diagnosis of a BRCA mutation and her stepmother's diagnosis of breast cancer when she was 18 years, which provided the tipping point for her to confront her father about her family history of breast cancer. She recounted how her father initially concealed her family history due to her age, but her awareness and desire for openness influenced him to share her family's breast cancer history. Her father's influence led to her testing as she detailed:

Right after my mother died, I started hearing more about a lot of her first cousins being diagnosed or dying of breast cancer.... When I was eighteen my stepmother was diagnosed with breast cancer. So I kind of had a flashback of everything. My dad started to freak out and it was about at the same time that my dad received information from one of my mother's first cousins in regards to this testing (BRCA positive). And he didn't really talk to me about it. I kind of knew that it was there but he thought that I was too young to deal with it. I believe I was eighteen or nineteen ... But eventually I got to the point where I told him, "You know I know that this is going on. I'd rather that you be honest with me with the information that you have rather than feel like this is something that you need to hide from me". So he at that point, opened up. Showed me the whole family lineage. Showed me some of the things that I was facing.... Told me that he wanted me to get tested.... So a few years later my dad...it wasn't serious pressure, but I knew that he wanted (me) to get it done.... So I just said, "All right Dad. I'm going to go and I'm going to get it done and we're gonna find out what's going on and we're gonna deal with it."

For others, it was the combination of breast and ovarian cancer that motivated them to get the testing, as this participant related:

I guess when my mom got ovarian cancer (after having breast cancer) and somebody said, well you know, there's a link with ovarian cancer and breast cancer, and the BRCA gene, and I started learning about that. That's when we

figured, well, we really should get tested. If anybody's got a family history, it surely does look like we do.

Media. The media was also a source of messages about BRCA testing for women who perceived themselves as high risk. A 29 year old mother with a genetic VUS, described how she gained awareness about BRCA testing by reading an article in a women's magazine. She started accumulating information and prepared by getting her insurance in order, as she explained:

After she (mother) died, ... I was reading a women's magazine, and it was the story of two sisters. And she had done a diary ... (about) getting (BRCA) tested. And she explained that if you have these particular newly discovered cancer genes, that they're separate. There are sporadic cancers that everybody else gets, or could get And then there's a gene, cancer gene where no environmental factor, no health factor, nothing can really stop it, prevent it type of thing, if you have this gene. And she and her sister were tested, and how she really didn't want to believe it at first, but then her sister was diagnosed with cancer around 38. And how she went through with the prophylactic mastectomy, and her sister as well, ... and that she was going to have her ovaries removed next, and all of this. And when I read it, ... my mom died about the year 2000. I put it away in a file that I labeled research, and I just began to gather more articles and more information, if I came upon anything on the latest screening or the latest this or that, always with the intentions of being tested one day when I had constant insurance.

Healthcare Professional. Sixteen percent (5) of the participants received messages about BRCA genetic testing from a health care professional; their gynecologist, breast specialist, primary care or other physicians based on their family history. As these two participants reported, they found out during their annual check up and follow-up after a needle biopsy, respectively:

I went for my yearly checkup. And I have a new doctor, and she's younger. And she said, "(name) have you ever thought about genetic testing?" And I didn't realize that genetic testing was happening. I mean, I've heard about it, but I didn't realize that was an option for people living in (state/province). And my GP told me about the genetics clinic and made a referral.

And I believe it was in meeting with her (breast cancer specialist) about needle biopsy stuff because that happened not too long after my mom passed away.... it

was in meeting with her that she said I might want to consider doing it (BRCA testing).

Another participant who had considered participating in a university BRCA research study thought initially there was no reason to test because she was doing all she could do using surveillance. As she recounted, her physician and breast specialist informed her of mastectomy as an option to prevent breast cancer and this provided the stimulus for testing:

The University (name)... They were doing a study on Ashkenazi Jews.... I contacted them and decided at that time not to test. Because I had already been doing every type of surveillance that I thought I could do and there was no reason for me to test... After my mother died, my medical doctor ...said, "This is very, very harsh, but I think the only thing that you can do to almost guarantee is the mastectomy". And I was...appalled. I went back to my breast specialist who then talked to me about statistics and mastectomy and testing.... She kept telling me to test. That there is... that next step. And her concern was ...the cancer in my family...it wasn't they got cured and lived. It was ... they were non-survivors.

In summary, this first category sets the stage and begins the managing susceptibility process, which participants continued throughout their BRCA testing experience. Whether awareness was gained in childhood or as an adult, participants viewed their risk of breast and ovarian cancer as a looming, inevitable threat. Reasons for seeking BRCA testing included clarifying their risk for breast and/or ovarian cancer, to learn about offsprings' and other family members' risks, and to do something which would reduce their risk of developing breast and/or ovarian cancer. Participants received messages to get tested from family events, media, and/or health care providers. Viewing this as an opportunity to reduce uncertainty about their risk, they proceed to getting tested.

Confronting Uncertainty And Getting Tested

Once participants had additional information from the media or confirmation of a higher risk status from family mutation results or health care professionals, they confronted their fear and decided to get tested. Hope of reducing the threat of developing breast and/or ovarian cancer and to reduce uncertainty about their risk were the main motivators for BRCA testing. As this participant explained, testing provided the knowledge to empower her to manage her risk:

As soon as I found out that there was a gene in the family and that it was testable, I knew I needed to know. It was...for me the decision to test was not a hard one. It was the decisions afterwards that were hard. To me knowledge is power. I know it's a trite phrase but it's...not trite.... I just didn't want to not know and be speculating about what my risks were. I thought if there was information out there that could make it clear to me whether I was or was not at high risk, I wanted to know that.

Participants described four steps in the process of getting BRCA tested (a) seeking clarification of their family cancer history, (b) overcoming professional, insurance, and bureaucratic barriers, (c) speculating on test results, and (d) obtaining and making sense of test results.

Seeking Clarification Of Family Cancer History

Those participants with a known BRCA family mutation came to the BRCA testing process with a good history of their family's breast or ovarian cancer, because it had been worked out previously by a mother, father, sister, aunt, or cousin. However, those first in the family to be tested, that self-referred or were referred by a physician, often needed to obtain more information from family members to determine specifics of their family cancer history. As one related, "That's when I started contacting relatives and trying to put together a family tree". Sometimes this included contacting those

estranged from the family. One participant described how she overcame “awkward” family dynamics, those who were “emotionally distant”, and feelings of guilt to obtain the family cancer history that she characterized as “not polite conversation”:

It was all this kind of vague, like well there’s a lot of cancer in my family, was about how I could sum it up. And it forced me to make contact with people who I hadn’t been in contact with in a long time and do research and actually talk to my father about some of this stuff.... Then they all suddenly died after I’d been out of touch with them for seven or eight years, and I had all these feelings of guilt and just weirdness.... So it was just... getting in touch with ...trying to find people to contact in the first place.... But it was just ... awkward stuff to talk about. To call up people you haven’t talked to in ten years and say, “So I want to know what kind of cancer he had”. And it doesn’t do to just say, “Oh, he had cancer.” I needed to find someone to tell me where this cancer was and that kind of thing. “I need...do you have an autopsy report?” Like just these things that aren’t polite conversation, especially with people who you’ve grown away from and who might perceive you as having kind of ignored them and blown them off for many years. So it’s mostly family dynamics stuff was awkward.

Overcoming Barriers

Some participants encountered external barriers that had to be overcome before obtaining genetic testing. These included health insurance, professional, and organizational/bureaucratic barriers.

Health insurance. Because of the cost of genetic testing (comprehensive test \$2950, multi site (3) \$450, and single site \$350) some participants had to get their health insurance in place before they could pursue testing. As one noted, “Once we got under this health insurance, then I went ahead and began to pursue talking to my new primary doctor about this (BRCA testing)”. Seventeen (57%) participants had insurance and used it to cover BRCA testing. Eight (27%) chose not to use their health insurance to cover testing for confidentiality reasons and paid out-of-pocket. Four (13%) had testing in a research sponsored program and Myriad Genetics paid for the testing of one (3%) participant with insufficient funds.

Despite having health insurance, two participants encountered problems getting their health insurer to pay for BRCA testing or surgeries. It was their assertiveness, perseverance, and Medicare changes regarding BRCA testing that helped them to win in the end. As these participants recounted:

They (insurance company) fought quite a bit and I fought back and I appealed it and they said, "We're not paying." And even after I did it they said they would pay and then they started, "It's done in a specialized lab." and they said, "This lab is not under our insurance." But I said, "This is the only one in the country that does it, you know, I took the blood here at (name), which is your clinic. But they have to send it. What can I do? They don't do it here" and, yeah, it was a big thing with them. ... at some point, somebody would have probably given up, but I didn't. ... I woulda probably paid it from my money but I just felt ... that you should do it. This is part of the testing that I should have.

(I) put off testing for 3-4 years due to insurance not paying. When I went to have the genetic testing, they told me they did not cover it, period, flat out. And that's one of the reasons I actually put off testing for probably three or four years, because I knew it was going to cost two or three thousand dollars. And then after my mother moved here...about a year and a half ago, we found out Medicare would cover her testing. I'm like, "let's you go ahead and do that, because that's covered. Then that will tell us ...where we want to go from there".

Bureaucratic. Another participant described "bureaucratic run-around" and "roadblocks" to testing created by her physician that caused her to delay testing until she moved:

I began to approach ...the idea of testing with him (OB-GYN physician), given my family history.... So I started approaching this and he said that it would be very difficult to get genetic testing, and I think that he probably thought that I was cancer-phobic at that point, just because of the family history. And he recommended that in order for me to get testing, I would have to cull my grandmother's records, my mother's records, I would have to get letters from their physicians, I would have to see a psychiatrist and the psychiatrist would have to say that it was in my best mental interest to have this test. I mean, it was incredible. And I thought that he was throwing roadblocks in my way on purpose, just giving me the bureaucratic run-around and fortunately, after that we moved to (city where the testing was easier to obtain).

Professional. Others encountered professional barriers, such as a physician who had not heard of BRCA testing, as this participant who inquired about testing in 2004 reported:

She (primary care physician) had never heard of it (BRCA testing) in her life -- kind of scary when the doctor hasn't heard of what you're talking about, and she directed me to call my insurance company... directly, which I did.... Finally I got in touch with a woman within the health insurance who did know and said that yes, they set up counseling and testing.

Speculating About Test Results

After clarifying their family cancer history and overcoming barriers to testing, participants who met their genetic counselor's or physician's criterion for high risk, had their blood drawn for testing. While awaiting their BRCA test results to come back from the lab, 23 (76%) participants speculated about what their results would be. Of those speculating, 17 (74%) thought they would be positive, 5 (22%) thought they would be negative, and 1 (4%) thought she would get VUS results. Participants developed theories about why they thought they would or would not be positive, such as (a) physical characteristics, (b) age, (c) family history, (d) luck or chance, and (e) degree of fear. By speculating about their results in advance, some participants were able to rehearse what it would be like as a BRCA mutation carrier and considered how they would manage their susceptibility.

Physical characteristics. As one participant who speculated she would be positive (and was) reported, "Since the day my sister got sick (breast cancer), I've always known it (my risk) was high. Yes. I figured I had it (the BRCA mutation)". When asked why, she responded, "Nothing concrete... I look like my sister, I'm built like my sister. It just seemed like genetics would carry through". In contrast, one who anticipated she would

not be positive like her sister, explained, “My sister and I, we’re just so different in character and the way we look and everything. So this was going to be in common? It just didn’t make sense.”

Age. For a 46 year old participant who thought she would be negative, age was the reason, she “had passed the age in which my sisters developed their cancer.”

Family history. Others thought they would be positive because of their family history: “And just because of my family history, I’ve always suspected some type of genetic relationship.” Another elucidated, “There was no doubt in my mind I’d be positive. My family history. Too many young women (with breast and ovarian cancer).”

Chance. A 27 year old speculated she would be positive because all 5 of her brothers and sisters were positive. She explained she would probably get retested if she got a negative result:

I would have been very, very, very surprised and probably would have got retested or I don’t even know if I would have ever been okay with the fact that I wasn’t positive, because I just would have been so surprised.

By way of contrast, a 54 year old who did not understand at that time how BRCA genetic mutations are inherited, speculated she would be negative because she thought there was little chance that “two sisters in the same family would come up with the mutation.”

Degree of fear. A 52 year old with a positive mutation speculated she would be positive due to her closeness to her mother’s death and her fear, “...I just had this gut feeling that I had the mutation. Because when our mother died... I’m the one that kind of watched all of this. So I’m the one that had more fear.” A 47 year old with a positive mutation who speculated she would be positive described “a sense of doom” and felt she would not live past 50 because her mother didn’t.

For those who did not speculate on results, one explains simply that she just didn't have a feeling one way or the other:

Until the moment she (genetic counselor) told me, I continued to think it was a flip of a coin. It could go either way. I hoped it would be negative, and I knew it could be positive. No, I never had any kind of feeling that I knew what it was going to be.

Obtaining and Making Sense of Test Results

About three to four weeks after testing, participants obtained their test results either in person or over the phone from their genetic counselor, physician, or geneticist. Some figured out their results before their genetic counselor or physician explained it. They figured out intuitively, based on the presence of certain people in the room when the BRCA test results were disclosed, and from inconsistencies in the way family members who tested together were given their results, as these participants related:

I got a phone call and they ... said, "We need to change your appointment." And I thought, "Hmm, that's kind of bizarre." And so my husband came with me and ... we were waiting The nurse said to me before we got into the room, "Do you mind if so-and-so observes because ... (name of new genetic counselor) will be taking over (current genetic counselor)'s spot when (she) has her baby." And I said, "Sure." So we walked in and this woman that I'd never met before comes to shake my hand. And I looked at her and I said, "Oh you must be (name). And she said, "No, I'm (name). I'm a psychologist." And I looked at her and I said, "Oh, I'm positive. I know ...my test results. I know I'm positive". And I thought, "Why else would they have this psychologist there?" And that was probably, I assume, why they postponed the appointment. Cause the psychologist couldn't make it at the original time. So we sat down and I said, "Look, just tell me, is it good or bad? I don't want any extra information.... Don't...be setting the stage. Just give it to me straight." And she said, "It's bad." And so ... my intuition was confirmed.

I think what happened ...the genetic counselor who did our testing, forgot that we were sisters. So when (sister)'s negative results came in, she (genetic counselor) was so excited she told (her)... So when I didn't get my phone call, I knew. ...she did call me and say that ...she wanted to see me. But I knew. And I said to her at that point when she called me, "I know that I'm positive."

Yet another participant knew when her pregnant genetic counselor came off maternity leave to give her the results:

I went in sort of knowing that ... it was positive. I know (genetic counselor) didn't intend for it to be. She was on maternity leave ... And when it came back ... (genetic counselor) wanted to come to meet with me and I said, "Nobody comes from maternity leave to give me a negative answer. This must be positive." ... I mean, I sort of figured one plus one.... I asked her, "Why did you come?" ... She was so wonderful. She said, "I started this with you. I want to also talk with you ... after the positive result."

Before receiving their test results from their genetic counselor or physician, participants made the decision to get their result in person or on the phone, and if in person, to go accompanied, or alone.

Going accompanied. Sixteen women (53%) had a family member (sister, father, husband/partner) or close friend accompany them when they obtained their results. A family member/partner's emotional support was very important at this time as this participant recounted, "My husband, he also was surprised and very supportive and has been, when I kinda crumbled a bit there hearing the positive results". Another example of a husband's support and what it meant is described in the following quote:

And my sister and I went in together. And also, ... I just knew I needed to have (my husband) there. And my sister didn't have a husband there. And I think, "Oh, gosh, thank goodness she was negative." Because I'm just so thankful that (name), my husband, was there to kind of go through that with me as well, because he got an opportunity to talk to the genetic counselor, as well as talk to the geneticist. And I mean, it was a huge shock.... I'll never forget her first line... And she just said that, "Your results are different".... And she looked at me and said, "(participant name), you do." And my husband and I just kind of clung to each other and cried. [voice breaking] It very much felt like a cancer diagnosis. I had to really remind myself that I don't have cancer....

Going it alone. Eleven (37%) of the participants were unaccompanied when they received their results and three (10%) received their results over the phone. One participant, whose husband was in a foreign country on business when she got her

positive results, explained that being alone was not problematic because she liked being self-reliant and relying on her inner strength:

Another participant expressed a similar self reliance: So, I was there on my own. But I liked that. I like getting my inner strength going sometimes when you're together you sort of rely on people, and although we have, I mean, we're together more than probably twenty-five years, but I don't know... there's some things you have to feel and do on your own, so I was sort of, I don't know if happy, but it was fine for me, him not being here.

I'm not a real emotional person. I don't get real stressed out, so I wasn't worried and felt like I needed support, like I think some people might ...even if she says there is a problem, I'm pretty level-headed. We'll just deal with it, so I didn't take support or anything and I just went to talk to her (genetic counselor).

By way of phone. Those who received their results by phone had mixed reactions.

One participant described it as "horrible". But when asked if she would choose to receive her results on the phone again she replied, "Yeah. ... because driving over there would be just dragging it out." She explains her phone experience:

Oh, she phoned me with them. I believe we agreed ahead of time that she could phone us with them. It was horrible. I was getting ready to take the kids to school and we were rushing out the door and the phone rang. And it was just...very overwhelming.

Another would prefer to talk with a counselor in-person, but it was the circumstances at the time and her desire to know that took precedence:

So I couldn't go in (to the clinic). I was going to be at (place of worship) all day, and there was no way that I was making it in there, so it was either receiving the results on the phone or not receiving them at all until the following week. I wanted to know. So given the situation, yeah, I would do the same thing again, but if I had the ability to talk to someone face to face, I definitely would do that.

This participant recalls how she received her results in her car on her cell phone.

This reminds us that timing and safety are important considerations. She related her experience:

I was in my car driving and I got the phone call. And she (physician) ... just was very, very like dramatic and almost like to the point where it was humorous a little bit. And she ...got on the phone, ... “you're going to be okay, you need to be sitting down”, and ...she told me that if I was driving, I really should pull over and maybe it wasn't a good time to talk.... at first I was kind of was like, “Okay, here we go, I definitely am positive.” ...I did pull over because she kind of ... forced me.... And she just said, “I'm calling to let you know that you're positive; your test results came back”.

Reactions to positive test result. Participants receiving positive BRCA test results described a range of emotional reactions: from relief, acceptance, and feeling empowered, to shocked, overwhelmed, distressed due to confusion and uncertainty, and feeling vulnerable.

Like several of the other positive carriers, these three women with positive mutation results described how having the positive result gave them a sense of “relief”, “it was an absolute gift to know”, as they had more certainty about their risk, and it empowered them to do something about it now:

Almost relieved. 32 year old. In a way, I was almost relieved to get a positive result because it was... I knew that having no specific mutation to test for, if I got negative results I'd be in the same boat I'd been in all along. And I wouldn't know if there was an unknown mutation that I had or if there had been something that my mother had had that I had managed to not get or what was going on. So in a way it was almost a relief to get some solid information about where my risk was coming from and what I could do about it.

35 year old. And part of me almost hoped they were positive, because then I could get the surgery and not have to worry about anything anymore. Even if my results were negative, I still would have felt that I was significantly at increased risk over an average woman.

Empowered. I had to really remind myself that I don't have cancer. So there was that. I had to also balance it with I do feel it was an absolute gift to know. And that's how my husband feels as well. Because we can do something about it. I don't have to die of cancer like my mom and my sister did.

Others were “shocked” and felt vulnerable. This 45 year old participant who thought her test results would be negative explained feeling shocked:

And so when I got my results back ...I kind of didn't think I was gonna be positive. I really thought it would be negative.... I remember my breath skipped, ...I just kinda went (gasps). It just took my breath away. I was just so shocked and I cried a little bit right there in the office.

Another a 41 year old who though she would be negative reported feeling shocked and overwhelmed. She described a delayed response to her positive test results and feeling "lost", "scared", and "angry", like working through the stages of grief:

The impact of the test results didn't hit 'til three days later. I was just, like, in shock. And I was on autopilot. I think I asked all the appropriate questions. I remember at that key meeting, that I was there with my husband, I finally had to say enough. I don't think I can take any more information.... And they very quickly respected that and said, "Ok". ...That was real helpful, but when you're first told that, especially like me...convinced that I did not have it (BRCA mutation).... And it just turned my life upside down. And I can remember so clearly watching a movie with my family three nights later.... It hit me so hard... that feeling ... I just had to leave the room, because it just all hit me then. Lost. Feeling really scared.... There's a point later where you just get angry, pissed off type.... And I think you're working through it like you do the death process. Go through those stages of grief.

For some, testing increased their sense of vulnerability. It provided an awareness that not only was their high risk of breast cancer confirmed, but the mutation conferred a high risk for ovarian cancer as well. This 32 year old participant described her "insidious" new worry:

However, it was also a big eye-opener.... I had no idea about the ovarian cancer link. That's something I'd never worried about.... And so suddenly I had this whole new sort of insidious cancer to worry about, even more so than breast cancer.

Reactions to variant of uncertain significance results. Among the nine participants with genetic VUSs, their interpretations and responses to their BRCA test results were very different. Some rendered the results problematic, like a positive result, and some did not. Some accepted the limitations of this test and said they had been prepared for the outcome. Others were disappointed due to their uncertain position with

regard to the magnitude of their risks of developing cancer and risk management. For clarification, in all cases, the BRCA test results documentation provided by participants indicated a “variant of uncertain significance”. Some had sought follow-up with Myriad Genetics, but none of the VUSs had been reclassified as deleterious mutations.

Rendering it positive. Some participants with BRCA VUSs were quite distressed by their results, because instead of the certainty they were looking for with this additional genetic information, they were left in a state of confusion. The hard part was “not having a definite answer”. Some felt that even though they were not positive for the BRCA mutation, they would still get breast cancer, as one reported below:

There was no question or not whether I would get it (breast cancer). And honestly, I still feel that way. I know I don't have that mutation, but also they haven't found, you know they know that there's another spot in that DNA, it has to be in.

One 34 year old, who was aware that a VUS result was a possibility during BRCA testing, expressed her confusion, frustration, and hopes for scientific advances “before it becomes an issue” for her. Although her genetic counselor “tried to emphasize the fact that ... it did not equal a yes result” she construed that it did equal a positive result, that way she would remain vigilant as she explained:

I was confused by the test results.... But I hate the fact that I don't know anything else more than I did before I went and did the whole thing. Very frustrating. So I think I'm just hoping that science will catch up with it before it becomes an issue for me, which I don't know if it's a realistic hope.... I have decided ... not to torture myself mentally, but that the healthiest interpretation that I can adopt is to go ahead and equate the variant as a yes, you tested positive for this gene essentially, so that I can do everything that I can and not slough off.

Although five (56%) participants with BRCA VUSs interpreted their results as positive, despite the fact they had been counseled that the results were uncertain, some explained that their physician had rendered the VUS problematic and “considered it

positive”. A 29 year old, who did not have genetic counseling prior to testing in a primary care setting, and was not aware that her test result could be a variant of uncertain significance, explained her confusion and how her physician rendered her VUS result positive:

So now the results are back. He (the oncologist) looked at me and looked at the chart and said, BRCA 1, nothing is detected. However, unfortunately I remember specifically him using the word “unfortunately on BRCA 2, there is a variation” -- and that’s the first I heard of that word, ‘variant of uncertain significance’. He said there is basically something different about that gene in my DNA panels. And he gave me... a little leaflet they had that said, “What does variant of uncertain significance mean?” And I let him know that I still didn’t understand what he’s telling me... that this is not positive, but it’s not like my BRCA 1, where there’s nothing detected. There’s something, but yet it’s not significant. And he said that based on my family history and this coming back the way it was, he considered it like a positive result. He really felt like now let’s proceed forward. He asked me who my gynecologist was and that he was sending my results to the gynecologist and wrote a note on my chart about the oophorectomy consult But it’s changed everything, because the test result isn’t clear. So it’s changed my life in the sense of without that clear result, I really feel confused a lot more than I think I would have with the positive result.

Negative rendering. Four (44%) with BRCA variants of uncertain significance did not interpret their results as positive and rendered their results less problematic. These participants explained their interpretations and reactions:

57 years old. It didn’t bother me at all to be honest. Finding out I had it (variant of uncertain significance), I’m a realist So finding out that my body may have a flaw is likely, not unlikely.... And I told my husband.... It didn’t phase either one of us that much. I wish I would have had more. I tried to find out... I wouldn’t have reacted like my sister. I know that. When she found out she was just really frightened, and maybe because I was older when I found out that I had it, I’m not sure or maybe it’s just we are different personalities. It just didn’t phase me one way or the other.

49 years old. Only I guess surprised that they found a variant, but then “We haven’t seen it before”, they said, “and we don’t know”. So it’s kind of like wait and see. So it’s been five years and I had a doctor fax over the report and checked with the lab a year ago..., and so far they have no further knowledge if that causes cancer or not.... I was fine with it. Just taking care of yourself, I guess, regular checkups and always being aware that you have a history ... that I’ll be fine.

She (genetic counselor) explained to me that it was really not a result that we could determine whether there was additional risk or not because it was, what did she call it? “Undetermined ... that there was some variation but it wasn’t the typical variation that we see with increased breast cancer risk”, so I thought, “Okay. so what do we do now?” There really isn’t much that you can do based on that.

In summary, getting tested encompassed seeking clarification of their family cancer history and overcoming such external barriers as health insurance, professional, and bureaucratic obstacles. In preparing for their test results, participants speculated based on theories they developed about whether their BRCA results would be positive, negative, or inconclusive. Influenced by this anticipation and rehearsal of results, when they received their results they experienced a range of emotional reactions: from relief, acceptance, and feeling empowered to shocked, overwhelmed, distressed due to confusion and the frustration of uncertainty, and feeling vulnerable. In dealing with these feelings, participants disclosed their genetic information to others seeking support and fulfilling family obligations. The next chapter will explore this disclosure of BRCA test results and how they made risk management decisions.

CHAPTER V

FINDINGS – PART 11

This chapter will focus on those components of managing susceptibility to hereditary breast and ovarian cancer that come after BRCA testing. These last three components are (a) disclosing results, (b) deliberating and making risk management decisions, and (c) reflecting on actions.

Disclosing Results

Seeking Support Through Disclosure

Shortly after receiving their BRCA test results, participants embraced the hard part, the feelings of “fear,” “sadness”, and “shock”, which brought the possibility of getting breast and/or ovarian cancer to the foreground, as these women recounted:

And I was like, “Well, I’m a mutant. It’s true. It’s real....” And it probably wasn’t for a few days until I sort of, I guess embraced or dealt with the hard part. The sort of fear and the sadness. It’s like ... I have this thing that could give me cancer and I really don’t want cancer.

It... all hit me like a ton of bricks. It’s like, “Oh my God, what am I gonna do?” ...and then that’s when I just, I had this sense of urgency. I mean...there will come a point where you just feel like a time bomb.... you’re sort of shocky at first and you don’t even realize the magnitude of it all immediately. I don’t think that you can.

The finality of it, cost of testing, questions about management, time absent from work, what they should tell their children, questions from family and friends, and the prospect of lifelong surveillance was overwhelming. Questions about the future were on their minds, as this 41 year old participant related:

...and then your thoughts just race, you know, what does this mean?... for my daughters? ...for my husband? ...for me? What is it? Who do I tell? Who do I not tell, like I don't know, just a mishmash of things.

In dealing with these feelings, participants disclosed their BRCA test results to others seeking “support” from family, friends, and peers. As one participant explained, “Someone’s gotta be in your corner to sort of have that little lifeline, at least for that first week or two.” They also selectively shared their BRCA test results to inform others so “they could take action” or “be more aware”.

Seeking support. Participants of all ages disclosed their test results to their husband or partner, close family members, and closest friends. Later on they selectively disclosed to extended family and a few disclosed it to employers, if they were planning surgery and would need time off from work. As this 25 year old single participant related, “The day I found out, my three closest friends, I really surrounded myself with them ... they were very supportive.” She further explained how she selectively disclosed her test information to immediate family and friends and her mother told extended family:

When I told my father - I might have told him the next day or a couple of days later. I told him in person. I told my stepsister ...I think in person, maybe on the phone. I told her a few days later. I didn't tell my stepbrother; we're not really close. My mother told my family. Well, we didn't tell my grandmother. But my mother told her sisters, and I guess my stepfather told his family. I really only told my immediate family and my friends.

In a deliberate and thoughtful manner, they disclosed the information to remaining family and friends in person, by phone, sent letters, and some found electronic

mail (e-mail) useful to relay the same information to several people simultaneously. One sought the assistance of a genetic counselor to help draft a letter for extended family that did not live close by. Another with a genetic variant of uncertain significance (VUS), explained a carefully considered strategy of first alerting her aunts of a possible HBOC risk by letting them know she was having the BRCA testing done. She then looked for an opportunity within the normal processes of her family life, a Thanksgiving family gathering, when she felt they were prepared:

Yeah, it was a family meeting of my mother's sisters. A lot of them knew that I was going to have the test.... And they all knew that I was coming for Thanksgiving, so when we went home after dinner, we were sitting around talking, and I said, ...“Well, why don't we talk about this now?” because the children were outside playing, and it was a good time. So we just talked and I told them, “You know I had the test and this is what my results were.” Told them what it meant or as far as I understood it meant, what the geneticist said it meant, and we kind of just talked.... And I was kind of like, so what would you guys do if you were me? And all of them were in agreement that they would try to do whatever the doctors thought would be best, the best step for prevention.

Most reported positive aspects of disclosing their BRCA genetic test results and overall found their family and friends supportive. Participants expressed a closer connection as a consequence of disclosing their results, “I think we're probably more supportive of each other” and “Maybe made us a little bit closer because we have a common problem. I mean, they actually don't know if they have the gene, but there's the potential”. For some it clarified why there was cancer in the family, as this participant related:

My mom's family was excellent; they just were really supportive. And it kind of answered some questions for them as to... why we were going through this stuff (cancer) with our family. And a lot of them didn't know that it was genetic.... So it was good.

A participant with a variant of uncertain significance explained telling friends, “I could tell them and I could have the support that I needed.” She described how she needed to get her true feelings, the frustration and “gloom and doom” about the VUS out of her system before she could move on. This she was able to do with her friend, but she could not be as honest emotionally with her father and brother.

One friend in particular, I just really was upset. Kind of reviewed the fact that (genetic counselor) had said it wasn’t really all doom and gloom because we couldn’t be 100 percent sure that it was a disease causing variant, but that I was sure that it was a disease causing variant, because it just had to be because my mom had breast cancer... And I think I was a little bit more at ease to share my true feelings, gloom and doom reaction.... Whereas my dad and my brother were so really good and mature about it. And I wouldn’t change the way they reacted. It was a very healthy reaction, really smart. But I think I needed to be dramatic about it and kind of get it out of my system before I moved on. They were supportive and they were sympathetic... and really did all they could.

There was a greater degree of support, a connectedness, felt when members of the family were going through the genetic testing together. They tended to have a family support network or “group up for bad news” as these participants explained:

I think in the family... we’re a very close family and we were all going through it at the same time, so it wasn’t any more “Oh poor, (name)” as opposed to “poor (my other sister)”.... It was sort of a group thing.... We usually group up for bad news.

I’m really grateful and thankful that I have other brothers and sisters that are in the same exact spot as me and that we can talk about it and stuff.

Finding insiders and outsiders. Not all the women seeking support by disclosing their BRCA genetic information found it. In telling others, participants encountered two responses from others: that of the insider and the outsider. The insiders understood about the meaning of the mutation and tended to “get it” - this was usually someone who had the experience of a mutation or a VUS. The outsider did not understand what the mutation meant to the participant and thus the support was lacking.

Outsider. As one participant described, her brothers responded less on an emotional level, she felt “they didn’t quite get it.... They just couldn’t quite relate to what it must feel like for me”. She said some people just “didn’t understand” when she explained it. Another participant experienced a good friend as an outsider:

And I guess it was hard, because I was trying to explain a lot to her, and she totally was not understanding ... because ... like going from my family who everybody is kind of involved in it, and then trying to tell somebody who is like on the outside, it was difficult. I don’t think she understood how serious it was when I was telling her ... 80 to 85 percent chance of getting breast cancer. She was supportive, but ... I don’t think it did a lot for me.

Insider. Most of the participants had some insider family members to whom they could speak freely about the hereditary breast or ovarian cancer in their families. Often this was a mother, sister, aunt, or cousin who shared the mutation. They talked about their fears of breast or ovarian cancer, passing the mutation to their children, and their experiences with risk management options.

Although surrounded by family and friends, there were times during the testing and risk management they felt alone, “isolated”. Disclosing it to other insiders who understood helped them to feel they were not alone in this, as one participant recounted:

I think there was just the sort of psychological component of not feeling isolated. Of feeling like, ...there were all these people out there (with a positive mutation on FORCE website), ... no matter how much your loved ones and friends care about you and want to understand, it’s just different to have people who are like, yes, like I am, who grew up, often many of them grew up afraid of cancer, the way I grew up afraid of cancer.

The insiders had a common ground of shared experiences and concerns and provided a sense of connectedness which helped the participants’ fears and isolation to dissipate. When they expressed their fears, they found someone who would listen with compassion. These insiders helped to legitimize the participants’ feelings and provided a

comfortable environment where they could speak out about how they felt. Participants found insiders in a chatroom on a BRCA support website, Facing our Risk of Cancer Empowered (FORCE), with the Hereditary Breast and Ovarian Cancer Organization in Canada, and the National Ovarian Cancer Coalition, as well as in hospital-affiliated support groups. One participant explained the personal significance of her mutation and the difference between the support of her husband and the women of FORCE:

And then my husband would say, ...“you’re no different today than you were yesterday (name)”. And it’s like, “You don’t get it. Yes I am”.... See, because it was real personal to me that way ... I think he was just trying to sooth me and to be there for me. I think there’s also a point in this whole process where husbands, as supportive as they all are... I guess more to one degree or another some are, but, you’re at a place where, unless you’re in the same boat, you don’t get it. You can’t possibly understand. And that’s the beauty of FORCE because they all get it.... and I don’t need anyone to ever tell me that they understand, because I know that they just can’t.

Support and empathy were unique benefits participants found with their relatives or support group peers who shared the BRCA mutation. They offered a special service that could not be duplicated by genetic professionals or non-carrier family members. Although they tried to use friends and family as a sounding board for their feelings about having the BRCA mutations and the risk management options available, several found that what they really needed was to draw on the strength and support of women who had been through similar soul-searching. These were people who could understand and share their fears of getting cancer and who knew about the risk management options to prevent the development of breast and/or ovarian cancer. They also shared the impact of their management decisions on their lives and understood issues such as employer attitudes about employment, and health and life insurance concerns.

However, few of the participants with BRCA variants of uncertain significance were able to find such support in their families (because with two exceptions they were the first in the family tested) and peers. Thus a few attended a high risk support group and others sought groups with BRCA positive carriers. Through these groups they were hoping to discover further developments about their VUS results.

Disclosing to Inform Others

In addition to disclosing their results to obtain support, participants wanted to inform others about the BRCA mutation. Underlying this disclosure to inform was the hope the information would stimulate relatives at risk to test for the BRCA mutation or to take action and be more vigilant about screening for breast or ovarian cancer. Other times it was to increase others' awareness about HBOC in general.

Duty to inform. Participants who were the first in the family to be tested, experienced a sense of obligation and responsibility, "a duty" to inform others in the family who could possibly share the mutation, for the other's personal sake and the sake of their children, as expressed by this participant:

So that's something that I feel so strongly, that I have a duty. It's not just whether or not I want to. It's I have to. And I have to do it in a way that's gentle and try to give the information, but allowing them to make choices.

She detailed how she assessed receptivity and fulfilled this duty to explain the BRCA information to her maternal cousins:

My mom's cousin, I called her on the phone. And I start off slowly. I just say "...I have some information about the breast cancer in our family... And is this something that you'd like? Can I tell you about it?" You know, to ask their permission first. And then I just tried to do it very gently and let them know what I've done and just again how wonderful the genetic counseling has been and how informative it is.

Implicit in their message was the need for the family member to find out their risk, to minimize their chances of developing breast or ovarian cancer:

When I'm passionate about something.... and I feel that it (testing) somewhat enlightened me, then I'm pretty good about saying, ... this is what I did and this is kind of what I got from it and maybe you should think about it also.... You don't want to push it on anyone, but you would just kind of like to put a thought in their head.

At first it (disclosing BRCA information) was ... just to my immediate family. My sister, my husband, my mother-in law, and then it was every member of my family who would listen. That was after ... maybe about six weeks when I started calling my family members. And telling them... that I tested positive. And, that I thought it was in their best interest... to test.

In addition to communicating their BRCA test results to their family, some wanted to be a resource for others, as they wanted as many people as possible to know about the mutation. One participant was on a mission and said she told "Anybody and everybody that would listen, because it's important for everybody to understand what it's all about ... it could be in other families". Some shared it by writing and publishing their story, speaking to lay and professional groups, and participating in chat rooms and in research. They hoped in this way someone in the future makes the connection with the genetic mutation as the possible cause for their familial breast or ovarian cancer. By being open, they hoped also to reduce other women's fears of testing due to concerns about discrimination.

Duty to inform, yet not upset. All who felt the duty to inform family members indicated it was a sensitive issue and needed to be handled carefully. There was a tension between their duty to inform their family members of their risk status and respecting their needs. As one participant explained, "You can't underestimate how complex the issues are in terms of family relationships and how it has tested family relationships". As

detailed below, she feared she had pushed her sisters away. This resulted in them being less open to talking about the BRCA mutation and increasing her sense of isolation:

My sisters have been very supportive of me. But our way that we deal with things are very different.... my oldest sister hasn't tested. My fourth sister tested after quite some time and kind of without telling anyone. And I've had a hard time accepting ... Like in my mind you do whatever you need to do to make sure that you stay healthy, and you gather all the information and it's pretty proactive I think. That's my way I wanted to deal with this.... And it's been very hard for me to accept that my oldest sister, who I love dearly, is terrified of being tested. So I try to give information.... I wonder sometimes if I've been pushing. And I've had to back off, because I think maybe I've pushed them away a little bit, so that they're not as open to talking about it with me. And I don't want to do that.

Another explained that pushing the issue on family can create a rift. So she learned to “kind of lay low”:

You run the risk of creating a rift in a family when it's an issue for you and it's not an issue for them and you want to make it an issue for them and they don't want you to make it an issue for them.

In disclosing her positive results another described that she sent “a shock wave through the family,” but was doing a necessary service for them. One who felt like “the grim reaper” describes that telling her extended family was one of the hardest parts of her BRCA testing experience. It was a balancing act, she wanted them “to see the importance” of testing, but at the same time she did not want “to step over the line,” as she recounted:

It (telling family) was absolutely horrible. I felt like... the grim reaper. Coming in and making phone calls.... And... it made everybody uncomfortable.... And my one cousin ... who I'm very close with...it took me a long time to get her to go test. You know, she kept saying, “Well, ...I'm older now.” And I kept saying, “No, you're not. You're right at that age. You have to go test.” So then it became... I didn't want to overburden anybody. I didn't want to step over that line, but I really wanted them to see the importance (of testing).

Others described how hard the disclosing was for them personally, as well as for their family to receive this BRCA information. One described her family's reaction on

two levels, “it upset them a little that I have it (the BRCA positive mutation)... and it raises their own fear”. It was also hard for her emotionally “just because I knew that not only are you absorbing (that) another one in the family (is at risk), but ...it trips that switch in you personally yet another time”.

Most participants preferred to disclose the family’s HBOC risk, unless there was a reason that inhibited them, such as the family member’s age, not wanting to worry someone that was sick, or to prevent guilt feelings in someone who was ill. While most thought it important that family members know their risk of a genetic mutation, they carefully assessed others’ receptivity. They did not anticipate the range or intensity of personal and family reactions they experienced while disclosing their genetic information. Some experienced a sense of reconnection with the family while others discovered not everyone, including sisters, are of their mindset and really did not want the information.

Blocking disclosure. A few families had members who weren’t receptive to hearing about the HBOC risk in the family and participants found their disclosure blocked. This participant explains her rejection:

...I started talking to her about it and she like pretty much slammed the door in my face. And I was pretty afraid to continue discussions with her. And I was concerned for her because ... she’s Ashkenazi and has had ovarian (cancer) so, she’s got a good chance of having it (BRCA gene)... But then I talked to one of her sisters. And her sister wasn’t very approachable about it, so I just kind of dropped it.

Another participant experienced indirect blocking of her communication. After drafting a letter with the help of her genetic counselor, she sent it to her distant relatives. There was a lot of silence, as she heard back from only two of them. This left her feeling “isolated”. She speculated that there is perhaps a fatalistic mindset in some of her family

and realized that some people are not interested in sharing in this BRCA information. She described trying to tell her non-receptive cousin with ovarian cancer:

One cousin... and ironically it's one who now has cancer, she hasn't been tested and she doesn't want to be tested and she doesn't really even want to talk about it. So I've just been really, really careful about pushing it on people. Or tried to be, cause I do have a sense that some people just aren't that interested.... there may be ... a mindset that says... "If I get it, I get it, and I just don't want to know". It's ... like, "why would I want to know this?" ...there's parts of my family that are very religious, and I think they kind of feel "...if it's God's will, then I'll get it or I won't and I won't interfere in that".... But the months and years after that, I did have a sense of isolation because people in the family very rarely brought up the subject and I tried not to because I didn't want to push it on them. And I think I felt a sense of isolation.

One foreign born participant with a genetic VUS also experienced indirect blocking of her disclosure, but this was due to language and vocabulary barriers. She expressed how difficult it was to share her VUS results with sisters in another country. "The vocabulary" to tell her sisters about her variant of uncertain significance was technical and the information she received was obscure and fraught with uncertainty. They had not heard of this new technology, nor could they afford it. Consequently, this family was subjected to additional stresses resulting from their lack of understanding.

Selectively disclosing to children. Most participants with young children did not disclose their BRCA test information to them, although, a few did. Two factors influenced parents' disclosure of BRCA results to children, the child's age and the parent's philosophy about communication. Most felt that they would tell their children when they were older, when they would understand and faced a potential risk themselves.

One participant who did not believe in keeping secrets because of her experience in adoptions, felt her daughters would overhear conversations and misinterpret her BRCA mutation as cancer. Thus, she shared the information openly with her 9 and 12 year old

daughters, as she wanted them to be a part of it. She gave this detailed account of why and how she and her husband disclosed her BRCA genetic results to them:

We sat down with our girls and talked to them about it.... And we had talked about this prior to as well.... we really felt that we could not keep it a secret. I knew that they would be overhearing me on the phone and I was worried that they would think that I had cancer or that they would be scared that it was something worse. I just didn't want them to feel that they weren't part of it. So in as simple a way as we could... or as age-appropriate way as we could, we explained to them what was going on. Tried to reassure them and I talked to them... And so I told them I had this gene that meant that I might get cancer. And to make sure that I didn't, I was going to have this surgery, just like grandma had, to make sure that I didn't get cancer.... I think it was a huge relief for them, because they had ...been going through my mom's death. And I mean, because we were caring for her... that wasn't hidden from our girls. They had been a part of everything.... So that was scary for them. So to know that I was going to have some surgeries to make sure that I didn't get cancer was a good thing. And they still say that they think it's just great that I'm doing this (prophylactic surgeries).

Although this mother did not disclose her BRCA positive results to her young daughter, she did tell her 17 year old son. She explained the implications for his children and recommended that he have the testing when older. She was concerned her son would see literature lying around and misinterpret it as she had breast cancer. She recounted:

My son, on the other hand, watched my mother (die of breast cancer)...he was seven, very close to my mother.... and it was very difficult for him. And I knew that there was going to be information coming to the house. I had seen somebody from reconstruction, and... he mailed me a book on ... mastectomies after cancer.... Once I saw that... I didn't want him (son) to think I had cancer. So, after my testing I went up and I told him ... that I tested positive, but that I do not have cancer. I wanted that to be very clear. And then I told him what it meant, and I told him that he, too, might have the gene and might not. And what that meant for him in terms of his children ... So I told him, when he was older, ... if he wanted to, that ... I thought it would be to his benefit to test, but not now. Not at this age.

Another mother who disclosed her test results to her 8 and 12 year old daughters, gave a detailed account of her daughters' emotions, from feeling isolated and different (stigma), repressed anger, and desire to be a "happy teen":

I think most of the time they like to just ignore it all and go on with their happy teen-age lives.... On the outside you don't see that it really affects them they're just totally normal kids, and yet I think somewhere underneath...and every so often ... it comes up.... And so they're thinking about it. They don't really have anyone else to talk to about it. I think they probably feel a little isolated and I think a little like the way I felt different because I didn't have a mother. I think they feel different because they have a family who has a gene, a cancer gene.... I think this is an experience that will, in some way, mold their lives I hope it's not a bad thing And it's only rarely that they'll come and divulge what they're thinking. And I did have one conversation when my daughter was sixteen She (was)... pretty emotional and I sat at the table and cried while she talked about some of the stuff. But she said, ... and we'd been not getting along for a few months..., "you know, yeah it's got to do with this gene thing.... I guess I'm kind of mad at you for this" ... because she said, "I want to be a happy teen. I want to be carefree and I know that this is going to hit me sometime".... every so often, you see a little glimpse that really it is in the back of their minds.... They push it far back and they don't think about it a lot, but it does... it sort of simmers there and it does affect how they think about things.

In contrast, most parents did not disclose their genetic test results to their young children. Instead they shared their surgical procedures and explained they were done to prevent cancer in the future, linking the explanation to a family member's cancer or surgical procedure they could relate to. These mothers recounted their experiences:

I told my kids about it (BPM). My mom had her implants replaced ... and stayed with us for two weeks after that surgery. So I told my children, who are 5, 9, and 11, ... "Remember (grandma) came and stayed with us and she had surgery to make sure she wouldn't get breast cancer, and I'm going to have the same surgery". And I said, "So you all are going to have to be real helpful for a while, just like we were with (grandma), and take care of me for a couple of weeks. And they kind of rolled their eyes and laughed.

My daughter, I told the truth (about BPM) as I thought she would be able to deal with it. ...she's nine years old and developing ... And she's having some body issues. I didn't want to make it worse. So what I said to her was that I had some cells in my body that could turn bad, and make me sick. And that I found out about them before that happened, so I was having them taken out. And then they were gonna put in different cells from another part of my body to make up for the ones that I lost. ...that's what I told her. As she...started seeing, she asked questions. And so I explained to her ...that it was in my breast area, and she's like, "Ooh, don't say those words." Cause she's dealing with ... her own body development and image and that satisfied her.... She could see that I was fine, and at nine, I think that's all they wanna know.

However, when parents decided not to disclose their BRCA mutation to the children, to keep the secret until they are older, they restricted the number of people to whom they disclosed their genetic information and received support from. In this way, there was less likelihood that someone would spill the information inadvertently. Parents who were less open and blocked communication with friends about the BRCA mutations, as this mother explained:

I talked to maybe four or five other couples, good friends of ours, and that was it. Nobody else knew about it (BRCA results) because ... I have three daughters and they were ... the highest priority for me.... I need support, but if I talk to a lot of people then somehow it will get to them and I didn't want it to get to them.... and then my husband and I talked about it and he said we shouldn't and once we'd decided we're gonna wait 'til eighteen or twenty, then I didn't want to make everything so common knowledge with everybody because ... some people are insensitive ... and might say something... I don't want the kids to know, about it from somebody else...

Fear of health insurance and employment discrimination were other reasons families decided to hold the information close, as this participant discussed:

But it was kind of a family decision to not really let many people know, because of worries about future insurance problems or work related prejudice or something like that. So we kind of as a family agreed to not really share the information with too many people.

In summary, disclosing results was a strategy to seek support from family, friends, and/or support group peers. It also served to inform others so they too would be more aware and take necessary precautions to prevent breast and/or ovarian cancer. In disclosing their results participants encountered two responses from others, that of the insider and outsider. The outsiders did not understand what the mutation meant to the participant and thus support was lacking. Insiders understood the meaning of the mutation, as they had experienced the mutation or VUS test result. They had a common ground of shared experience and concerns and provided a sense of connectedness which

helped to reduce the participants' fears and isolation. They found insiders within the family, as well in BRCA support groups, such as FORCE, HBOC, and hospital-affiliated support groups. Value differences regarding openness in communication, sense of duty to inform, age and maturity of children, and differences in receptivity led to selective disclosure to family members and others about their BRCA mutations. Disclosing of their BRCA results was an important step in breaking down their feelings of fear, isolation, and vulnerability so they could proceed to further manage their susceptibility to breast and ovarian cancer.

Making Risk Management Decisions

For unaffected women with BRCA positive or VUS mutations, making a risk management decision was a strategy to manage susceptibility to breast and ovarian cancer, as well as the consequence of their knowledge and experience. Although participants experienced a wide range of emotional reactions after receiving their BRCA test results, they sought support and confronted their fears, pulling themselves together in order to move forward, and regain control. In considering treatment options of prophylactic breast and ovarian surgery, chemo-prevention, and vigilant surveillance participants were choosing either to reduce their risk of breast and ovarian cancer or "catch it early." Risk reduction options included the prophylactic surgeries and chemoprevention with tamoxifen. Early detection ("catch it early") involved annual vigilant breast and ovarian screening. Both the certainty and uncertainty of their BRCA test results set in motion a series of interpretations and actions leading to risk management decisions. This set of actions included seeking information, drawing on resources, sustaining relationships, and deliberating and making decisions.

Seeking Information

Decision making to try to reduce their risk of breast or ovarian cancer or “catch it early” was “hard,” “tough,” “scary,” “agonizing,” “daunting” and made some feel like they were on “an emotional roller coaster” because of the presymptomatic nature of the testing and the uncertainty involved. Few risk management options are black and white and the gray areas are many, due to a paucity of long term follow-up of unaffected women with BRCA mutations and variants of uncertain significance. These decisions had to be made by both those with positive and VUS results. As this 34 year old with a variant of uncertain significance explains: “If it’s an inconclusive result like mine was, that doesn’t mean that there’s no decisions to be made. You still have the same decisions as everybody else”.

After obtaining information from their genetic counselor or physician on risk management options available, participants sought information from lay and professional sources to assist them in making informed choices. Knowledge gained in information seeking after their testing was effective in reducing the bewilderment they felt in discriminating between alternatives. Preferences for both the type and amount of information varied among participants. For some, their physicians taking a non-directive stance, not prescribing what to do, left them disappointed because they felt they must make a decision when they did not have the requisite information. As this 32 year old with a BRCA mutation explains:

You get the results of your mutation testing and they sit there like, well there’s ...you could do this but there’s not really any data to support that, that really helps, and some people think you should do this.... It’s like ...no one is going to sit you down and say, here’s what you need to do. So it’s all up to you. And that can be daunting.

Acquiring information, figuring out “where to go and who to see” took initiative and time. As this 47 year old who sought testing in 1999 and described her experience as “agonizing”, like “pushing a big boulder up a hill” explained:

They kept saying, “there is no clear thing to do here. We don’t have a specific direction”. And they continued to go on with “these are very personal choices and you’ll need to discuss this with your doctor”. And that was it.... I felt like I had to push this big boulder up a hill. Because I had to push to get into doctors and figure out where to go and who to see and I didn’t even know what order to see the doctors in. (I saw) ... a gyn oncologist who I talked to about having my ovaries removed and all the things that go along with that. I saw a breast surgeon ... about the option of having my breasts removed. Then I saw a plastic surgeon and ... later I ...saw an endocrinologist to talk about hormone replacement ...actually I saw two different endocrinologists afterwards. So it was kind of bouncing between those four doctors.... And I didn’t get a lot of guidance honestly from the genetics clinic.

Like the above participant, some preferred obtaining information about treatment directly from professionals in clinical settings; including breast surgeons, plastic surgeons, oncologists, gynecologists, and gynecologic oncologists. They also used written materials provided by their physicians and genetic counselors. Others sought information from members of support groups and read popular media sources and scientific reports on the internet. This participant’s belief that “knowledge is power” and the key to “life saving decisions” was shared by several participants:

I think the most important thing is the information. Knowledge is ... so important. And with this knowledge, you can make lifesaving decisions. And then you have to remember even if you're positive, it does not mean you have cancer. You have to keep reminding yourself of that. But that you can make such good decisions for yourself and for your family. And that knowledge is power.

For some, gathering and sorting through all the information left them feeling “very overwhelmed many times....just the amount of information that I had to try to process.” Some sought professional publications, but found the medical language was technical and hard to interpret. This 47 year old participant described her information

search, difficulty understanding the medical language, and need for someone to translate it for her:

And I wish I'd had easier access, clearer access to the medical and scientific knowledge out there. I felt like it was really difficult to dig it all out and try to make sense of it. Cause I'm not a doctor. A lot of this was written in medical language. And it just wasn't there translated in ways I could understand. Well, I could. I mean I did my best, but I often felt like I would have liked to have somebody translate it for me.

Others compared their information with that given to a sibling with the same BRCA results, as this 43 year old recounted:

I did online research, as much as I could find. I met with oncologists. I met with breast surgeons. I met with my gynecologist, my family physician. And then, my sister was meeting all of those same people, so we had doubled the input with half the legwork.

Several of the participants emphasized how valuable it was for them to meet with or talk (by phone or in chat rooms) with women who had similar histories and had been through the decision making process and were happy and have moved on in their lives. These women were role models who openly shared their knowledge and experience about issues that seemed insurmountable at the time for the participant. Participants found BRCA peer support groups not only had up-to-date information on research and treatment, but also had links to other internet sites, so they could do their own information gathering. These three participants describe their support group experiences, the "wealth of information", including physician referrals:

The HBOC group... these women have just been a wealth of information.... And so I got to see, not only to talk to them, but I got to see the whole range of options, women who had chosen surveillance, ... to have the surgery, ... who had had trans flaps, ... had reconstruction through implants. And from them I could hear different information, and then I got names of different surgeons.... So I immediately had referrals. I went to my GP (general practitioner) and got referrals to those doctors.

(Through FORCE) I learned all about ... Medline I didn't realize that I could just go and find abstracts of journal articles on line and read these studies first-hand myself as opposed to reading the interpretation of it in *U.S.A Today*, where everything gets blown out of proportion....

These support networks helped participants regain a sense of control over their lives and reassured them about the decisions they were considering. As these two participants recounted:

(FORCE has) been ... a good source of information for me. ... I'm finding out from other women what procedures they've had, what the effects have been, and how it's gone, what surgeons they recommend, and what the pros and cons are of the different procedures.... Yes, a lot of what I'm saying is like well... I've been right all along in knowing I should do these things.... It's making me see that, yeah, this is the right thing.

(FORCE) was an incredible source for me, to help me feel more comfortable about my research and my decision and second opinions and things like that.

The decisions for prophylactic surgery were multifaceted, not just for BPM and BPO. Bilateral prophylactic mastectomy included options for breast reconstruction. The decision for BPO involved deciding whether fallopian tubes and uterus should also be removed, and if hormone replacement therapy would be used to mitigate the symptoms of menopause.

Participants discovered several different breast reconstruction options available and described how their views on autonomy, trust, and control influenced their decision making process. They either decided on what options they wanted and found a physician to perform it or found a physician they could trust and went with what he recommended. One participant discovered that plastic surgeons do not explain all the options for reconstruction, just those they perform:

There's so many different types of reconstruction, it's sort of a whole sub-world unto itself.... Silicone versus saline is the least of it. There's keeping your nipples, loosing the nipples, you know the diep flap, the glut flap, the back flap,

expanders.... There's just a ton of decisions to make. It's not enough to decide that you're having a mastectomy. That's like the least of it.... Every plastic surgeon has their own surgery that they push. And you ...go online and you do some research and you can make up in your mind what ...you want to do, and then you find a doctor who does that. Or you can go to doctor after doctor 'til you find a doctor that you like and trust and go with what they say.

It was through the FORCE website that some discovered complete information on all the types of breast reconstruction available, as this 47 year old mother recounted:

...that (reconstruction) was the most difficult part of this.... cause the doctors don't tell you the different options if they don't do them.... Through FORCE I sort of feel like FORCE was such a lifesaver, because I really did feel like I was drowning.... Everybody was giving me limited information as to what area of expertise they had. And they all had an agenda.... But there was no one outside FORCE ... who was just gonna be willing to give me information and let me make a decision for myself.

Drawing on Resources

Decisions made for any of the risk management options were complex and grounded in the trust, support, beliefs, and values from each individual's life experiences and relationships with others. Past family and personal experiences, present sense of self identity and their relationships, and aspirations for the future were all part of a participant's decision making.

Participants who were interviewed at longer time intervals from their decision making process, related clear accounts of their life experiences influencing their decision making, which were consistent with the experiences of those who had made more recent decisions.

Past experiences. Participants made decisions about their risk management options by interpreting their past and applying it to their current situation. Family history, past personal experiences, and the experiences of family, friends, and others provided the

groundwork for participants' expectations about their susceptibility and risk management effectiveness.

Family history influenced their decision, as breast or ovarian cancer represented death or loss of quality of life, as their family member(s) with these cancers did not survive or had suffered through traditional treatments like surgery, chemotherapy, and radiation. Participants chose risk management options they perceived gave the greatest risk reduction and thus enhanced their chance of living out their lives. This allowed them choices that their family member(s) did not have, as expressed by these participants:

If I had survivors in my family, I might have said, "... what's the worst that happens is, with all the surveillance, they catch it early". But they didn't survive. I mean ...with my mother's cancer it was a...tiny little lump. I mean, no affected tissue, no affected lymph nodes, nothing.... It (BPM) would put me into the 90th percentile of not getting this illness that I saw my family members die of.

50 years old with a VUS who chose BPM. Well, if I have to make a decision because I'm living with the unknown, and I'm feeling, thinking of my sister (died recently of breast cancer) so close to me, telling me, "I didn't have that test, I didn't have the opportunity, don't think twice what you have to do".

Other important elements in decision making were the participants' values and beliefs about their quality of life and its impact on their children, should they develop breast or ovarian cancer. This mother expressed her beliefs about quality of life:

46 year old mother who chose both BPM and BPO. I'd been so involved with all my sisters (three with breast cancer), what they've been through and their scars and their drains ... I've seen it all and it's not pretty and I just didn't want to have to go through the chemo and the radiation or whatever.... I didn't want my son to have to see me go through that... (On getting her positive BRCA test results): I knew right then and there what I had to do.

Past personal experiences. Past personal experiences also provided participants with a framework in which to judge their options. Most participants had been doing vigilant breast surveillance prior to testing. Both those who chose prophylactic surgery

and surveillance expressed an awareness that mammograms were going to “find” breast cancer, not prevent it, as expressed below:

Chose BPM and BPO. I’ve had mammograms every year since I was in my late twenties. Well, I mean, it’s better than not having them, but it’s not going to prevent cancer. It’s only going to find it.

Chose surveillance. I feel like my surveillance is such that ...I have the best possible chance of catching it early and I have great faith that if it’s caught early and dealt with early, it’s a very treatable disease. And I know dozens of women who have survived it and I think that I could be one of them, worst case scenario.

Although participants who chose surveillance and surgery had reservations about mammography, those who chose surgery expressed the greatest distrust. Even as one participant who chose vigilant surveillance explains, she only felt she didn’t “have breast cancer” and had “peace of mind” when she had an MRI.

I do annual mammograms, annual breast ultrasound, monthly breast self-exam, twice annual clinical breast exam and then I’ve had one MRI as part of a clinical trial and I’m trying to have another one because that was definitely the screening that gave me the most peace of mind. When that came back clear I was like, “Ok, that’s good”. Because when I get a mammogram and even an ultrasound and certainly the clinical exams...when they say, “Well I don’t see anything” or “it looks ok”, ...I’m glad, but I don’t feel like “Oh, whew, I definitely don’t have cancer”. I don’t have that much faith in those technologies. I have some faith but not exclusive faith. The MRI, when they didn’t find anything I was like, “Ok, I don’t have breast cancer”.

Equipped with their additional information about a BRCA mutation, which confirmed an even greater risk for breast and ovarian cancer, those who chose surgery wanted a treatment that was more definitive than surveillance. Several participants with “lumpy breasts” or those who had previous breast biopsies were not willing to continue with the cycle of “anxiety, tests, waiting, and results” that mammograms or biopsies perpetrated on their lives. A 47 year old mother with a BRCA2 mutation articulated this view:

Mammogram, I just was not comfortable with the error rate and given the density of my breasts and the difficulty that I've had with the lumps and the difficulty mammogramming them, I just didn't feel that that was an option.... And I've had lumpy breasts all my life and I never knew what was a lump and what wasn't.... And I'd ... been doing surveillance those three years prior to (testing).... The six-month checkup thing, that was not a lifestyle that I was willing to carry on with for the rest of my life.... You have anxiety for a week before the appointment.... You get your mammogram and all these tests done and for a week later until all the results come back ..., you're wondering what they're going to find. And every six months you go through this cycle of anxiety, tests, waiting, results.... And then the day after you get the ... good result back saying there's nothing there, you think, well what if it's starting to grow now. And I just didn't want to live like that.

Most women also did not have a favorable impression of the CA-125 test for ovarian cancer screening, which was confirmed by their physicians. This participant described her lack of trust in this test:

And the C-125.... But what I've heard is it's not particularly effective. By the time they actually see something, it can be quite advanced. And that was confirmed kind of by Dr. (name), the gynecologic oncologist. And so although that's something that I do, I don't feel really good about that as a screening method. And ovarian cancer is, from what I've heard, so much more fatal and progressive.

Relatives past experiences. Participants' decisions were also influenced by the past treatment experiences of relatives, friends, or support group peers. They were distrustful of mammography because it had missed their mother's or sister's breast cancer. For others, ovarian cancer went undiagnosed until an advanced stage. These participants explained their distrust:

41 year old who chose prophylactic surgery. Mammography, for my mother, did not help. That cancer was not there then all the sudden came up in two months. It ... in my opinion, it laid shadowed in a mammogram and was never picked up. She has dense tissue, I have dense tissue. And I said mammograms, for me, are not enough. And I don't want to end up like this because I'm trusting mammograms.

41 year old who chose prophylactic surgery. You know, I've been doing surveillance. I have very dense ... breast tissue. And I know my sister (with breast

cancer) had a mammogram and it didn't show up anything. So I know how ineffective that can be. It had spread already. And they couldn't even see cancer on the mammogram. So for me surveillance was not an option, because I had been doing that, and I lived with fear.

47 year old who chose the surgical options. ...my sister was diagnosed (with ovarian cancer) at stage III. I looked at the statistics for survival. They're horrible. I looked at the surveillance methods. They were inadequate. It's a deadly, horrible disease that isn't found early enough. And I looked at what the potential research was coming up. And I just didn't see that something was going to be popping out of the woodwork in the next year or two.

Maintaining self identity. Just as past experiences influenced participants'

treatment decisions, they also chose to maintain their current sense of self, personally, as well as in their relationships with others. This was especially true for women in their twenties and thirties. Most of these young women chose surveillance and to postpone surgery to sustain normal roles for themselves and to preserve normal relationships with family and significant others. They were concerned about their sexual experience, wanted to marry, make decisions with a husband, have children and the opportunity to breastfeed them before they made any surgical decisions. In the meantime, they wanted to get on with a normal life. One 32 year old participant felt breast reconstruction "would take over my life.... It would be a very major life altering choice and it's just not worth it to me at this point." These young women felt they had the "gift of time" and set a time in the future to reevaluate their risk management decisions or have the BPM and/or BPO. The choice of age for reevaluation was related to their mother's age at diagnosis of breast or ovarian cancer, or was suggested by a genetic counselor or physician. As one 22 year old positive mutation carrier explained, she wanted to move this from the foreground, live her life until she was 25 years, then she would start surveillance:

I just want to keep living my life as a twenty-two-year-old and not overdo things. But I haven't even been to a doctor yet and ...it's been nine months, but I want to

live my life until I'm twenty-five ... I say that year because that's the year the genetic counselor told me that that's when I need to get serious about things. So, I've been talking to people. I've been finding out what other people have done.

A 32 year old participant with a variant of uncertain significance who chose surveillance, wanted more children and the opportunity to breastfeed them. She related that her decision making was shortened as a result:

Well, me being so young, I mean, I know you can do a mastectomy ... or a hysterectomy.... See, I don't know if I'm through having children, so with my case there was so much that my options were shortened to, because I wasn't ready to go on to tamoxifen or any other type of medication. I wasn't ready to have a mastectomy. I was getting ready to nurse a baby.... So it didn't change anything ...until I become older and I know I am through having children, only then would my options pretty much change on what I needed to do.

Unlike the younger participants, women who were older did not plan to have more children or were beginning menopause were ready to make their risk management decisions. As described by these women in their mid-forties, the decision for removal of their ovaries was not a difficult one:

And, so, all of us sisters just looked at each other and said, "Well we don't need them (ovaries) anymore." ... and the other ones are already going through menopause, and I was starting, so I thought, "Get them out of here." There's no testing for it (ovarian cancer)..., so I just said, "Take 'em. Wasn't gonna have any more babies anyway."

I mean, being 47, I could say, "You're gonna go through this (menopause) anyway." You know, had I been younger, I don't know if I would have done it so readily. But I think because of my age, and I just wasn't willing to take the risk (for ovarian cancer).

Overcoming past beliefs. In contrast to most participants in their twenties and thirties, one participant in her twenties did not postpone her surgery and had BPM. She explained how she overcame a past belief, a fatalism which she attributed to her Catholic faith. She consulted a priest who helped her understand she was not harming her body with surgery, that she could change things so cancer was not her destiny. She then

decided to become more proactive, “using the knowledge I was given” and the scientific advances available to her, as she recounted:

Another big reason that I was hesitant to have the surgery (BPM) was because of my religion I’m Catholic, and I kind of had this belief in my mind ... that I would be harming my body ..., this body that I was given, and that I shouldn’t really change things. If I was meant to deal with having to have cancer, then I should kind of accept that and deal with it and kind of suffer through that I actually felt that pretty strongly for a long while until I talked more closely with one of my good friends who is a priest. And he kind of convinced me otherwise that it wouldn’t be. And then I started looking at it more in the sense of that I would be doing something proactive for myself, and that it would be using the knowledge that I was given, and using the advances scientifically. The fact that I can know that I have the gene and know what my risks are and do something to try to change myself from having that in the future. So that started leaning me more towards doing it (BPM).

To help her deal emotionally with concerns about body image, dating, and what BPM would mean for her, this single 28 year old consulted a psychologist, as she explained:

I was referred ... (to) a psychologist who is on the staff at the breast center that I go to. And I started meeting with her weekly and then biweekly, just kind of talking through my concerns and my thoughts and feelings, and kind of tackling the issue of what it’s going to be like to try to date, start dating somebody after having a mastectomy and reconstruction, how that would be really different and difficult.

Considering future self. A desire to maintain their self concept, including their “desire not to get cancer,” led participants to think about their future. Participants made decisions that permitted them to remain hopeful about the future, especially regarding their roles as mother and wife. One single 28 year old participant viewed her risk management decision as weighing whether she should have the BPM now, without chemotherapy and a family, or do it later while taking chemotherapy and caring for her family. She related:

And I could either do it (BPM) now without having to go through chemotherapy and going through all the other things, or I could do it later, when I could potentially at that point have a family or kids to take care of, and have to go

through chemo, and weighing out my options that way. It kind of made me decide that I should probably do it now while I am just kind of on my own and not have to worry about all that other stuff”.

These women also wanted to be alive to support their husband and family, “see my kids raised” or “graduate”, “becoming a grandmother” as these three women recounted:

One thing that played a part in making the decision was when somebody said to me about, “Don’t you want to be there for your kids in the future”, and this is a way to do that, ... potentially be there longer for them by preventing myself from having cancer.

46 year old with an autistic husband and 9 year old autistic son who chose BPO and BPM. Her husband attempted suicide earlier in the year: I worry about my son, ... if I’m gonna make it to see him graduate So I worry that if something happens to me, who’s going to take care of my son? And then my husband can’t take care of my son because he’s just not that material.

47 year old mother who chose both BPM and BPO: I think about becoming a grandmother.... women in our family just hadn’t been grandmothers my mom never got to be a grandmother. She didn’t even get to see any of her kids get married and for me now to be able to think about... yeah, I do think that I will be around if my kids have children. I’ll be around there to see it and be there and know them. And that’s why this (prophylactic surgery) is all worthwhile.

Sustaining Relationships

Most participants sustained personal relationships by involving their husbands/partners and other family members in their decision making. They sought various levels of input about the risk management options from family, but most felt in the end it was a personal choice, they had to make the final decision. These women found their spouses/partners supportive, which made decision making easier and helped reduce their distress. This 30 year old with a 6 month old baby explains how she involved her husband:

He’s (husband) kind of “whatever you want to do is fine with me”, if I want to have my ovaries out immediately and not have any more kids, I think it would be

a little disappointing, but he seems very understanding about whatever, do whatever is the safe choice. So he's kind of leaving it more up to me, he's not steering me to do one thing or another ... and I keep him involved, I tell him, he talks things through with me, so he's on the same page as I am as far as having my ovaries out at 35.

Sometimes hearing professional confirmation that their wife's decision was a good one helped a spouse be more supportive, as this 47 year old mutation carrier and wife explained:

I think that he (husband) was very reluctant, although he would have done whatever I thought best, until he came with me to the breast surgeon who said to him, "Your wife's testing just saved her life." And I think after listening to her, he was very comfortable with my decision. He's been incredibly supportive.... I would have done what I did (BPM, BPO), but I think it would have put a strain on our relationship, if he wasn't as supportive as he has been. And I think that once he received professional confirmation of my decision, he was more comfortable with it.

Conversely, while trying to sustain family relationships, some of the women in their twenties felt pushed by family members toward a certain treatment option. This was met with resistance, as these young women, too, felt it was important to maintain independence in this "personal decision". One recounted her situation:

And most difficult for me was that my whole family was seemingly pushing me to... have a mastectomy, especially after ... I tested positive (and) my one sister went in for her prophylactic mastectomy and then was found to have cancer. That kind of put this whole push from my family for the rest of us to all have our mastectomies, and no real personal kind of decision seemed to be playing into it, which was kind of difficult for me to handle because it's a pretty personal decision. And having people tell you what to do was kind of a difficult thing for me.

Some participants found family members less supportive of their decisions to have surgery and participants were faced with strong emotional responses. Because of the strong reactions, they were more selective in who they told, and by not telling they protected themselves from different opinions. A 41 year old participant, in trying to

sustain her relationship with her mother and affected BRCA positive sister, found them unsupportive, initially. They felt her choices of the prophylactic surgeries were too extreme. As she explained, she “didn’t need advice,” their response hurt her so she excluded them from her decision making, and told them after the fact:

By the time I told people, I had already thought about a few things, where I should go with this and I’ve already talked with my husband and I’ve already decided that we still have to research everything, but if it means taking out the ovaries and taking out both breasts then I’m gonna opt for it. So when I did tell people it was already ... “I don’t need advice” At that point, my sister, ... having gone through full blown cancer, said she hurts me really easily and she said..., “Don’t panic. Think about it, I don’t think you have to go to such extreme measures.” And this was not even deciding, it was just sort of I wanted it to be out there so that she would know that this is a possibility for me I’ve never relied on somebody else’s thinking or doing the decisions for me.... I talked to my mother, ... she said... , “Well, did you talk to doctors and did they talk some sense into you?” ... “Did they calm you down?” ... So, actually my sister and my mother, I didn’t let them in on this process at all after that. I was really hurt from both of them and they were the first ones I told. ... they didn’t know about my ovary removal ... and I only told them about the mastectomy two weeks before, sort of giving them a hint, you’re not part of my process any more. I just want to let you know if you feel like supporting, be there for me, ... They’ve been very supportive since then, but, when I first started to talk about it they were so unsupportive.

Relationships with healthcare team. Finding the right physician(s) was a key element of participants regaining control and decision making. They felt it was up to them to put together a team they could relate to in a personal way. They wanted someone they could “trust” and felt “comfortable” with and was highly competent. Physicians helped in participants’ decision making by providing information about the risk management options (both advantages and disadvantages) and establishing rapport. As one 41 year old participant described, finding a team she felt “comfortable” with was hard work:

What was difficult about the decision-making process was actually deciding on the kind of surgery and getting in to see the specialist and feeling comfortable

with that team. That was hard! ...I knew once I saw my breast surgeon, Dr. (name), I just really liked her and I felt instantly very comfortable with her. But the original plastic surgeon that I saw, I just was not comfortable at all with him. So I had to get a referral to another one I had to wait a long time to get in So kind of once I had chosen my team and I knew the kind of surgery... (she proceeded).

Reconnecting with a physician she trusted was the first step for this newly diagnosed 51 year old with a positive mutation:

Then the first thing I did was I had an Ob/Gyn who I loved; she had moved from the area ... about 45 minutes away from here, so I had stopped going to her because of the distance. But I totally trusted her. So when this (BRCA positive test) came, I called and made an appointment to meet with her again, and I'm back seeing her.

Physicians also provided ways to focus and frame the decision attributes which helped participants to make a decision. This 52 year old with a positive test result, who felt she had "this sword of Damocles hanging over my head," explained how her physician framed the issue around her anxiety:

...she (physician) looked at me and she said, "Well, what I would do and what you would do might be different..." And this was like probably the most helpful piece of advice that anybody gave me throughout all of this. ... "(participant's name), you have to weigh the anxiety you have about disfigurement with the anxiety that you have about developing breast cancer." And I just went to her and I said, "...now I get it. Ok. I'm gonna have to have surgery." Because the anxiety of developing breast cancer was just increasing. You know, it was like I have this sword of Damocles hanging over my head.

Deliberating and Decision Making

After having obtained sufficient information about risk management options, and having drawn on personal, family, and professional resources and relationships, participants began deliberating or examining the alternatives to select a particular option. The participants looked at the advantages and disadvantage of the alternatives and sorted out their feelings, weighing alternatives based on personal experiences, beliefs, and

values. The time required for deliberation after testing was related to how much they had considered various options before BRCA testing and their preferred decision making pattern. Four decision making patterns emerged (a) acting on apriori decisions, (b) following expert advice denovo, (c) following some and rejecting other advice, and (d) postponing the decision until older.

Acting on apriori decisions. Several participants had considered having a BPM prior to BRCA genetic testing. They had heard about prophylactic mastectomy in connection with a sister or mother's breast cancer. For one 41 year old participant, her physician recommended BPM when she was 25 years old. Once she had the additional information of a positive BRCA mutation she knew she would elect BPM:

I was very determined that I was going to have the mastectomies.... In 1989 when my sister died, we had a breast surgeon that the girls, we all saw. And he ... recommended that we have prophylactic mastectomies done And so we were all thinking about it then. I hadn't even had kids. I was 25 years old. So that had been something that I had thought about for a long time. And then once I knew I was positive for the BRCA gene, it was like ... of course I'm going to do this.

For another 41 year old who had seen that her mother's BPM prevented breast cancer, testing provided the certainty she needed to proceed with this surgery. She recalled:

And since probably my early thirties we've known there was a strong family factor, whether we had any proof of it or not, and saw that my mother had a prophylactic mastectomy and did not get breast cancer. And I figured that's what I needed to do, too. And so I have been considering doing it for ten years. I've been putting it off, though.

After discovering her mother's positive BRCA results, this 41 year old mother waited two years to test. She had already decided to have prophylactic surgery because of the looming threat, "ticking bomb" of breast cancer, as she described:

Even before I had this (BRCA) test, ... it had been almost two years from when I found out about my mom (BRCA positive) to when I decided to test. And I had pretty much had my mind made up even before the test that if it came back

positive ... that I would probably do the surgeries, just because, otherwise I'd feel like a ticking bomb way too much without doing something.

For a 34 year old mother with a variant of uncertain significance, she decided to have a BPM if she received any BRCA test result but a negative. She realized that a VUS result was a possibility, as she reported:

I actually made the decision the day that I went home after my doctor's appointment, before I even knew what the test (BRCA) results were. I knew that I was going to have the surgery (BPM). Either I said that because he told me that there was a chance that the test could come back inconclusive, and he explained to me ... what that meant, as far as he understood it. And I came home and I talked with my husband, and we decided at that point that if it didn't come back no (negative), that I would have the surgery. And that was the end of that. In the weeks before my surgery (BPM) I still had that fear that something could pop up in those weeks, and I didn't feel good until I woke up from the surgery.

Following expert advice denovo. Those who took their physician's advice, acknowledged the risk management decision was theirs, but deferred to their physician's expert judgment. This trust in their physician's expertise was noted particularly in those who chose surveillance as their final choice. One 32 year old mother with a variant of uncertain significance who chose surveillance explained that she felt confidence in her physician's ability to monitor her and thus did not choose BPM:

So the only comfort that I have and the only reason why besides I'm not quite sure I'm through [having] children, is Dr. (name). That is the only comfort that I have in waiting is knowing that she's totally qualified in helping me make decisions But if I didn't have her, then my options I'm sure would have totally changed ... it's totally because of her the reason why I haven't drastically maybe did a mastectomy.

This 43 year old mother with a positive BRCA mutation, who chose both BPO and BPM, initially felt prophylactic surgery was too extreme. She related how her physician helped her see the advantages of prophylactic surgery, which changed her perspective:

Well, I never for a second considered having a mastectomy. I just thought it was so out there and extreme, until I met with my ... sister's oncologist.... And that was really the first time that I even considered it (prophylactic surgery) He said I could get my ovaries removed and then I wouldn't be able to go on hormones because I'd still have my breasts. So I would be 43 and going through menopause without any relief. And ... the type of cancer that runs in our family has a high ... penetrance. ... even taking out my ovaries, while that would eliminate the risk of ovarian cancer, I was still faced with the real probability I was going to get breast cancer then you have to go through chemotherapy and radiation and whatever. He sort of let me see the light or the advantages to taking care of it prophylactically.

Another participant, a 51 year old mother who chose both BPM and BPO, describes how she wanted her physician to tell her she “had to do” the surgery, as this was a “huge decision”:

The mastectomy was harder.... what I was really looking for was the doctor to say, “you have to have this done”, because a double mastectomy, that was a huge decision. I mean, I kept looking at my breasts and going, “Oh my God, they're going to be gone”.... And finally the doctor I decided to go with, she took my hand and she said, “(participant's name), this is something you need to do. Your chances of getting breast cancer are in the 80 and 85th percentile. You need to do it.” And you know, I needed to hear that. All the research I was looking at was telling me I should do it, but I needed a doctor to tell me I had to do it.

Following some, rejecting other advice. Those who analyzed and deliberated over the options took control in a way that was more purposeful than the previous patterns. These participants were more thorough in laying out the advantages and disadvantages of each alternative. This participant described her deliberations with her husband:

We spent the weekend at this resort ... looking out at the beautiful scenery and going through all this stuff. Talking about the options. Made little charts about ... if I did this combination of things, what would the pros and cons be. If I did this combination of things what would...and what should the timing be ... And I mean money wasn't an object because we don't have to pay for our surgeries And insurance wasn't an object. It was just a matter of what was the right decision for me. And by the end of that weekend it was very clear to me that I was going to go ahead with the prophylactic surgery it just kind of solidified for me that I felt ok about making that decision despite what other people were warning me about and telling me.

This group gathered additional information to answer questions about the options and sought professional consultations with various experts, sometimes seeking second opinions to validate information they had obtained. Having a physician who could inspire confidence, was nondirective, yet helped them make a decision or affirmed their decision was important as this participant related:

And she (physician) just said, I think this is a good decision for you as well. She said, "I thought I would have to talk you into it, but I can see that I don't have to do that."... But again, it was after they knew where my state of mind was and what I wanted kind of going into it. I don't know that they would have done that had I not been pretty sure that that's what I ... was going to do.

These participants were aware there was uncertainty in any option they chose. They made a choice when they were confident they had considered all the relevant information and found an alternative that satisfied their most important requirements. They required the most resources of time, expert opinion, and information to make their decision. They also identified potential problematic outcomes that were the source of some conflict, but reported a sense of control and confidence over the process of selecting a risk management option. As this 54 year old participant who chose both prophylactic surgical options detailed:

The pros (of BPM) is pretty much along the same line as the ovaries ... it was going to give me the satisfaction, as much as possible, of removing the possibility of getting the breast cancer and living longer without getting cancer, in those areas, anyway. And, being able to live a life, not having to wonder at each mammogram or at each physical, doctor's appointment or at each time I would check myself, which I did monthly pretty faithfully, whether I was going to be finding something. And, so the pros were I could see myself sleeping better, living a life better, not having to worry about it and living longer. The cons became more a thing of, a sexual thing between my husband and I of ... I'm not going to have nipple sensitivity ... I might not even feel him touch my breasts ... And so that ... really caused me a lot of, probably surprising amount of emotion, that I said, "God, I'm really gonna miss that. That part of our, our bedroom life ..." And yet, it wasn't a matter because I was upset about that that I was thinking of not doing it, it was just realizing that that was gonna be gone.

Postponing the decision until older. As indicated in the previous section, maintaining self identity, women with positive and VUS results in their twenties and thirties wanted to sustain normal roles for themselves and to preserve normal relationships with their family and significant others. They wanted to move this issue from the foreground until a later point in time. This 25 year old single student reported her desire first to marry and have children, was hoping for better options in the future, but was planning on having prophylactic surgery in the future:

I'm not getting married any time soon, but I think that maybe once I get married and, I don't know, maybe have my kids, maybe not have my kids, if I'm going to do this before I have my kids, I'm going to have some kind of prophylactic mastectomy.... If I end up having children younger, I might do it after, in which case, I'm going to see. It's just so far in the future that they might have a cure for it, or they might have so many new treatment options, that I'm really not inundating myself with information about the current ones, because they're going to be outdated by the time I make these decisions. I do plan on having prophylactic hysterectomy after I have my children. And that's it. I mean, (in the meantime) I'm just going to follow up really closely.

These women felt they had the gift of time and set a time in the future to reevaluate their treatment decisions or have the BPM and/or BPO. One 30 year old new mother with a family history of ovarian cancer, who wanted more children, planned on having a BPO at 35 years. This age was recommended by her gynecologist, as she related:

So when I told my doctor (gynecologist) the (test) results He said, "Thirty-five, you need to have your ovaries out" and he said, "I would take your tubes and uterus, you don't need them at that point" So, I am planning to do that at thirty-five, if not before. If I am able to have more children and I'm done at thirty-four, I'll have it out at thirty-four.

In the final analysis, it was the option's risk reduction capability that made it salient or attractive to the participant, causing the option to overshadow consideration of other available options. Participants balanced the gains of risk reduction, relief of breast

and ovarian cancer worry, and fulfilling their obligations as mother and wife to remain cancer free, against the potential losses of surgery; such as menopause, infertility, changed body image, the continuing risk of developing ovarian and breast cancer in residual tissue, and surgery(ies)' effect on family and employment. Some who chose vigilant surveillance anticipated further consideration of their choice in the future. Table 3 provides a summary of the advantages and disadvantages of the various risk management options (in order of precedence) expressed by participants.

Decisions made. Sixty percent of participants (71% of those with a BRCA mutation) chose one or both of the surgical options (BPM or BPO) to prevent breast or ovarian cancer; 40% chose both BPM and BPO. Participants clearly preferred prophylactic surgery over early detection measures to reduce their susceptibility to breast and ovarian cancer. See Table 4 for a summary of participants' risk management options chosen. Of the twelve participants that chose both surgical options (BPM and BPO), three chose concurrent procedures and nine chose to spread the surgeries out over time. Six additional participants chose only one of the surgical options (BPO: 2, BPM: 4). The mean time from testing until surgery for BPM was 10 months (range 1-40 months) and for BPO was 6.9 months (range 0-27 months).

The sequence in which participants had their surgeries varied with each participant. Participants made this sequencing decision around their greatest fear or perceived risk, as this 52 year old woman with a father with breast cancer recounted:

And I think what you find is that those of us who have lost a family member to one or the other cancer, our fear is what we lost our closest loved one to.... I didn't know my grandmother that much, so I wasn't fearful of ovarian, as much as I was fearful of breast. So, I focused all of my mental energy at first on breast cancer.... I could not deal with my ovaries until I was done with my breasts they didn't have the ticking time bomb aura to them that my breasts did.

Table 3. Participants' Perception of Advantages and Disadvantages of Risk Management Options (attributes of options listed in order of precedence)

Bilateral Prophylactic Mastectomy (BPM)

BPM Advantages

- Greatest risk reduction for breast cancer – most effective option
 - Reducing breast cancer risk to the 90's of not getting it
 - No tissue, less chance of cancer
- Will save my life
 - Preventing/avoiding cancer in the future
 - Living longer without getting cancer
 - Avoiding chemotherapy
- Being there for my children
- Peace of mind, knowing I had done all I could physically
 - Rid my life of this fear of cancer
 - Not having to worry so much about breast cancer
 - Reduced anxiety of getting breast cancer
 - Not having to be scared like I was from May-July from mammogram to ultrasound to MRI
- A better quality of life
 - Not having to go through mammograms and biopsies again due to fibrocystic breasts
- Breasts don't serve a physiologic purpose
 - It's not a sexual thing for me
- Getting breasts that don't sag (planning reconstruction)
 - Reduce weight in breasts

BPM - Disadvantages

- Risks of surgery and anesthesia
 - Recovery and inconvenience
 - Healing after surgery; pain, scars
 - Extensive surgery
- Reconstruction hassle - long recovery
 - Possible complications like infections
 - Getting exchanges, getting nipples put on
 - It would take over my life
- Removing healthy tissue
- Not completely reducing the risk; 90% reduction, due to no long term studies
- Body image concerns, not wanting to alter body
 - Fear of feeling ugly, hideous
 - Self image – “Not a woman anymore”
 - Having something fake inside my body. Fake nipples
 - Fake boobs, they don't feel the same or look the same
 - Clothes don't fit properly

Table 3. Participants' Perception of Advantages and Disadvantages of Risk Management Options (attributes of options listed in order of precedence)

Bilateral Prophylactic Mastectomy (continued)

BPM - Disadvantages (continued)

- Relationship concerns with male/partner, family
 - Look good in clothing but not naked
 - Feeling less feminine
 - Loss of nipple sensation
 - Not able to nurse my babies
 - Explaining to the kids about the surgery
 - Would impair feminine role modeling for teenage daughters
- Cost - Need health insurance to cover cost
 - Insurance doesn't cover
 - Cost in terms of time off work
 - No nearby family resources to assist with children after surgery
- Too extreme at my age of 32

Bilateral Prophylactic Oophorectomy (BPO)

BPO Advantages

- Greatest risk reduction
 - Prevents or reduces risk/chances of ovarian and breast cancer
 - Prevent ovarian cancer, the silent killer
 - Survival; I'll live forever
 - Better than dying of ovarian cancer
- No good early detection program for ovarian cancer
- Ovaries no longer of value
 - Don't need ovaries any more, no more babies planned, starting menopause
 - Control onset of menopause
 - Wanted menopause early so it would be over with
 - Be finished with menstruation
- Provide peace of mind
 - Rid the fear of ovarian cancer
- Found ovarian cancer and thus saved her life

BPO - Disadvantages

- Surgical menopause
 - Menopausal symptoms, loss of estrogen and subsequent health effects
 - Increasing risk of significant diseases: osteoporosis, heart disease, libido concerns, vaginal lubrication, hot flashes, night sweats
 - Surgical menopause is an assault – Premature aging, mind ages, memory loss
- No standard of care for treatment of surgical menopause in BRCA women
- Not able to have children

Table 3. Participants' Perceptions of Advantages and Disadvantages of Risk Management Options (attributes of options listed in order of precedence)

Bilateral Prophylactic Oophorectomy (continued)

BPO – Disadvantages (continued)

- Ovaries serve a physiological purpose
- Removing perfectly good body parts on a “what if”
 - May be unnecessary
 - Another female organ ripped from me
- Loss of femininity
- Impairment of maternal role to role model for daughters

Surveillance – Breast and Ovarian

Surveillance Advantages

- Breast screening - MRI are definitive
- Ovarian screening- surveillance is better than surgical menopause
- Confidence in physician checking breasts q 3 months
 - Being followed closely, and getting follow up on anything that is suspect

Surveillance Disadvantages

- Breast screening- Don't trust mammograms, didn't show sister's cancer
 - Missed mom's cancer
 - Screening not sensitive enough to find small cancers
 - Error rate too high due to breast density
- Didn't reduce my risk enough
- Didn't relieve the breast and ovarian cancer worry
 - Based on my chances, I'd live my life in fear
 - Takes my time, my mind, and my emotions
- Ovarian screening - not particularly effective
 - By the time they actually see something, it can be quite advanced
 - No good early detection of ovarian cancer
 - CA-125 too many false positives - testing not that great
- Radiation with mammograms may be harmful in high risk women

Chemoprevention with Tamoxifen

Tamoxifen Advantages

- None stated

Table 3. Participants' Perception of Advantages and Disadvantages of Risk Management Options in order of precedence (continued)

Chemoprevention with Tamoxifen (continued)

Tamoxifen Disadvantages

- **Bad side effects, increased risk of uterine cancer, cataracts, deep vein thrombosis**
 - Hot flashes
 - Risk of cervical cancer
 - Weight gain -Had hard time losing weight while on tamoxifen
 - Allergic to tamoxifen
 - Couldn't tolerate it
- **Minimal reduction of risk, more for BRCA2 women**
 - Questionable results for BRCA 1 women
 - Didn't reduce my risk enough
 - 50/50 chance of preventing breast cancer
 - Not as effective as BPM
- **Adverse to experimental meds**
 - Taking a drug which is on a clinical trial basis for women with BRCA mutations
 - Not sufficient research for women at risk in "previvor" (unaffected with positive mutation) community
 - Don't like taking medication or pills
- **Not a permanent fix; I'm young, protocol for 5 years, then what?**
- **Potential harmful effects on fetus. Not supposed to have kids for next couple of years**
- **Tamoxifen didn't help mother, her cancer recurred in 5 years.**

Table 4. Genetic Testing and Risk Management Options Chosen

	Total Sample n= 30	BRCA Positive n= 21 BRCA1= 12; BRCA2= 9	Variant of Uncertain Significance n= 9
Year tested (median) (range)	2002 1994-2004	2002 1997-2003	2002 1994-2004
Surgery Chosen	Both BPO and BPM n= 12 BPO n= 2 BPM n= 4 None n= 11 Other n= 1	Both BPO and BPM n=11 BPO n= 1 BPM n= 3 None n= 5 Other n= 1	Both BPO and BPM n= 1 BPO n= 1 BPM n= 1 None n= 6
Decision Option Chosen	Both breast and ovarian screening n= 9 Ovarian screening n= 4 Breast screening n= 2 Tamoxifen, breast and ovarian screening n= 2 BPM and BPO n= 12 None n= 1 (age 22)	Both breast and ovarian screening n= 3 Ovarian screening n= 3 Breast screening n= 2 Tamoxifen, breast and ovarian screening n= 1 BPM and BPO n= 11 None n=1 (age 22)	Both breast and ovarian screening n= 6 Ovarian screening n= 1 Tamoxifen, breast and ovarian screening n= 1 BPM and BPO n= 1
Time until BPM (mean) (range)	10 months 1-40 months	11 months 1-40 months	2.5 months 2-3 months
Time until BPO (mean) (range)	6.9 months 0-27 months	6.9 months 0-27 months	2 BPOs done prior to testing
Estrogen use	Birth Control n= 3 Hormone replacement n= 7 None n= 20	Birth Control n= 1 Hormone replacement n= 6 None n= 14	Birth Control n= 2 Hormone replacement n= 1 None n=6
Menopausal status	Surgical menopause n= 14 Pre-menopause n= 3 Menopause n= 2 No n= 11	Surgical menopause n= 13 Pre-menopause n= 1 No n= 7	Surgical menopause n= 1 Pre-menopause n= 2 Menopause n= 2 No n= 4

Others made their sequencing decision around the capability of medical technologies and their physicians' recommendations. As this 41 year old mother who had the BPO first reported:

The entire team there pretty much said that oophorectomy was like ... a no-brainer. The medical oncologist, as far as the breasts said, "... we have diagnostics that we can catch this, we can catch it better than ovarian cancer, which when it's discovered it's usually very late" and my family history certainly indicates that, and they...seemed more urgent in the oophorectomy part.

Some needed time between prophylactic surgeries to "grieve" the loss of their breasts and ovaries. As this 52 year old woman who chose both prophylactic surgeries, but spaced them 2 years apart, related:

I was not going to do this oophorectomy until after I had done my breast work. I could not bear losing more than one body part at the same time. And I really felt I needed some grieving time I mean this was really hard.

Thirty seven percent of the women (5 BRCA positive and 6 with VUS) chose vigilant surveillance. This included women in their twenties and thirties who were delaying their decision until they were finished with childbearing and breastfeeding. Four in these age groups (3 BRCA positive and 1 with a VUS) indicated they planned on having prophylactic surgery in the future. In the interim they chose vigilant surveillance. Trust in the competence of their physician, use of MRI, and hope for new research discoveries were influential factors in their decision for vigilant surveillance. They found reassurance in their belief that with surveillance, if they developed cancer it would be found early. It is important to note that the advantages and disadvantages of the surgical options identified by those choosing vigilant surveillance were similar to those choosing prophylactic surgery(ies). However, how they weighed the advantages and disadvantages of the alternatives, based on values, beliefs, and desired outcomes were different.

Only two participants chose the option of chemoprevention with vigilant surveillance. Participants received less information about this option and it was not usually recommended by their health care providers. Participants did not identify any advantages of taking tamoxifen, but delineated several disadvantages, including risk for endometrial cancer, cataracts, and thrombosis, its experimental nature in BRCA carriers, and insufficient risk reduction. Several younger women did not choose this option because of implications for child bearing.

Whether participants arrived at a risk management decision based on apriori decisions, followed the expert advice of their physicians, accepted some and rejected other advice, or postponed risk management decisions until older, they reflected back to evaluate their decisions and put things in perspective.

Reflecting On Actions

Viewing Decisions With Satisfaction

All 30 participants expressed satisfaction with their decision to have genetic testing, indicated they would do it again, and would recommend it to other women who were at high risk due to a strong family history of breast and/or ovarian cancer. Although the youngest participant, age 22, was satisfied with her testing decision, she indicated that she “wouldn’t have gone through it at the age that I went through it.” And her recommendation for other young women, “Do it (testing) for yourself and not for your parents.”

In addition, without exception, all those who had the prophylactic surgeries were satisfied with their decisions. For two of the women, the prophylactic surgery “saved” their lives, as early breast and ovarian cancer was discovered during surgery. Few had

problems with their prophylactic mastectomy or oophorectomy itself, however some had difficulties with breast reconstruction. Several agreed they were not prepared for the time that reconstruction took. One participant encountered several problems which have taken five years to resolve and she still has a nipple reconstruction revision to complete.

Despite this, she was satisfied with her decision for surgery as she explained:

Overall my experience ... it was difficult, but I don't regret it. The reconstruction was the one decision that I, at times, wondered if I'd made the right decision. I think if I had not had complications ... it would have been an easier journey. I am happy to have reconstructed breasts. I'm really happy about that. But it was a lot harder than I thought it would be. And I was prepared for a year What I wasn't prepared for, was for it to go on ... for five years to be continuing.

Viewing Decisions As Personal

Participants acknowledge that having testing and managing susceptibility to breast and/or ovarian cancer are “individual decisions,” yet ones set in context. This participant advised using an intuitive approach, doing “what feels right for you”:

You need to do what feels right to you. You will get advice. You will hear people say, do this, do that, don't do this none of that matters You need to listen to your gut and do what's right for you. When you get your results, if they're positive, it's not the end of the world. And it's not a fun experience but there are people who have gone through this. You are not alone. You can get through it, no matter what you want to do Because so often people project their own fears and their own thoughts onto you, and that's not doing people a service.

Viewing Decisions As Unsupported

Although most felt things had been taken care of medically with the surgeries, one pointed out her emotional and psychological support needs were not met by her surgeons. “They talked only about the procedures”. The “whole loss part and grieving were not addressed.” But in the final analysis, prophylactic surgery offered an “amazing sense of relief, this huge risk is gone.”

After the prophylactic surgery(ies) and reconstruction were finished, most felt there was not a “cohesive plan” for follow-up. The reason follow-up was important was their lingering uncertainty about the small risk of peritoneal cancer or cancer in residual breast tissue. As this woman explained, she did not know how to do breast exams after her breast reconstruction:

I think the ball has been dropped with me.... I have silicone implants I don't know how you do breast exams on implants ... You know everyone keeps telling me you're not gonna get cancer, although there's always the slightest risk. And that it would be right at the surface, because the implant is behind the muscle, and anything you have is right up front and that you would know immediately. But certainly I need to see a doctor for peace of mind.

One seeking care at a comprehensive breast center explained, “They're not exactly sure where I fit in. I don't have cancer. I don't have breasts. They didn't do my surgery”. She had been working with her genetic counselor to get a process set up for follow-up, but it was moving very slowly. She explained:

It (follow-up) is non-existent. And I talked to my genetic counselor.... They talked about putting together a task force of doctors, at least for the institution where I was...so when you leave the hospital they tell you what to do. They don't tell you what to do. And I find that very worrisome. I went through all the surgeries to be proactive, and I'm sort of in this 'now what'? category where I'm not sure what to do. Neither do my doctors, I've gone to some specialists. I just feel I have to take what they say and make my own decisions. I've yet to find one doctor who I feel comfortable with. I'm looking I find that very ... disheartening. But the problem being that nobody wants to follow I don't know. Do I go to a breast surgeon still? Do I go to a breast oncologist? Do I go to a gynecologic oncologist? ... I don't know. There are just no guidelines.

Others concluded, “You have to take personal responsibility”, be “very self directed” in follow-up. Another participant who felt the women in her generation were “paving the way” indicated there is no “cohesive plan” for follow-up, because “nobody knows what to do ... everybody has a different opinion.” Still others appealed to the American Society of Clinical Oncology (ASCO) and other professional organizations to

develop protocols for screening of women with BRCA mutations who have had prophylactic surgeries:

I still feel pretty bad about the care that I've received as follow-up. Nobody ever presented me with a plan of how to follow-up. And I am totally in charge of my own follow-up.... So I do go back to my regular gynecologist every year for an annual exam. I try to check my own breasts. I do ask my gynecologist periodically to have a CA-125. But nobody has ever said, "Ok, this is what you should do for follow-up." I even actually insisted on having a ... transvaginal ultrasound So, I'm doing that myself too You know, it would really be nice ... for ASCO or somebody to come up with some recommendations about screening for women who have had prophylactic surgery....

Some women sought follow-up with their primary care physician, gynecologist, gynecologic oncologist, breast surgeon, or breast oncologist. However, there was not "a single place" to go for follow-up that addressed all their concerns about the BRCA mutation. An issue for those that had had a BPO was whether they could safely take hormone replacement therapy. They questioned what the long term ramifications of the BRCA mutations were and how they could stay current about new information, as this woman articulated:

You know, like when I buy a toy or something for my kids, I have to register it, like the car seats. So if there's recalls, they'll find me. But with this BRCA thing, there's no like national registry, and they're not going to find me. So I feel like I have to be more on the up and up with ... what the long-term ramifications are.

Viewing Decisions As Supported

Some of the participants found they could keep up-to-date about the BRCA mutations by joining a BRCA support group which met regularly to discuss such topics as new BRCA related research, nutrition, exercise, and stress reduction, and also provided emotional support. These topics were important because several indicated they felt maintaining a healthy lifestyle was important for their future without breast or ovarian cancer. Several had engaged in "exercising", "eating right (lowering fat in their

diet)", "seeking a balance in their life", "reducing stress" and wanted to "role model healthy behavior" for their daughters.

Celebrating The Positive

In reflecting on the consequences of their decisions, participants pointed out the following aspects (a) achieving peace of mind, (b) remaining vigilant, (c) a need to "pay it forward", and (d) strengthened family relationships.

Achieving peace of mind. Some participants achieved "peace of mind". This was especially true for those who had prophylactic surgery (ies) and those with variants of uncertain significance who interpreted their results as negative. They were less worried, less anxious about getting breast and ovarian cancer, as indicated by the comments from these two participants:

34 year old with a VUS who chose BPM. I don't have nightmares anymore. It's given me a peace of mind and it's made me more of an outspoken advocate for it (prophylactic surgery).

35 year old who chose BPO and BPM. I am much less anxious about it (positive mutation). I don't worry constantly about breast cancer. Like a load has been lifted off.

Those with peace of mind also felt they had done all they could to prevent breast and/or ovarian cancer, so they would not feel guilty if they should develop these cancers in the future. If they are present to raise their children, it will have been worth it, as this participant reported:

What I'm glad about is that I don't think about cancer I'm not afraid of cancer any more. Now I feel like if I got cancer, and I could, and given my family history, I mean who knows where I could get it, I would feel like I've done what I could and I think I would be able to deal with it Cause I did the most that I could at the time and now I'm living my life. And I'm raising my kids and ... as they get older I will feel like a big motivation for having done the surgery will have been fulfilled. I mean, I've been around to raise my kids. They didn't have to grow up without a mom.

They also started envisioning themselves getting older, becoming a grandmother:

47 year old. I really never envisioned myself as an old person either And I guess because there was no reason for me to. All the woman in my family died in their 50's. And it wasn't until my testing and my surgery that I can envision myself as a grandmother one day.

Women who had achieved peace of mind had also let go of their fear and their worry and were now more externally focused, on daughters, sons, sisters, and nieces as these two mothers related:

41 year old. I worry about my daughters. I worry about my sisters, my nieces. I don't worry about myself anymore. That feels so good to be able to say that But I don't worry about getting breast cancer any more. And I know that my chances, it's not completely gone. But you know what? I don't need to hold on to that fear anymore. I've done all that I can do to let that go.

52 year old. But I worry about my daughter. Oh, just the thought of her having to go through this is pretty ... scary. And I think it's difficult, not because I know she'd have the same experience. But I worry about her because I actually fear that if she's positive she won't look at it with the sense of urgency that I did, because she's not watched her mother die of breast or ovarian cancer. And I really fear that she's gonna minimize it because of that.

Remaining vigilant. Those who chose surveillance, both with positive BRCA mutations and with variants of uncertain significance, had a heightened awareness of their body and felt a need to be vigilant about surveillance. As one 41 year old participant related, "I know that I need to be vigilant and be serious and take it seriously, because I've seen cases where women were not doing anything and got cancer, and that's just the last thing I want to have happen".

They also felt they had to take responsibility and be their own advocate for follow-up, as expressed by this 54 year old participant: "I think it's, you have to take personal responsibility to make sure you get physicals frequently enough and to pay attention to symptoms and things like that".

For those choosing surveillance, especially those who were delaying their decision making for prophylactic surgery, there was a lingering uncertainty that maybe they didn't make the right decision or were not giving it enough attention as expressed by these participants:

45 year old. I don't think it's really changed my life, other than making me be more diligent about my screening, although I feel like I was pretty diligent already. It's maybe made me even more so ... And also, maybe one day I might change my mind about the surgical prophylactic, if more research, if things would change. I mean, this genetic testing is relatively new still. [Hardest part?] The worry that I didn't make a right decision and have a surgical prophylactic ... It's just because I'm having just the medication and the frequent screenings. What if that's not enough?

25 year old. I mean, I'm worried this whole breast cancer thing is not in the forefront of my mind. I've got so much else on my plate right now.

Paying it forward. A third consequence experienced after BRCA testing and risk management decision making was a sense of altruism, some participants wanted to pay back by helping others. One 41 year old participant, motivated by her loneliness when making her decisions, described her desire "to pay it forward" by helping support and share information with others going through this:

I am involved in circles that I never thought I would be involved in. I never thought I'd be doing this... on the phone with researchers My breast surgeon here in town calls me and says, "... would you speak to so-and-so. They just found their results." Because when I found out my results and I was here in (city) with all my treatment team, I offered my name and my number to the people in mammography, to my breast doctor,.... genetic counselors I wanted to be able to help other people that came up with this, because it was a real lonely place when I found out I sort of have that mentality of that you have to pay it forward. You know that there will be others that will come with this. That whether ya find out in 1999 or you find out in 2004, the issues are going to be so similar.

Another, a 52 year old, did not want others to feel unsupported and isolated as she had. The mutation had become part of her identity. She viewed her experience as spiritual, one of personal growth:

I vowed when I went through this, that if I had any power, ... I would not let other women be so unsupported and isolated in their (experience) I think this is a way that I can give back to the world I'm not a religious person but I think I've gotten much more spiritual throughout all of this ... And to be honest with you, if somebody said, "Oh, we could take away your mutation", if it meant that I had to give back all the personal growth that I've achieved because of this ..., I wouldn't. I would keep my mutation. Because it's been that much of a learning and growth experience for me.

Strengthened family relationships. Several of the participants felt that the testing and risk management had brought them "closer together" with family members, especially spouses who had been through the decision making and difficult times with them. The experience had helped them to reevaluate their priorities and realize that "family is more important" as this 50 year old grandmother with a BRCA variant of uncertain significance explained:

I don't go out like I used to go before with friends and enjoy. I don't know why. I'm more closer to home.... It does change you. And I think that the family is more important now. And my grandkids And sure, that's for the family's sake.

One participant gave a detailed account of her difficult times, the changing feelings and emotions during her surgeries, and how working through these brought her and her husband closer together:

In some ways it's brought my husband and me closer together. In some ways it's been difficult because there were times when I was trying to sort out hormones that I felt emotionally very volatile and I'd get into fights with him. I felt like kind of asexual for a while where I had these big lumps on my chest that didn't feel like breasts and I was probably kind of angry that I had to go through this. So there were difficult periods but all in all, if I look back now, the overall sense I have is that we're closer It was a very bonding experience to have to make those decisions together and to have to work through that difficult period when I

had that big surgery I think we feel pretty optimistic, really optimistic about our future together.

A 32 year old participant also described a strengthened relationship, this one with her father:

As far as my family, I think my father and I get along better now because ... his view being that I wasn't dealing with the cancer thing and now I'm dealing with it and he is, I think, very happy with that. And I'm happy that's it's not this weird thing that we can't talk about without getting in a fight about it anymore.

Giving Advice To Other Women Seeking BRCA Testing

Participants had the following advice for other women seeking BRCA testing and treatment: (a) get your insurance in order, both health and life insurance before testing; (b) seek genetic counseling first, don't get testing done in a physician's office without genetic counseling; (c) there is a community of women, "a sisterhood of a unique kind" that is willing and able to provide support; and (d) "be prepared for any type of result, if it's inconclusive, you have the same decisions as everyone else;" and (e) "take your time, keep an open mind and try to make sure you have people you can talk to openly and honestly about how you're feeling."

In summary, making a risk management decision to manage their susceptibility to breast and/or ovarian cancer was a strategy to manage their embodied risk from a BRCA mutation or a genetic variant of uncertain significance. It was the consequence of their knowledge and experience, as these women confronted their fear and regained control. Both the certainty and uncertainty of their test results set in motion a series of actions and interpretations leading to risk management decisions. After having obtained sufficient information about risk management options, and drawing on personal, family, and professional resources, participants' began deliberating, examining the advantages and

disadvantages of alternatives and sorted out their feelings, weighing alternatives based on personal experiences, beliefs, and values. Four decision making patterns were used (a) acting on apriori decisions, (b) following expert advice denovo, (c) following some and rejecting other advice, and (d) postponing the decision until older. In the final analysis it was the options risk reduction capability that made it salient or attractive to the participant, causing the option to overshadow consideration of other available options. After making a risk management decision they reflected back to evaluate their decisions and put things in perspective. Participants viewed their decisions as satisfied, personal, supported, and unsupported in follow-up. They celebrated the positive consequences, including peace of mind, remaining vigilant, a desire to “pay it forward”, and strengthened family relationships.

These last two chapters explored the categories, actions and interactions, and consequences of managing susceptibility to hereditary breast and ovarian cancer. The following chapter will discuss these categories in the context of existing literature.

CHAPTER VI

DISCUSSION OF FINDINGS

This chapter discusses the grounded theory of managing susceptibility to hereditary breast and ovarian cancer in relation to other research and explores the categories in the context of existing literature. Interrelationships within the categories will be discussed as well.

This study examined ways in which unaffected women with positive and VUS BRCA mutations assigned meaning, made interpretations, and responded to their susceptibility to hereditary breast and ovarian cancer. A grounded theory was developed based on the perceptions, beliefs, and feelings of these women. The study findings add to the initial body of knowledge about BRCA genetic testing and risk management decision making and provides some insights for assisting women in the decision making process.

This study used an inductive analytic technique to identify social and contextual factors in decision behavior, took a systematic approach to the study of risk management decision making in the context of participants' past and present experiences, and shed some light on the complex processes occurring when participants made decisions and appraised those decisions over time.

*Grounded Theory of Managing Susceptibility to Hereditary Breast and Ovarian
Cancer*

Managing susceptibility, the social process identified in this study, broadly describes the genetic testing and risk management decision making experiences of unaffected women who are BRCA positive and variant of uncertain significance (VUS) mutation carriers (hereafter referred to as BRCA mutation carriers). Managing susceptibility pervades all of the actions, from gaining awareness of HBOC risk, getting tested, disclosing results, making risk management decisions, and reflecting on the decisions. Although the theory is categorized, the process is integrated and interdependent. In this study, given the benefit of hindsight, unaffected women who are BRCA mutation carriers described their experience of discovering their mutation and making risk management decisions related to that new knowledge.

Gaining Awareness

The first condition in managing susceptibility, gaining awareness, embodied the preparatory work to managing their risk of breast and/or ovarian cancer. Most participants grew up with an awareness of their susceptibility to cancer because of the loss of a mother, grandmother, or aunt to breast and/or ovarian cancer. This childhood awareness left them feeling cursed and fearful, with a sense that these same cancers were their fate. This is consistent with research on elevated perceptions of risk in women with a family history of breast cancer (Hallowell et al., 1998a, 1998b; Lerman, Kash et al., 1994; Lerman & Schwartz, 1993; MacDonald et al., 2002) and research by Wellisch, Gritz, Schain, Wang and Siau (1991) who found daughters of mothers with breast cancer perceived their chances of getting breast cancer to be much higher than a well-matched

comparison group. Daughters reported that their life courses were altered by their mothers' illness, which had ongoing emotional effects, in particular, integration of the image of a dying mother into a sense of self. Emotional reactions were even more serious for adolescents. As noted in this study, identification with mothers evoked recollection of interruptions in family life during the mother's breast or ovarian cancer experience (e.g. loss of parent and subsequent family disruption).

These heightened perceptions of risk may be explained by Aspinwall and Taylor's (1997) theory of proactive coping, which indicates that once a potential stressor like familial risk is detected, it must be appraised. Heuristics such as salience, accessibility, representativeness, affect, and past experience influence the way in which people interpret potential danger signals and match them to available schemas or scenarios in long-term memory. This is a preliminary effort to understand what a warning sign such as a strong family history may mean. In this study, family history provided salient cues for interpretation of participants' breast or ovarian cancer risk, they were personally relevant, and several histories were dramatic. Assessible cues, such as a mother or sisters recent death or diagnosis, were fresh in their mind, thus a breast cancer diagnosis was ominous. In addition, breast cancer represented loss of life or loss of quality of life based on their past experiences. These experiences with family and friends were incorporated into their risk assessments resulting in a heightened risk perception (Katapodi et al., 2005).

However, a few participants gained awareness of their risk as adults, as the result of a daughter's or sister's cancer. This lack of awareness of breast or ovarian cancer risk occurred when the cancer came from the paternal side of the family or there was a death or divorce which caused the family to lose touch with affected family members. This

sample had more affected maternal relatives than paternal relatives. The asymmetry in reporting of maternal rather than paternal history reinforces previous research which indicated that those with a paternal history of breast or ovarian cancer are very unlikely to have an affected parent and their affected relatives are thus more distant (Green, Richards, Murton, Statham, & Hallowell, 1997).

Consistent with previous research, the most reported reasons that brought participants to BRCA genetic testing were: (a) the presence of a known familial mutation, (b) desire to help other family members by undergoing testing, (c) to relieve doubts about whether a diagnosis of breast or ovarian cancer is or is not likely, (d) to learn about their children's risks, (e) and to make health care decisions to reduce risk (Lerman, Seay et al., 1995; MacDonald et al., 2002; Matthews et al., 2000; Shiloh, Petel, Papa, & Goldman, 1998). In contrast to the study by MacDonald et. al., this work found that reproductive decision making was an element considered by women with BRCA mutation carriers in their twenties and thirties. This difference may be explained by the fact that this previous work dealt with women at high risk based on family history who were speculating on factors considered important.

In addition to an awareness of their increased risk of breast and/or ovarian cancer, participants also received messages from family, the media, and health care providers which brought them to genetic testing. Sixty two percent of participants with positive BRCA mutations sought testing after notification of a mutation in the family. Two with VUSs were tested because a VUS had been identified in the family. For others, it was a family member's diagnosis of breast or ovarian cancer that triggered self or physician referral about a genetic mutation. Although a motivation for testing was to clarify their

own risk and gain more certainty, it was also done for other family members and children.

Confronting Uncertainty and Getting Tested

Confronting uncertainty and getting tested was characterized by seeking further clarification of their family cancer and overcoming professional, insurance, and bureaucratic barriers in the health care system. Lending support to research by Lim et al. (2004), participants believed “knowledge is power,” the knowledge of their BRCA mutation was seen as an advantage. Their concern about high risk had been validated and this empowered them to take necessary action to reduce their risk of breast or ovarian cancer.

When a BRCA mutation had been identified in the family, the family history required was little more than background information in the genetic counseling session. But when the determination of the presence of a BRCA mutation had to be made, the cancer pedigree was essential because it was the data with which the counselor had to work in order to assess the likelihood of a BRCA mutation in the family. Thus the information that participants obtained about their family histories from other family members was crucial and family communication was key. As in research reported by Green et al. (1997) female relatives were much more likely than males to be asked for family history information, supporting the idea that women are the “kin-keepers” in the family. Obtaining family cancer history was at times difficult because of participants’ desire not to upset people with painful memories, having lost touch with a branch of the family due to death or divorce, or not wanting to upset a vulnerable family member with painful memories.

External barriers which participants had to overcome before getting genetic testing included obtaining health insurance, as well as professional and bureaucratic barriers. Lack of health insurance and cost of BRCA genetic testing (nearly \$3000) have been identified in previous studies as factors that influenced uptake of genetic testing in a clinical setting (Geer, Ropka, Cohn, Jones, & Miesfeldt, 2001; Lee, Bernhardt, & Helzlsouer, 2002). As reported by Lee et al. (2002), participants in this study identified fear of insurance discrimination as a barrier to testing. Twenty seven percent (8) of participants chose not to use their health insurance and paid for BRCA testing out-of-pocket for confidentiality reasons.

Women in this study, as well as those presenting at academic centers for BRCA1/2 testing, arrived with a strong belief they had a BRCA mutation (Winer et al., 1997). While awaiting their BRCA test results from the lab, 76% of study participants speculated about their test results. Of these 74% (17) thought they would be positive, while only 5 thought they would be negative. They developed theories about why they thought they would be positive, such as physical or personality characteristics, age, family history, chance, and degree of fear. This rehearsal helped participants to conceptualize their experience and prepare for their forthcoming BRCA test result. In other circumstances, rehearsal has reduced distress and helped with coping after stressful events (Lazarus & Folkman, 1998; Taylor, Pham, Rivkin, & Armor, 1998).

Obtaining Test Results

The majority of participants in this study chose to receive their BRCA test results in person, however three obtained results by phone. As reported in previous research, those who received their results face-to-face from a genetic counselor or physician were

better able to deal with the results (Frost et al., 2004). Receiving BRCA VUS test results in a primary care setting, from other than a genetics professional, was problematic for the only participant who sought care in a primary care setting. This participant was not informed that a variant of uncertain significance was possible, which proved very distressing for the participant and her spouse. All other VUS participants recalled a discussion with their genetic counselor or physician regarding the possibility of uncertain results. However, when it was the genetic counselor's first experience with a VUS, confidence in the advice regarding a VUS result was diminished.

A majority (53%) of participants had a husband/partner, sister, father, or close friend accompany them to the post test counseling session for emotional support. Subtle clues from the genetic counselor allowed some participants to figure out their results in advance. They knew their results were positive by the presence of unexpected professionals in the post-test counseling session, or by noting a difference in how their counseling session was set up in comparison with a sibling with a negative result.

Consistent with previous short and longer-term research, participants described a range of emotional reactions to their BRCA test results: from feeling relief, acceptance, and empowered to shocked, overwhelmed, distressed due to confusion and uncertainty, and feeling vulnerable (Cella et al., 2002; Dorval, Patenaude et al., 2000; Dorval, Paternade et al., 2000; Lerman et al., 1996; Lim, Macluran, Price, Bennett, & Butow, 2004; Schwartz et al., 2002; van Oostrom et al., 2003). Some with a positive mutation felt it was "a gift to know" because it offered an explanation for their family breast and/or ovarian cancer and they could now do something about their susceptibility. Some indicated that if they had been negative, they would still have felt at higher risk than the

average woman. Others were shocked, scared, felt lost, and angry, especially those who thought they would be negative. This lends support to observations from a previous study that mutation carriers, who did not expect to receive positive test results, reported heightened distress after results disclosure (Dorval, Patenaude et al., 2000). After disclosure participants' thoughts raced with questions about the meaning of a mutation for themselves, their children, especially daughters, their husbands, and wondering who to tell, and what it meant for them in terms of their lives and jobs. Testing increased their sense of vulnerability. It provided an awareness that not only were they at increased risk for breast cancer, but an ever more insidious problem, ovarian cancer.

For the nine unaffected women who received a result of "variant of uncertain significance", five rendered it problematic, like a positive result, and four considered it negative or uncertain. Those who interpreted the results as positive or problematic were very distressed by their results, because instead of the certainty they were looking for, they were left with more uncertainty. For participants in the current study, the hard part was "not having a definite answer," which confirms observations by Frost et al. (2004) in a study of affected women with test results of uncertain significance. Some felt they would still get breast cancer with the VUS, that science had just not found other genes in the DNA that was linked with breast and/or ovarian cancer. Others chose to consider it positive so they would remain vigilant in their breast and ovarian cancer screening. For one, it was a physician who rendered the VUS result problematic, considered it positive, and made referrals for a BPO and BPM. Consistent with qualitative research by Frost et al. (2004), dealing with a VUS result was even more problematic when they did not receive genetic counseling to help prepare for this possibility.

Those who interpreted their VUS result as negative and not problematic, chose a wait and see approach. They continued with annual breast surveillance and followed up with their genetic counselor, physician, or Myriad Laboratories to see if new evidence had been found regarding their variant. This interpretation may have been age related, as the mean age of those who rendered a VUS problematic was 36 years, while the mean age for those who did not was 53 years. Perhaps those who rendered it less problematic felt they were beyond the age in which other relatives had developed breast or ovarian cancer.

Shortly after receiving their BRCA test results, participants embraced the finality of their results, their feelings of fear and sadness, shock, and the reality of possibly getting breast or ovarian cancer. Their emotional turmoil and anxiety were not prolonged and decreased over subsequent months, which supports findings in previous research on psychological distress levels after testing in individuals in BRCA families (Butow et al., 2003; Lim et al., 2004; Lodder et al., 2002; Schwartz et al., 2002). Like other healthy at-risk women, they were able to accommodate the information of their increased risk of developing breast or ovarian cancer over time. Deciding to overcome their distress, participants in this study confronted their fears, sought support, and took action they viewed as taking back control of their lives.

Disclosing BRCA Test Information

After obtaining BRCA test results, in an effort to cope with their susceptibility, participants sought emotional support by disclosing their genetic information to others. Findings confirmed other researchers' observations that shortly after testing participants disclosed their results to their husband or partner, sisters, other immediate family, and

closest friends (Claes et al., 2003; Green et al., 1997; Hughes et al., 2002; Richards, 1999; Wagner Costalas et al., 2003). Later on they disclosed to extended family and a few disclosed results to their employers. In addition to seeking emotional support, participants indicated other reasons for disclosing BRCA test information was to inform others of their potential risk, to encourage testing in others, and to get advice about risk reduction measures.

Findings that family factors including pre-existing relationships, patterns of interaction, and tensions acted to promote or hinder disclosure of BRCA genetic information supports other research studies (Claes et al., 2003; Forrest et al., 2003; Hughes et al., 2002). Little and/or superficial contact seemed to be the major subjective barrier to informing distant relatives. Findings confirmed observations by Hamilton, Bowers, and Williams (2005) that disclosure of test results was a deliberate process of selecting family members, the content of information disclosed, and style for telling. Participants carefully considered the family member's receptivity and vulnerability before disclosing. They were pragmatic and not like the 'prevaricator' style of telling found by Forrest et al. (2003) who wait for 'the right moment' and try to squeeze the information into an already scheduled event. They took a more proactive approach and talked about disclosure as a more active, carefully considered, and thoughtful process.

Most unaffected women with positive and VUS test results reported positive aspects of disclosing their BRCA results and overall found their family and friends supportive. Some families felt closer, because of the bond of a common problem or vulnerability. This finding lends support to other researchers' observations that there is a higher degree of support, a connectedness, felt when members of the family were going

through genetic testing together and shared the same results (Smith et al., 1999). Findings also reinforce previous studies which showed that support of the partner, relatives, and friends was important in coping with genetic testing and elevated HBOC risk (Kenen, Arden-Jones, & Eeles, 2004a; Thompson, Gustafson, Hamlett, & Spock, 1992; Wylie et al., 2003).

In disclosing their results to others for support, participants encountered two responses: that of the insider and outsider. Insiders understood the meaning of the mutation and were usually someone who had the experience of a mutation or VUS result and provided emotional support. For participants from families in which a mutation had been identified previously, they usually had insider family members they could speak to freely. Often this was a mother, sister, aunt, or cousin who shared the mutation. They talked about their fears of breast or ovarian cancer, fear or guilt about passing the mutation to their children, adopting, and their experiences with various risk management options.

The outsider did not seem to understand what the mutation meant to the participant and thus support was lacking. Outsiders were found among family and friends, but were noted here in brothers. Kenen et al. (2004a) also found that brothers were more distant and seemed almost in a space apart when discussing HBOC in their families. Research studies indicate that men and women frequently have different communication styles, give and receive various amounts of social support, and differ in their interest and willingness to discuss such matters as cancer (Kenen, Arden-Jones, & Eeles, 2004b; Moynihan, Burton, Huddart, Dearnaley, & Horwich, 2003; Pretorius, 1996).

Although family and friends were sounding boards for participants' feelings about having the BRCA mutation or VUS, there were times they felt alone, isolated by the responses they received. These findings lend support to Foster et al. (2004) observations that support by some relatives is limited by differing opinions about BRCA testing and different ways of coping with the threat. Several participants in this study found what they really needed was to draw on the strength and support of women who had been through similar soul-searching. These insiders had a common ground of shared experiences and concerns and provided a sense of connectedness which helped the participants' fears and isolation to dissipate. They helped to legitimize the participants' feelings and provided a comfortable environment where they could speak out about how they felt. They also understood issues such as employer attitudes about employment and health and life insurance concerns. Participants found insiders in a chatroom on a BRCA support internet website, Facing our Risk of Cancer Empowered (FORCE), the Hereditary Breast and Ovarian Cancer Organization in Canada, National Ovarian Cancer Coalition, as well as hospital-affiliated peer support groups.

These peer support networks helped participants regain a sense of control over their lives and confirmed the decisions they needed to make for themselves and their loved ones. These findings lends support to the need for peer support groups identified by women at high risk for breast cancer in research by Appleton and colleagues (2000). These findings also support previous studies that indicate that professionally led supportive-expressive group therapy facilitates psychological adjustment in cancer and in women with BRCA mutations (Esplen et al., 2004; Goodwin et al., 2001; Spiegel, Bloom, Kraemer, & Gottheil, 1989; Spiegel & Classen, 2003). This is the first study to

date to address the importance of peer support groups in unaffected women with BRCA positive and VUS mutations.

In addition to disclosing their results to obtain support, participants also wanted to inform others. Participants who were the first in the family to be tested experienced a sense of obligation and responsibility to inform others in the family who would possibly share the mutation, for the other's personal sake and the sake of their children. These findings support previous studies (Foster et al., 2004; Green et al., 1997; Hallowell, 1999; Kenen et al., 2004b). Implicit in the participants' message was the need for the family member to find out their risk to minimize their chances of developing breast or ovarian cancer. As in other research, participants disclosed to inform female relatives more than male relatives and to immediate family members more than distant relatives (Claes et al., 2003; Hughes et al., 2002; Wagner Costalas et al., 2003). In addition to communicating their BRCA test results to their family, some wanted to be a resource for others, as they wanted as many people as possible to know about the mutation. Some shared their stories by writing and publishing, speaking to lay and professional groups, participating in chat rooms and in research. By being open they hoped to reduce other women's fear of discrimination with testing.

Those who felt the duty to inform family members indicated it was a sensitive issue and needed to be handled carefully. Findings confirmed other researchers' observations that some people desire as much information as possible about their genetic cancer risks, while others prefer to avoid the information due to the emotional impact on self and/or family members (Geer et al., 2001; Kenen et al., 2004b; Lerman, Peshkin, Hughes, & Isaacs, 1998; Lim et al., 2004). Women in this study had to balance their

sense of obligation to inform family with their family member's need to be protected from upsetting information. Most preferred to disclose the family's HBOC risk, unless there was a reason that inhibited them, such as vulnerabilities due to age, sickness, or to prevent guilt feelings in someone who was terminally ill. The desire to protect family members from harm while disclosing has been noted in other studies (Forrest et al., 2003; Foster et al., 2004; Hamilton et al., 2005; Hughes et al., 2002; Kenen et al., 2004b).

Participants in this study did not anticipate the range or intensity of reactions they experienced while disclosing their genetic information to family members. While some experienced a sense of reconnection with the family, others found their communication blocked, because not everyone, including sisters, are of the same mindset and really did not want the information. Family communication patterns such as open and supportive, directly blocked, indirectly blocked, self censored, and use of third parties were noted, lending support to research by Kenen, Arden-Jones, & Eeles (2004b). Those with VUS results were more uncertain about their own risk and experienced more difficulty in explaining their results to family. Some participants experienced difficulties telling relatives because they worried about its impact on family members and it also reopened their own sense of vulnerability. These findings lend support to research that disclosure of positive BRCA test results to relatives may result in increased psychological distress for the discloser (Costalas et al., 2003; Foster et al., 2004; Lerman, Peshkin et al., 1998; Lim et al., 2004).

Most participants in this study did not disclose BRCA test results to their young children. In comparison, research by Hughes et al. (1999) and Tercyak, Peshkin, DeMarco, Brogan, and Lerman. (2002) found that 50% and 53% respectively of mothers

chose to disclose their BRCA mutation status to pediatric age children (under 19 years of age for Hughes et al. and 8-17 years for Tercyak et al.), with older children more likely to be informed than younger children. Findings support previous studies which showed that disclosure of BRCA results to children is influenced by the child's age and developmental phase, and the parent's philosophy of communication (Segal et al., 2004; Tercyak et al., 2002). In this study, some mothers who did not believe in keeping secrets, or wanted their daughters to share in this experience, disclosed to daughters as young as 9 year old. Consistent with reports by other researchers (Forrest et al., 2003; Hamilton et al., 2005; Kenen et al., 2004a; Segal et al., 2004), there was a desire to do the right thing for children, protecting them for as long as possible, but also knowing they needed to be told in time. Most mothers of young children did not plan on disclosing their BRCA test results until they were old enough to understand the information and faced the potential risk themselves. Participants in research by Segal et al. (2004) felt the 'ideal age' for a child to be told about BRCA mutations was 19 to 25 years.

Although most parents with young children in this study did not disclose their BRCA genetic information, they shared their prophylactic surgical procedures and explained they were done to prevent breast and/or ovarian cancer in the future, usually linking the explanation to a family member's cancer or surgical procedure that the child was aware of. However, when parents decided not to disclose their BRCA results to children, it restricted the number of people they told. This served to prevent information from being inadvertently revealed to children at gatherings of family and friends. Other families agreed to hold the information close to family due to fear of health insurance or employment discrimination.

In summary, these findings indicate that disclosure of BRCA genetic information was a coping mechanism to obtain support from family, friends, and peers. It also was viewed as a mechanism to fulfill a social obligation to inform other family members of their potential risk and encourage testing in others. Disclosing was a complex issue, a family affair, and was influenced by both pre-existing familial communication patterns and the individuals' understanding of and responses to their susceptibility for hereditary breast and ovarian cancer. Disclosing of their BRCA carrier status was an important step in breaking down participants' feelings of fear, isolation, and vulnerability so they could proceed to further manage their susceptibility to breast and ovarian cancer.

Making Risk Management Decisions

Although participants experienced a wide range of emotional reactions after learning their BRCA carrier status, their disclosure to family, friends, and peers seemed to provide the support they needed to cope with their fears, regain control, and gain a sense of mastery over their fear of breast and ovarian cancer. Findings reinforced previous longitudinal studies that indicated BRCA testing generated specific concerns and psychological reactions, however there were minimal adverse psychological effects (Appleton et al., 2000; Croyle, Smith et al., 1997; Cull et al., 1999; Dorval, Paternade et al., 2000; Lerman et al., 1996; Schwartz et al., 2002).

In considering risk management options of prophylactic breast and ovarian surgery, chemoprevention, and vigilant surveillance, participants were choosing between two outcomes, either to reduce their risk of breast and ovarian cancer or "catch it early." Risk reduction options included prophylactic surgeries and chemoprevention with tamoxifen. Early detection ("catch it early") involved annual vigilant breast and ovarian

screening. Both the certainty and uncertainty of their BRCA test results set in motion a series of actions and interpretations leading to risk management decisions. This set of actions included seeking information, drawing on resources, deliberating and making decisions, and reflecting on actions

After obtaining information from their genetic counselor and physician on risk management options available, participants sought information from both lay and professional sources to assist them in making risk management decisions. For some, their physician's non-directive stance, not prescribing what to do, left them disappointed because they felt they must make a decision when they did not have the requisite information. Most participants had an information seeking coping style, as they expressed their belief that "knowledge is power" and the "key to life-saving decisions". Findings support previous studies that indicated alternatives that were not immediately salient (preferred) tended to stimulate a need for more information or consultation, so the participant was better able to evaluate it (Slovic, 1975; Tversky, Sattath, & Slovic, 1988). Participants sought more information about the surgical options, especially breast reconstruction options. Knowledge gained in information seeking was effective in alleviating the bewilderment they felt in discriminating between the risk management alternatives. These findings lend support to research by Pierce (1993; 1996) who found that breast cancer participants making decisions for surgery sought information when they were unable to discriminate between alternatives and experienced conflict.

Participants sought information from various sources, some preferred obtaining information only from professionals in clinical settings, while others used written materials ranging from scientific reports to popular magazines. The use of the internet for

scientific reports and online support groups, such as FORCE was prevalent. Participants emphasized how valuable it was to meet with or talk (by phone or in chat rooms) with women who had similar histories, had been through the risk management decision making process, and openly shared their knowledge and experiences about issues they were working through. These support networks provided links to research and other internet sites for gathering information about their risk management options and were knowledgeable referral sources for genetic professionals, oncologists, gynecologists, and plastic and breast surgeons. Peer support networks helped participants regain a sense of control in their lives and provided reassurance about the decisions they were considering. These findings confirmed other researchers observations that social networks help alleviate the damaging effects of stressful life events by providing potential coping resources such as emotional, informational, and practical support (Fawzy, Fawzy, Arndt, & Pasnau, 1995; Leszcz & Goodwin, 1998).

In making risk management decisions, participants drew on the resources of their life experiences and relationships with others. Their decisions were based on a broad set of values and beliefs about the perceived effects of risk management on their personal lives and their families. Past family and personal experiences, present sense of self and their relationships, and aspirations for the future were all part of the participants' decision making considerations. These findings reinforce previous studies which reported that patients' decisions to accept treatment were personalized, to match their views of themselves within the context of their life stories (Hallowell et al., 2004; Kelly-Powell, 1997). Pierce and Hicks (2001) indicate that individuals make decisions filtered through a personal understanding of the problem and potential solutions. For several participants,

risk management decision making resurfaced memories of interruptions in family life during an earlier cancer-related experience of their mother, sister, or daughter. Also, the sense of loss associated with their susceptibility to breast and ovarian cancer impacted their concept of current and future self. Concepts used by other researchers to demonstrate the perceptions of participants regarding the effects of risk management on everyday life and reliance and trust on health care providers were salient (Appleton et al., 2000; Frank, 1990; Hallowell, 2000; Hallowell et al., 2004; Lim et al., 2004; Pierce, 1996; Ward, Heidrich, & Wolberg, 1989).

Most participants sustained personal relationships by involving their husbands/partners and other family members in their decision making. They described both supportive and unsupportive responses from extended family members to their decisions about risk management, especially prophylactic breast and ovarian surgery. They sought various levels of input about the risk management options from family, but most felt in the end it was a personal choice. They described their spouses as supportive, which made decision making easier and helped reduce their distress. This lends support to findings by Wylie et al. (2003) which showed that the role of spouses in BRCA mutation carriers' social support system is significant. Perception of their spouse's anxiety and spouse's support at the time of testing were predictive of the women's psychological distress up to 2 years after BRCA testing. The interaction of the two variables were even more predictive. Findings from this study further support the importance of family context in understanding risk management decision making for susceptibility to HBOC (Lim et al., 2004).

After obtaining sufficient information about risk management options and drawing on personal and professional resources, participants' began deliberating, examining the alternatives to select a particular option. For some, decisions were immediate while others were more deliberative, depending on the salience of the alternative and its respective attributes.

Four decision making patterns emerged: acting on apriori decisions, following expert advice denovo, following some and rejecting other advice, and postponing the decision until later. These patterns were related to the way participants approached the decision, gathered information, and the amount of control they preferred in decision making. Two of these patterns are similar to styles of decision making identified by Pierce (1993; 1996), that of the deferrer and deliberator. Deferrers selected an alternative with relative ease, choosing a recommendation made by their physician, deferring to his or her expert judgment. Participants' reports indicate that their physician's presentation of risk management options may have influenced their choice through framing effects, as has been reported in previous studies (Llewellyn-Thomas, McGreal, & Thiel, 1995; Pierce, 1993; Tversky & Kahneman, 1981). It is important to note here that as new medical evidence on the effectiveness of the risk management options for BRCA carriers evolved during the period participants were making decisions, physicians' knowledge and willingness to make recommendations also may have changed.

Deliberators were similar to participants who 'followed some and rejected other advice'. These participants' decision making was similar to the normative models of decision making and resembled the vigilant decision maker identified by Janis and Mann (1977). They carefully considered the risk management alternatives, identified

advantages and disadvantages of the alternatives, weighed attributes based on personal preferences, and validated the information with expert consultation.

However, two styles were dissimilar to Pierce's (1993) research. One, 'postponing a decision until older', was not similar to the delayer identified by Pierce, as the decision to postpone prophylactic surgery(ies) included the decision to comply with vigilant breast and ovarian cancer surveillance at the present time. It was not because they could not arrive at a decision because the alternatives were close together in a valued dimension. In this study, participants' plans for surgery(ies) were delayed until they had completed a family or an older chosen age, based on when a mother or sister had developed breast or ovarian cancer, or an age recommended by their genetic counselor or physician.

The fourth style, 'acting on apriori decisions', did not have a corollary in this previous work (Janis & Mann, 1977; Pierce, 1993). These findings, however, support previous research in breast cancer in which Hughes (1993) found that treatment selection was related to the amount of information subjects received prior to their clinic visit from informal sources such as friends, family, and media. This suggested that patients' treatment decisions may be influenced or biased by early information, regardless of the source. These findings also confirmed other researchers' findings that some participants had been strongly considering their chosen risk management option before genetic testing and the additional BRCA test results provided the certainty that this was the right choice (Lerman, Daly, Masny, & Balshem, 1994; Meiser, Butow, Barratt et al., 2000).

Most participants chose risk management options they perceived gave the greatest risk reduction and thus enhanced their chance of living out their lives. Other important

elements were the participants' values and beliefs about their quality of life and its impact on their children should they develop breast or ovarian cancer. Testing and risk management allowed them choices their family members did not have.

Sixty percent of participants (positive and with VUS) chose a prophylactic surgery options (BPM or BPO). Seventy five percent of those with positive mutations and 33% of those with variants of uncertain significance chose one of the prophylactic surgical options. Forty percent of participants chose both surgical options. Positive mutation carriers clearly preferred prophylactic surgery over early detection measures to reduce their susceptibility to breast and ovarian cancer. Although the preference for prophylactic ovarian surgery over early detection to reduce the risk of ovarian cancer has been reported in other studies (Lodder et al., 2002; Meijers-Heijboer et al., 2000; Scheuer et al., 2002), this has not been the case for prophylactic breast surgery in the United States where choice for BPM has ranged from 0%-15% in unaffected carriers (Botkin et al., 2003; Lerman et al., 2000; Scheuer et al., 2002). However, two studies in Holland (Lodder et al., 2002; Meijers-Heijboer et al., 2000) found that 51%-54% of unaffected female carriers had a BPM within 2 years after testing (50% - 64% had a prophylactic oophorectomy). Variability in uptake of BPM may be explained by differences in recommendations across locations, cultural differences, population characteristics across studies (differences in age distribution), value differences toward body integrity, femininity, and preventive surgery, and differences in health care funding systems in different countries. In addition, collectively all previous studies reporting on uptake of prophylactic surgeries in unaffected carriers, recruited participants from 1994-2000 when

research reports on the effectiveness of BPM and BPO in unaffected mutation carriers were not available. Also, most studies report on following women for only 12-24 months.

As reported in studies of high risk women, factors that influenced participants' prophylactic surgical decisions included their sense of susceptibility (subjective risk perception) to breast or ovarian cancer (Hallowell, 1998; Hallowell, Jacobs, Richards, Mackay, & Gore, 2001; Hatcher et al., 2001; Hurley, Miller, Costalas, Gillespie, & Daly, 2001; Meiser et al., 1999; Stefanek et al., 1995), sense of family obligation (Hallowell, 1998), witnessing a mother, sister, aunt, or daughter's experience of breast or ovarian cancer (Hallowell et al., 2001; Hatcher et al., 2001), age, fertility, and menopause (Hallowell et al., 2001; Meiser et al., 1999; Tiller et al., 2002), fear related to body image changes (Hallowell, 1998), breast reconstruction availability (Contant et al., 2002), and fear of surgical procedures and complications (Hallowell, 1998; Hallowell et al., 2001). Findings that several participants choosing prophylactic surgery(ies) regarded screening modalities for ovarian cancer (transvaginal ultrasound and CA 125 testing) and mammography for breast cancer as having limited predictive power, and wanted to avoid the anxiety associated with yearly screening, confirmed other researchers observations (Lerman et al., 2000; Tiller et al., 2002).

Thirty seven percent of the women (5 BRCA positive and 6 with VUS) chose vigilant surveillance. This included women in their twenties and thirties who were delaying their decision until they completed childbearing and breastfeeding. Four (3 BRCA positive and 1 with a VUS) in these age groups indicated their plans for prophylactic surgery in the future. In the interim they chose vigilant surveillance. Trust in the competence of their physician, use of MRI, and hope for new research discoveries

were influential factors in their decision for vigilant surveillance. They found reassurance in their belief that with surveillance, if they developed cancer, it would be found early. It is important to note that the advantages and disadvantages of the surgical options identified by those choosing vigilant surveillance were similar to those choosing prophylactic surgery(ies). However, how they weighed the advantages and disadvantages of the alternatives, based on values, beliefs, and desired outcomes were different.

Unlike previous studies these women reported adhering to recommended breast and ovarian cancer screening guidelines (Lerman, 1998). Perhaps this is because participants' psychological distress in this community sample was not as great as those in earlier studies conducted in research institutions. This also may be due to personality characteristics or style of coping with threatening information.

Only two participants chose the option of chemoprevention with vigilant surveillance. Participants received less information about this option and it was not usually recommended by their health care providers. This finding may be partly related to the lack of scientific evidence of the effectiveness of tamoxifen in women with BRCA mutations (King et al., 2001). Participants did not identify any advantages of taking tamoxifen, but delineated several disadvantages; including risk for uterine cancer, cataracts, and thrombosis, its experimental nature in BRCA carriers, and insufficient risk reduction. Several younger women did not choose this option because of implications for childbearing.

Cameron and Leventhal (1995) have suggested that perceived control over disease influences beliefs of vulnerability. Most women in this study believed they had some control over their susceptibility to breast and ovarian cancer and some women believed

that adherence to vigilant surveillance and having a close relationship with a health care provider could result in early detection and thus survival from breast or ovarian cancer. These findings lends support to research by Katapodi, Facione, Humphreys, and Dodd (2005) which indicated women that had a trusting relationship with their health care provider had a sense of control over breast cancer and perceived they were at a lower risk.

Making a risk management decision was a strategy to manage susceptibility as well as a consequence of their knowledge and past and present life experiences and relationships with others. Decision making involved consideration of both individual and social factors. Participants balanced the gains of risk reduction, relief of breast and ovarian cancer worry, and fulfilling their obligations as mother and wife to remain cancer free, against the potential losses of surgery; such as menopause, infertility, changed body image, the continuing risk of developing ovarian and breast cancer in residual tissue, and surgery(ies)' effect on family and employment. Whether participants arrived at a decision based on apriori decisions, followed the expert advice of their physicians, through deliberation - accepting some and rejecting other advice, or postponed treatment until older, they reflected back to evaluate their decisions and put things in perspective.

Reflecting on Actions

All 30 participants expressed satisfaction with their decision to have genetic testing and indicated they would do it again, as well as recommend it to other women at high risk for breast and ovarian cancer. In addition, those with prophylactic surgeries expressed satisfaction with their decision choice(s), despite some difficulties with breast reconstruction, which is consistent with finding from other studies (Frost et al., 2000;

Hatcher et al., 2001; Lloyd et al., 2000; Meijers-Heijboer et al., 2000; Stefanek et al., 1995; Tiller et al., 2002). A few experienced complications after surgery, such as a “frozen shoulder,” pain due to overstretching the capsule during saline injections, and one described replacement of her implant due to an infection. Overall, it tended to take participants longer than they had expected to recover from surgery. Findings of a sense of relief from the fear of developing breast and/or ovarian cancer after prophylactic surgery supports a 5 year follow-up study of BRCA carriers by van Oostrom and colleagues (van Oostrom et al., 2003). Findings also reinforce previous research findings that high risk women perceived the benefit of anxiety reduction outweighed the potentially adverse effects of prophylactic surgical procedures (Tiller et al., 2002).

An additional finding was the importance of social context in determining participants’ experience of prophylactic surgery. Although telling others in the family about their decision for surgery was motivated by a desire to obtain support and reassurance, for some it brought strong emotional reactions, adverse opinion, and lack of support. As a result, some decided not to tell certain family members or reduced the information communicated. This meant their support network was decreased, which contributed to feelings of isolation. Several developed contacts with BRCA carriers in peer support groups who shared a similar experience.

Participants viewed their decisions as personal, supported by professional and peer support groups, but unsupported in follow-up. Most indicated there was not a cohesive plan for follow-up; they just did not fit, as they did not have cancer, had prophylactic surgeries, and the medical systems they encountered did not know what to do with them. They had to be self directed and take personal responsibility, as there was a

lingering uncertainty about the small risk of peritoneal cancer or cancer in residual breast tissue. This finding regarding follow-up has not been reported in other literature on unaffected positive or VUS mutation carriers. In reflecting on the consequences of their decisions participants indicated they had achieved peace of mind, were altruistic and working to “pay it forward”, experienced strengthened family relationships, and those who chose screening, remained vigilant.

Conclusion

In this age of genetic technology and discovery of new genes, it is important to continually assess the impact of this technology on the lives of those who engage in them. The women who participated in this study regarded breast and ovarian cancer as a predictable outcome, given their family history, and felt they had a responsibility to their family to prevent this danger if possible. Their attendance at genetic counseling was the first step in taking responsibility for their perceived susceptibility and was influenced by feelings of obligation to their children or other family members. Their BRCA information was perceived as information about the family. Thus participants disclosed their test results because of a sense of duty to inform their family members of their risks and risk management, no matter how difficult it was for them personally. They also felt they had a responsibility to persuade their family to act on the information. Their risk management decisions stemmed from a feeling of duty to children, having a responsibility to remain healthy so they could nurture their children and protect them from seeing a mother die or having to care for them. Engaging in risk management was seen as providing them with control over their susceptibility to breast and/or ovarian cancer. Those choosing prophylactic surgeries wanted to prevent cancer, as they were not satisfied with the

limitations of vigilant surveillance which provided only early detection of cancer. By taking these measures they not only gained some control over their lives, but as importantly, could maintain their identity as mother and nurturer of others. Their strengths and abilities may serve as indicators needed for successful coping with BRCA genetic information.

The similarities of previous studies related to genetic testing and risk management serve to validate this study. This discussion of the findings presented the shared discoveries and added the unique experiences of unaffected positive and VUS mutation carriers, to provide a cohesive theory of managing susceptibility to hereditary breast and ovarian cancer.

At this point, I am including two powerful and poignant poems, written by two participants which so eloquently express the experiences of women managing their susceptibility to hereditary breast and ovarian cancer (see Figure 3). May these be cogent reminders to each of us of the need for emotional support as we continue to grope, cope, and understand our humanity, as life is lived in our genomic age.

The final chapter provides a critique of this study and offers implications and recommendations for health policy, nursing practice, and research.

Legacy of Fear

“Are you scared, Mom?”
 My seven-year-old daughter asks.
 “Yes, honey, a little bit”, I respond,
 Trying to keep my voice sounding calm and reassuring.
 More than you’ll know I think, quietly, inwardly...

My fear comes in so many ways.
 That my beautiful daughters, now seven and ten,
 Will know this fear, just as I have.
 Just as my mom, my sisters, my brother, my husband have.
 More than anything,
 I prayed that this would not be there for them.
 But I can’t stop it
 And the fear continues.

It feels like a time bomb, waiting to explode in my life.
 Should I have children? ...tick, tick...
 Did I book my mammogram?...tick, tick...
 Should we have another child?tick, tick...
 What was that I felt in my breast? ...tick, tick...
 Do I need life insurance? tick, tick...
 What about the ultrasound? ...tick, tick...
 Should I get tested? tick, tick...

And we wait
 And we wait
 And we wait

BRCA1 – my fear has a name.
 I hate this name, but now there is a face to it.
 Part of me knew it would be there.
 I can look this fear in the face now.

I hate you.
 I hate what you have done to my beautiful mother, my beautiful sister, and my
 beautiful daughters.
 I hate what you have done to me.

But I have power over you now.
 I will cut you out of me
 And you will not hurt me anymore,
 Or scare me.
 You will not explode in my body.
 I will win.

“Mommy, are you scared?”
 Yes, my beautiful, precious, innocent daughters.
 For you, I am scared.

“Will this happen to me?”
 My daughter asks.
 Tick, tick, tick....
 FKJ December 2002

Figure 3. Participant’s Poem: Legacy of Fear

Skin
Stretched
Over saline sacs
Breasts
They call them
Reconstructed

Numb
Cold, scarred
But in a breast-obsessed world
They succeed
To deceive
Them

I remember
Her small hungry mouth latching on
The urgent sucks slowing
Her tiny fingers'
Soft touch
Sleeping

Warm
Wet kisses
Breasts rising, responding
Reaching for his touch
Naked skin embracing

Feeling
Gone now
To save my life

I remember
And I cry
Joanne
October 25, 1999

Figure 3. Poem of BRCA 2 positive participant coming to terms with her BPM and Reconstruction

CHAPTER VII

IMPLICATIONS AND RECOMMENDATIONS

Growth in our knowledge of the human genome over the last decade has resulted in the availability of genetic testing for familial susceptibility to hereditary breast and ovarian cancer. Knowledge of genetic mutation status potentially provides high risk women with the needed information on which to make decisions about options to reduce their breast and ovarian cancer risk. This study has formulated a grounded theory of Managing Susceptibility, reflecting the experiences of unaffected BRCA carriers' testing for hereditary breast and ovarian cancer and subsequent risk management decision making. This chapter provides a discussion of the critique of this study and offers implications and recommendations for health policy, nursing practice, and research.

Critique of the Study

Strengths

A strength of this study, over previous studies related to BRCA mutation testing and risk management, is that it was a community sample, from 14 states and Canada. It represented unaffected BRCA carriers' experiences in several different cancer genetics institutions, not just in a single research institution. The sample size was sufficiently large to allow saturation of categories to be reached and the purposive sample of women

reflected a range of perspectives. Other strengths include the range of ages of participants, from 22 years to 60 years. This age diversity of unaffected women participants served to enhance the validity of the resulting theory. Women in this study were unaffected prior to testing, thus preventing confounding by cancer status. In addition, this study explored genetic testing and risk management decision making in both BRCA positive unaffected women and those with BRCA VUS results, which have not been reported previously in the literature. Furthermore, it explored the impact of time since receiving test results and making a risk management decision, as some women had received their test results and had their surgery several years ago, while others were in the process of their decision making (median year tested - 2002, range 1994-2004; median time from testing until BPM surgery - 10 months, range 1-40 months; median time until BPO - 6.9 months, range 0-27 months).

The methodology enabled rich data to be gathered, with findings derived from participants' own accounts. Additionally, adherence to methodology to establish trustworthiness was a strength of the study. Credibility was established by using consistent interviewing techniques, using reflexive analysis, keeping methodological and theoretical memos, and using peer examination. Peer examination and discussion of the data also helped to achieve dependability and trustworthiness. Transferability was enhanced by providing dense background information to allow others to make comparisons.

Limitations

The predominance of Caucasian, well educated participants of middle to upper middle income status, who volunteered to be interviewed might be considered a

limitation. However, due to the cost of BRCA testing and risk management options, this represents the socio-demographic features of women in general seeking genetic testing. The interview data represented retrospective accounts and was influenced by what participants remembered. However, women were able to give rich and relatively unprompted accounts of their experiences of both their BRCA testing and decision making experiences. While the high decision satisfaction for BRCA testing and risk management options chosen may be real, it may also be due to positive response bias from cognitive dissonance. This phenomenon has been documented in unvalidated patient satisfaction measurement (Carr-Hill, 1992) and is relevant to surgical decision making (Homer, Sheard, & Jones, 2000).

Implications

Implications and recommendations are indicated in the following three areas: health policy, nursing practice, and further research.

Health Policy

Findings suggest that participants were concerned about both insurance and employment discrimination, to the extent that some participants paid for the testing out-of-pocket rather than use their health insurance. Fear of health insurance discrimination represented the greatest barrier to utilization of cancer genetic counseling in a study by Geer et al. (2001). Legislation protecting individuals from genetic discrimination by health insurers and employers might help clients feel that they and their relatives will not be harmed by seeking BRCA genetic testing. Although recent federal and state legislation, such as the Health Insurance Portability and Accountability Act (HIPAA) of 1996, have begun to provide protections against genetic discrimination in health

insurance, more needs to be done. The HIPAA prohibits group health insurance plans from treating most genetic information as a preexisting condition and from using genetic information to determine eligibility (Anderlik & Lisko, 2000). However, it does not apply to those not insured under group plans, e.g. individual plans. It also does not prohibit other discriminatory practices, such as mandatory testing, raising premiums, or setting caps on insurance, if these conditions are equally applied to all persons enrolled in the plan (White, Callif-Daley, & Donnelly, 1999).

To prevent genetic discrimination in insurance and employment, federal legislative reform is needed. Nurses can advocate for such legislative reform through the legislative arms of their professional nursing organizations. Current legislation, Senate Bill 306, the Genetic Information Nondiscrimination Act passed the Senate in February, 2005. However, the companion U. S. House of Representatives bill, HR 1227, was introduced in the 109th session but awaits consideration in committee. This bill seeks to create a national standard to bar genetic discrimination by health insurers and employers (Genetics and Public Policy Center, 2005). Similar legislation has been introduced in previous sessions of Congress, but has not made it to the joint conference committee. Since genetic discrimination has been less of a threat than anticipated (Hall & Rich, 2000a, 2000b), education of clients about this information may help remove barriers to genetic testing services.

Findings of this study also suggest that individual economics played a role in decision making both for genetic testing and risk management options chosen, which confirmed other research (Geer et al., 2001; Olopade, 1996). Policy makers must recognize the implications of employing technology that is costly to families who seek

this testing. As discussed, some of the participants and their providers spent a good deal of time negotiating with insurance payors to cover costs of testing and surgical treatment. Future goals should include the provision of funds for genetic testing and risk management options by third party payors, including Medicaid. Nurses can advocate for just distribution of health and insurance costs, so that people are not penalized for genetic attributes over which they have no control.

Nursing Practice

Although the value of genetic counseling by cancer genetics health professionals in preparing for genetic testing is supported by participants in this study, counseling did not fully prepare them for the emotional strain they would experience in making risk management decisions. Few indicated there was contact with the genetic counselor or other genetics health care professional after disclosure of their test results. After testing, participants sought information from multiple sources regarding risk management and indicated that putting together a surgical and oncology team was difficult work, like “pushing a boulder up a hill.” A coordinated effort between the genetics, oncology, surgical, and nursing teams to provide informational support is important to prevent further distressing these women during their decision making experiences.

Emotional care must also be provided for unaffected BRCA carriers as they take on risk management decisions. Many of these women, at least initially, were unaware of support systems outside their immediate family and had to discover support systems independently. The prevalent use of peer support groups to obtain emotional support and information about risk management options underscores the need for professionally led supportive-expressive group support programs for BRCA positive and VUS carriers, to

facilitate psychological adjustment and decision making (Espen et al., 2004). Advanced practice nurses in genetics can conduct such groups, providing information and clarifying or correcting factual misinformation during group sessions.

Study findings also stress the importance of long-term follow-up for both breast and ovarian cancer for unaffected carriers who have chosen the prophylactic surgery(ies) risk management options. Advanced practice nurses, in concert with other health care providers, need to develop a comprehensive approach to follow-up for unaffected BRCA carriers within their health care facilities.

The observation that participants speculated about their BRCA test result, which helped in preparing for their test results, may indicate there is a role in counseling to help individuals predict their results and reactions, to protect against emotional stress. Identification of those who anticipate negative results, but receive positive results, will allow for targeting of those individuals for further evaluation and counseling and for follow-up. Those women with variants of uncertain significance who render their results problematic may experience more distress and need follow-up evaluation and counseling.

Communication about BRCA test results within the family should receive special attention during counseling, especially when tested clients are the initial messengers. It is important that clients give consideration to whom and how they will communicate this information to their family members. It is important that they are able to provide correct information to their family. This study found that variant of uncertain significance test results are not easily understood and consequently difficult to communicate. Even when patients can recall some of the counseling information, subtle personal communication may color the communication. These findings reinforce that written material from genetic

counselors may be a helpful aid in this process (Green et al., 1997; Hallowell & Murton, 1998; Richards, 1999).

Training of advanced practice nurses and other health care professionals needs to include information regarding the impact of BRCA test results on risk management decisions, particularly the impact of variants of uncertain significance test results. This study found that some VUS carriers perceived their test result as positive, others rendered them negative. These perceptions impacted their risk management decision making and long term follow up.

Future Research

The experience of risk management decision making in unaffected positive and VUS BRCA carriers remains a fertile area for further research. First, studies could examine decision making with a larger sample of unaffected positive and VUS carriers. With more statistical power, significant differences between positive and VUS carriers may be revealed regarding disclosure of test results and risk management decision making. All of the women in this study volunteered to be interviewed and voiced satisfaction with their decisions. The voices of women who do not feel satisfaction with their decision making needs to be contrasted with those who were satisfied. Another modification would be a prospective study to examine decisions of young unaffected women over time. This study used data from participants from 22 to 60 years of age. Looking at decision making across multiple time points, such as age 35 and 40 years in those diagnosed with BRCA mutations in their twenties and early thirties, would reveal whether those who indicated their desire for prophylactic surgery in the future, after having children and meeting family obligations, have changed.

There are limited data on uptake of genetic testing and risk management decision making among nonwhite populations. Further research is needed to define factors influencing testing and risk management decision making in these populations. The decision making styles of other ethnic groups may reflect differing viewpoints on the same issues or other issues may emerge in the risk management decision making experience.

Future research is needed to define which individual or familial qualities encourage adaptation and mastery by those undergoing genetic testing. Results of this study and that of Dorval (2000) suggest that the ability to anticipate accurately one's distress reactions to test disclosure may be a predictor of psychologic outcomes after genetic testing. Identification of those at increased risk for heightened distress after disclosure will allow for targeting those individuals for further evaluation. Future research is also needed to examine the use and effectiveness of internet and online chatrooms to provide emotional and informational support and help women make sense of their experience of susceptibility to HBOC.

This study of BRCA genetic testing and risk management raises questions about the process and content of communicating with children about cancer risks, and the impact of these communications on parent-child well being. Research is needed to evaluate the process and content of post-test disclosure and the impact on participant, child, and family functioning. Given the complexities of risk management decision making and psychological adjustment associated with genetic testing, a better understanding of the consequences of disclosure to family members can help nurses and other clinicians provide better counseling to these individuals. Research is also needed on

the impact of BRCA positive and VUS results disclosure on relationships with relatives. Research from the perspective of relatives who do not want the genetic information is needed to better understand their perceptions about informing family members and factors influencing their decisions to block disclosure.

All the women who volunteered for this study were unaffected with breast or ovarian cancer prior to testing. Most had made a risk management decision and had moved on with their lives. The decision making voice of women affected with cancer, before genetic testing, needs to be contrasted with these unaffected women. The development of quantitative decision making aids or tools with subsequent intervention studies also present further options.

In summary, indications for changes in health policy, nursing practice, and further research have been identified. Health policy must be changed through federal legislation to prevent insurance and employment discrimination. Legislation protecting individuals from genetic discrimination by health insurers and employers might help clients feel that they and their relatives will not be harmed by seeking BRCA genetic screening. Policy makers must recognize the implications of employing genetic technology that is costly and provide for funding of genetic testing and risk management options by third party payors, including Medicaid. Advanced practice nurses must recognize the informational and emotional support needs of women seeking BRCA testing, during disclosure and during risk management decision making, and ensure a coordinated approach to follow-up care. Lastly, research possibilities include risk management decision making in unaffected carriers of other cultural and economic backgrounds, as well as women affected with cancer prior to genetic testing. Future research is needed to define which

individual or familial qualities encourage adaptation and mastery by those undergoing BRCA genetic testing. In addition, the development of quantitative decision making aids or tools with subsequent intervention studies present further options.

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