

ABSTRACT

Title: NEGOTIATION OF HEALTH RISKS AND RISK MANAGEMENT FOR YOUNG ADULT *BRCA1/2*-POSITIVE WOMEN: IMPLICATIONS FOR PARTNERING AND FAMILY FORMATION

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In the mid-1990s, genetic testing was introduced for two breast and ovarian cancer predisposition genes: *BRCA1* and *BRCA2*. For mutation carriers, lifetime risks of breast and ovarian cancer approach 90%, and 54%, respectively, *versus* general population risks of 12% and 2%. Women testing positive for mutations during young adulthood face numerous challenges related to navigating basic life course tasks, including establishing permanent couple relationships, family formation, and risk-management decision-making. These complex choices require young carriers to balance personal and family desires and provider recommendations for health maintenance and disease avoidance against their own desires/plans for personal, relationship, and family fulfillment. How they accomplish these tasks is significantly influenced by their experiences with cancer in close relatives, personal cancer risk assessment, and partner presence and support. Although the experience of older carriers and cancer survivors has been thoughtfully explored, little attention has been paid to the experiences of younger women.

Using qualitative methods and grounded theory, I analyzed in-depth interviews with forty women aged 18-35 who knew themselves to be *BRCA1/2*-positive. Participants

were recruited from: 1) an ongoing NCI Clinical Genetics Branch Breast Imaging Study; 2) the membership of FORCE, an online support network for mutation-positive individuals; and 3) snowball sampling, whereby participants referred others whom they knew to be mutation-positive. Using a semi-structured, open-ended interview format, participants were asked about family relationships and background; couple relationships; experiences and perceptions regarding family formation; and experiences and perceptions related to utilization of risk management strategies over time (*i.e.*, surveillance, chemoprevention, risk-reducing surgery). Questions were developed using sensitizing concepts from the biopsychosocial perspective on health and illness and life course perspective, as well as attachment and feminist theories. Interviews were conducted by telephone, digitally recorded and transcribed, and analyzed using QSR N-Vivo software, version 8. The data-driven model indicates that risk perception and management decisions are closely tied to family and couple relationship experiences. Young mutation carriers aggressively and courageously utilize agency to alter their life trajectories while minimizing sacrifice to their family and relationship ideals, thereby freeing themselves from mutation-related emotional and physical constraints, and regaining control over their genetic destiny.

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IMPLICATIONS FOR PARTNERING AND FAMILY FORMATION

by

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Dedication

For “Aaron,” “Acacia,” “Ally,” “Anita,” “Annie,” “Audrey,” “Beth,” “Branson,”
“Brooke,” “Charlotte,” “Chris,” “Danielle,” “Dawn,” “Elaine,” “Elisa,” “Ellen,” “Ellie,”
“Gail,” “Grace,” “Heather,” “Isabelle,” “Jane,” “Julia,” “Kate,” “Kay,” “Kristy,”
“Leigh,” “Libby,” “Lilly,” “Linda,” “Lynn,” “Madaline,” “Maelie,” “Marie,” “Marjory,”
“Maryann,” “Maureen,” “Melanie,” “Michaelina,” “Monique,” “Nichelle,” “Noelle,”
“Pam,” “Pauline,” “Rachel,” “Reina,” “Rose,” “Ruby,” “Ruth,” “Rylan,” “Sadie,”
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speaking with you all were fascinating beyond description; at times joyful, at times
heartbreaking; and always left me eager to find out what will happen next in your lives.
Each of you has enriched my understanding of what it means to live with knowledge of
risk and in families so deeply touched by breast and ovarian cancer. You taught me a
great deal about strength, perseverance, courage, and grace. If I have conveyed here even
a small portion of what I learned from you, then together we have made the world a better
place.

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clinician. I have always appreciated your faith in me and your encouraging me to pursue my passion. I hope to someday emulate your outstanding teaching, mentoring, and the balance you bring to your lives.

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CHAPTER 1: INTRODUCTION

Mutations in *BRCA1* and *BRCA2* predispose carriers to increased risk of developing both breast (50-85% lifetime risk) and ovarian cancer (11-54% lifetime risk) (Antoniou et al., 2003; Brose et al., 2002; M. C. King, Marks, & Mandell, 2003; Suthers, 2007); the corresponding risks in the general population are 12.8% and 1.5%, respectively (DevCan, 2005; Fay, 2004; Fay, Pfeiffer, Cronin, Le, & Feuer, 2003). Individuals with mutations in *BRCA1* and *BRCA2* comprise a small but notable proportion of all breast and ovarian cases, about seven in 100 cases of breast cancer in women with a family history (Ford, Easton, & Petro, 1995) and approximately 10% of all ovarian cancer cases, with more of these resulting from *BRCA1* (about 5.7%) than *BRCA2* (about 3.8%) (Boyd, 2003; Claus, Schildkraut, Thompson, & Risch, 1996; Pavelka, Li, & Karlan, 2007; Prat, Ribe, & Gallardo, 2005). However, the proportion of cancer cases in *young* women attributable to mutations in *BRCA1/2* paints a very different picture: these mutations explain

approximately 33% of [breast cancer] cases age 20-29 years compared with approximately 2% of cases age 70-79 years. The proportion of ovarian cancer cases predicted to be due to the susceptibility gene ranges from 14% among patients diagnosed in their 30s to 7% among those diagnosed in their 50s (Claus et al., 1996, p. 2318).

The prevalence of *BRCA* mutations in the general population is quite low; for example, the gene frequency of *BRCA1* as been estimated to be between 0.0006 (Ford et al., 1995) and 0.002 (Struewing, Lerman, Kase, Giambarresi, & Tucker, 1995); exact population prevalence for these genes is difficult to determine because widespread screening is impractical (Brody & Biesecker, 1998). While the overall carrier frequency for *BRCA* mutations is thought to be about 1 in 280, it is significantly higher in several

distinct populations. In the Ashkenazi Jewish population, carrier frequencies of approximately 1 in 40 are attributed to the prevalence of three founder mutations, two in *BRCA1* and one in *BRCA2* (Boyd, 2003). Additionally, among native Icelanders, about 0.4% of the general population carries a specific mutation in *BRCA2* (Johannesdottir et al., 1996). Further information regarding the historical and biological background of the *BRCA1/2* mutations can be found in Appendix A.

Strategies available to *BRCA1/2* mutation carriers to mitigate risk include intensive cancer screening, chemoprevention, and risk-reducing surgery (prophylactic mastectomy and/or salpingo-oophorectomy) (ACS, 2007; Babb et al., 2002; Eisinger et al., 1997; Foster et al., 2002). These strategies vary in the extent to which they are effective, physically and emotionally acceptable to women at high genetic risk of cancer, and practical at various life-course stages. For example, with regard to ovarian cancer, women who have not completed childbearing and wish to have biological children are not candidates for risk-reducing salpingo-oophorectomy, but relying on screening (generally regarded as ineffective) may leave them susceptible to uncertainty and continued vulnerability. Carriers must also consider the risk of passing the *BRCA* mutation to their future children. *BRCA1/2* are passed from parent to offspring in an autosomal dominant pattern; each offspring (of either sex) has a 50% chance of inheriting an abnormal copy of the gene from the parent (of either sex) who carries the mutation. Consequently, female *BRCA1/2* mutation carriers may face cancer-related distress including worry and anxiety (Watson et al., 1999), and may have both acute and chronic needs related to informational, tangible, and emotional support (Appleton, Fry, Rees,

Rush, & Cull, 2000; Babb et al., 2002; Kenen & Peters, 2001; Koehly et al., 2008; Peters, Hoskins, Prindiville, Kenen, & Greene, 2006).

These latter issues have been extensively explored, as have the bidirectional influences between marriage and the mutation positive experience in women of middle age (Manne, Audrain, Schwartz, Finch, & Lerman, 2004), and between marriage and the breast cancer experience of women in the same age cohort (Dorval et al., 2005; Osborne, Ostir, Du, Peek, & Goodwin, 2005). However, less attention has been paid to the personal and relationship implications of living with increased genetic risk of breast and ovarian cancer in early adulthood. Young female *BRCA1/2* carriers face unique challenges vis-à-vis informing partners of their risk status as new relationships develop, because they are less likely to be in stable relationships or to have completed childbearing when they undergo genetic testing (Oktay & Walter, 1991). An early literature review revealed only a single publication regarding the impact of being a *BRCA1/2* mutation carrier on couple relationships (Werner-Lin, 2007); since then, additional research has elucidated the complex manner in which the mutation disclosure process occurs in young couples (Hoskins, Roy, Peters, Loud, & Greene, 2008). We still do not fully understand how young *BRCA1/2*-positive women and their partners reconcile their mutation status within their couple relationship over time, or how partners effectively provide support to these women through the myriad stressors that accompany the mutation-positive experience.

Further, there has been little research regarding how young women living with increased breast/ovarian cancer risk reconcile their risk status with their plans and decisions regarding family formation. Werner-Lin (2008) observed a “great sense of urgency” (p. 428) in a group of 11 women aged 22-35 who were planning to start or

expand their families, largely related to their desire to have risk-reducing surgeries as early as possible. Young female *BRCA1/2*-mutation carriers may be faced with difficult dilemmas when, for example, their plans for having biological children are in conflict with physicians' recommendations regarding the timing of pregnancies and risk-reducing surgeries, or when their desire to breastfeed their biological children is at odds with a desire or perceived need to undergo risk-reducing mastectomy (unpublished data). To what extent do mutation-positive women consider making use of alternatives to "natural" biological family formation (*e.g.*, adoption, gamete donation, surrogacy), and what factors are involved in these decisions? What determines whether and how mutation-positive women alter their plans with regard to family formation as a result of their knowledge of their mutation status?

Finally, young mutation carriers face unique challenges with regard to decision-making about risk management and/or reduction. There are significant psychological and physical ramifications of some risk-reduction options, and the impact of these procedures on young women is often very different from the impact on women in middle age. For example, electing to undergo risk-reducing removal of the ovaries brings about full and immediate menopause; doing this at age 50 would seem relatively "on time" and normative, while commencing menopause in one's thirties might present challenges with regard to sexuality, body image, and grieving the early loss of fertility potential. Similarly, choosing to undergo risk-reducing prophylactic mastectomy challenges women and their current and/or future partners to grieve the loss of their natural breasts and the inherent capabilities therein; to cope with the often challenging process of surgery, recovery, and reconstruction; and to reacquaint themselves with their altered bodies. How

do women think and decide about risk-reducing surgery? How does the prospect of risk-reducing surgery impact young mutation carriers psychologically and emotionally? How do they and their partners cope with this stressor as a couple unit? What is the experience of young female mutation carriers who are not yet in couple relationships and/or choose not to be?

The current study is an attempt to elucidate this understudied area and contribute to our understanding of the lived experiences of women who are aware of their positive *BRCA1/2* mutation status during young adulthood. These qualitative findings can contribute to the knowledge of medical, mental health, and other healthcare providers who work with these women throughout the mutation-positive experience, from pre-testing through surgery and beyond, and assist them in better preparing women to successfully cope with the challenges and struggles they may face.

CHAPTER 2: REVIEW OF THE LITERATURE

Psychosocial Issues in Breast/Ovarian Cancer Predisposition

Cancer pre-vivorship as a newly legitimate health status. Advancements in medical technology have resulted in the recognition of “at-risk” as a legitimate health status warranting both medical and research attention. The availability of genetic testing and personal cancer risk assessment, combined with media reports lauding the benefits of genetic testing (H. T. Lynch & Lynch, 1999) has resulted in increased awareness among physicians and demand among consumers for testing of individuals and families with significant histories of cancer (Bottorff, Ratner, Johnson, & McCullum, 2000). As a result, there now exists a growing population of women who have never been diagnosed with cancer, but are aware of their own very high risk of developing breast, ovarian, and other cancers during their lifetime, due to an inherited mutation in *BRCA1/2*. These women are now part of an ever-expanding group of individuals who have already struggled with a sometimes difficult decision regarding testing, experienced a potentially stressful and complicated genetic testing and mutation test result disclosure, and may be coping with a variety of psychosocial and relational issues as a direct result of their membership in hereditary breast/ovarian cancer (HBOC) families and their individual decisions to learn their mutation statuses. For an expanded review of these complex issues, please refer to Appendix B.

In an effort to highlight the unique characteristics and needs of this cancer- unaffected, mutation-positive population, the Facing Our Risk of Cancer Empowered (FORCE), an online support network for *BRCA1/2*-positive women, has coined the term “cancer Pre-Vivor,” (FORCE, 2005). This term is meant to represent individuals who are

survivors of a *predisposition* to cancer. In the decade since genetic testing for *BRCA1/2* mutations became available and began to be used by members of high-risk families, a great deal of clinical and research energy has been focused on this group, but only a small subset of this research is psychosocial. Research has largely focused on the individual, has been largely quantitative in nature, and has targeted women in their forties and fifties. There is a noticeable absence of research focusing on the lived experiences of young pre-ivors, who face an altogether different set of challenges related to their high-risk status.

Breast and ovarian cancer risk-perception. Much of the psychosocial research that has been focused on women in HBOC families has examined cancer risk perception, and multiple studies have demonstrated that an individual's *perceived risk*, rather than objective scientific risk, plays a significant role in decision-making regarding genetic testing, screening and prevention options, and other *BRCA1/2*-related issues. Perceived risk also plays a large role in the psychological ramifications of those decisions (Absetz, Aro, & Sutton, 2003; French, Kurczynski, Weaver, & Pituch, 1992; Klein & Stefanek, 2007; Price, Butow, Lo, Wilson, & KConFab, 2007; Schwartz, Lerman, Miller, Daly, & Masny, 1995; Smerecnik, Mesters, Verweij, de Vries, & de Vries, 2009). High levels of cancer worry are associated with increased anxiety, depression, general emotional distress, family distress, and impaired role functioning for women in high-risk families (Audrain et al., 1997; Baider, Ever-Hadani, & Kaplan De-Nour, 1999; Lerman, Rimer, & Engstrom, 1991; Trask et al., 2001; Van Oostrom et al., 2003). Accurate risk perception is essential for women making potentially life-altering decisions regarding their breast and ovarian health (Hopwood, Howell, Lalloo, & Evans, 2003), and those with inaccurate risk perceptions may be more likely to have difficulty using empirical risk information to

guide decision-making regarding risk management and reduction (Kelly et al., 2005). Further, women with high levels of cancer risk perception may continue to experience negative emotional, psychological, and relationship implications of high risk perception even after completion of counseling, testing, and risk-reducing surgery (Lerman et al., 1998; Lodder et al., 2001; Van Oostrom et al., 2003).

Although several instruments, including the Claus Model, BRCAPRO, Gail Model, and Myriad II, have been developed to calculate scientific cancer risk based on family history and presence of a genetic mutation (Berry, Parmigiani, Sanchez, Schildkraut, & Winer, 1997; Claus, Risch, & Thompson, 1994; Pavelka, Li, & Karlan, 2007), and although genetic counselors and other health professionals interacting with high-risk women use these instruments and collective knowledge about genetic risk to communicate scientific risk to patients, research indicates that women are commonly unsure about their scientific risk and/or perceive their cancer risk to be different from what any of these instruments would calculate (Hopwood et al., 2003). Prior to genetic counseling and testing, women with family histories of cancer commonly describe their cancer risk as above-average to high (Hopwood et al., 2003); this is consistent with research suggesting that individuals with family histories of cancer tend to overestimate their own cancer risk (Andrykowski, Munn, & Studts, 1999; Croyle & Lerman, 1993; Struewing et al., 1995). It is striking that fully 76% of women with family histories of breast and ovarian cancer (most of whom are *not* mutation carriers) report that they perceived themselves to be at significantly higher risk of cancer than women in the general population, and 20% of them perceived cancer as inevitable (Hallowell, Statham, & Murton, 1998).

Further, many women subscribe to relatively simple and/or illogical explanations about risk, such as describing risk as “fifty-fifty,” or as connected to “bad luck,” (Bottorff et al., 2000). Conversion of objective risk figures into categorical terms such as high/low or likely/unlikely has been identified as a primary way that genetic counselees simplify their understanding of risk by interpreting it in binary form (Lippman-Hand & Fraser, 1979), allowing them to “mentally construct the worst-case scenario of the risk event occurring, and gauge the acceptability of this to themselves,” (Pilarski, 2009, p. 304).

The extent to which individuals’ risk estimations approach actual risk after genetic counseling varies with method of communication and several other factors. Moreover, the communication of risk information is complex and involves multiple distinct but related pieces of information, such as lifetime risk, levels of risk that change with age, separate risks for different cancer sites, etc. (Hopwood et al., 2003). Understanding of perceived risk is vitally important because “across all genetic-testing domains...participants’ decisions about testing are influenced less by their actual risk than by subjective risk and emotional factors,” (Lerman, Croyle, Tercyak, & Hamann, 2002, p. 792). The presence of cancer-specific worry and distress is associated with motivation for test use (Durfy, Bowen, McTiernan, Sporleder, & Burke, 1999; Lerman et al., 1997; Vernon et al., 1999), while general distress seems to reduce the likelihood of testing for cancer susceptibility genes (Lerman et al., 1999).

The common sense model “posits that individuals’ understanding of illness is based upon somatic symptoms and life experiences and thus may differ significantly from the biomedical view of illness,” (Kelly et al., 2005, p. 34). In a recent study of women at potentially increased risk of breast/ovarian cancer, participants indicated that their

perceptions of their own cancer risk were based on their understanding of the causes of cancer, their ability to control or cure cancer by having surgery or changing behaviors, and the timeline on which cancer would occur in their lifetime. The researchers argue that in order to better understand perceived risk and bring it in line with actual risk, genetic counselors and other health professionals working with high-risk women must understand these variables and how they fit together in the minds of women at risk (Kelly et al., 2005). A brief review of relevant variables includes age, family cancer history, and personal and family myths regarding family cancer history.

Age. Various studies have confirmed or failed to confirm the existence of an association between age and breast/ovarian cancer risk perception. Scientifically, cancer risk at both sites does increase with age (NCI, 2006, 2009) and most carriers are aware of this widely known fact. A study of women with significant family histories of breast cancer who had not yet undergone genetic testing found that older women were more likely to overestimate their risk (Cull et al., 1999). However, in families where breast and ovarian cancer are common experiences, age-related risk takes on a secondary meaning because individuals often associate the specific ages at which loved ones were diagnosed with increased risk for themselves. While simple numerical age is not always a reliable predictor of one's risk perception, there is an important relationship between age and the awareness, fear, and worry that comes along with it, especially when at-risk women are acutely aware of the age at which a loved one or loved ones were diagnosed with cancer. Werner-Lin (2007) reported that many participants in her qualitative study of young *BRCA* mutation carriers "believed that [their] path toward cancer would mirror those of family members who had traveled the road before them," (p. 342). Studies utilizing both

American and British samples have reported that feelings of fear, worry, and vulnerability increase as this age approaches for high-risk women, and that once that age passed the feelings would gradually subside (Appleton, Fry, Rees, Rush, & Cull, 2000; d'Agincourt-Canning, 2005; Werner-Lin, 2007).

Family illness narratives. At base, the way that members of a family collectively understand and discuss their cancer risk is based on their knowledge of what has occurred in other members of their families. Thus, family breast and ovarian cancer *history* describes the set of known information about biological relatives that applies to *BRCA*-related cancers. The extent to which an individual is familiar with his/her family cancer history is determined by levels of communication within the family group, social and demographic variables that determine whether or not s/he has had contact with family members on the relevant side of the family, and whether or not members of previous generations lived long enough that a natural history can be understood.

The narratives that families develop around their understanding of cancer risk may be more important than the hard facts surrounding examples of cancer in the family. As *BRCA*-related cancers move through families from generation to generation, a powerful shared history emerges that impacts each member of that family (Rolland, 2005). Negative images of cancer are commonly held by healthy family members as well as those who are personally affected by disease (Die-Trill, 2000). This shared history influences individuals' current behavior and responses to new *BRCA*-related stressors, which are associated with organizational shifts and coping strategies utilized within the family group (Rolland, 2005). Meanings attached to illness may also directly affect quality of life, individual and family system-level psychological responses, and

interpersonal relationships throughout the pre-cancer and cancer experiences (Die-Trill, 2000).

Research indicates that women take family experiences with cancer, including the number of affected individuals, positive and negative cancer experiences, and interpersonal relationships with affected family members, into account in evaluating their own risk and making decisions about management (Kenen, Arden-Jones, & Eeles, 2003). Family cancer history and experience, then, is a lens through which women in high-risk families examine and integrate the new genetic information they receive (Emery, Kumar, & Smith, 1998; Mellon, Berry-Bobovski, Gold, Levin, & Tainsky, 2006). Daughters of breast/ovarian cancer patients who experienced their mothers' cancer during adolescence, and especially daughters of women who had poor prognoses or who died as a result of cancer, are particularly vulnerable to high levels of cancer-related stress (Wellisch, Gritz, Schain, Wang, & Siam, 1992). It has been proposed that these unique challenges, including phobia about one's own breasts and inability to view breasts as sexual, are related to the experience of puberty and breast development during the period in which one's mother was experiencing and perhaps dying from breast cancer (Wellisch & Lindberg, 2000). In addition, women of any age who are currently dealing with cancer in another family member (*e.g.*, they have recently lost a relative to cancer, have a relative in treatment for cancer) are vulnerable, as they may be actively grieving and/or may identify more strongly with the cancer experience (Dudok de Wit et al., 1994).

In communicating risk information to individuals with significant family histories of cancer, the meaning attributed to risk is impacted by both social and familial meanings that act together to create causal schemas in the minds of individuals (Bottorff et al.,

2000). That is, regardless of the quality of risk information received in the healthcare setting, personal experiences relevant to the family history contribute to the formulation of a 'fixed' understanding of personal risk (Bottorff et al., 2000), creating a barrier in understanding and assimilating objective risk information (d'Agincourt-Canning, 2005). This is consistent with other research indicating that family cancer history is salient in constructing a personally meaningful explanation of one's own level of risk (Geller et al., 1997; McAllister, 2003). Feelings of vulnerability to cancer based on family history and family illness narratives shape attitudes toward behaviors that may mitigate risk, such as genetic testing and breast and ovarian screening (d'Agincourt-Canning, 2005). Risk perception is then related to the manner in which some mutation carriers create meaning regarding their positive mutation status and communicate with significant others, including partners and children (if applicable), in their lives. Breast and ovarian cancer risk perception is also brought to bear on women's decision-making with regard to family formation and cancer risk-reduction strategies over the life course (Hoskins et al., 2008).

In terms of a family's handling of the stress of this shared history, maladaptive management of cancer-related stressors and issues may lead to an increased sense of vulnerability to cancer, or heightened perceived cancer risk; conversely, healthy and functional family coping may lead to a sense that cancer is conquerable and lower perceived cancer risk (Rolland, 2005). These dynamics are fluid and likely to shift over time as the family cancer experience continues to develop.

Individual and family myths. Individuals and families often construct stories to help organize their illness experience, especially in the face of insufficient medical knowledge or understanding (Die-Trill, 2000; Rolland, 2005). It has been proposed that

such stories emerge as a result of a cultural emphasis on understanding cause and effect, of having a logical explanation for an event that threatens or modifies one's status; further, they may fulfill a need to recover basic assumptions about life that are threatened by the cancer experience (Kelley, 1971). These can be shared by multiple members of a family system or can vary from person to person, and can have significant bearing on individuals' perceptions of their risk both for being a mutation carrier and for developing cancer (Rolland, 2005), for decision-making with regard to prevention and treatment, for emotional and psychological responses, and for interpersonal relationships.

In some cases, causal explanations of illness or high-risk status can be quite irrational (Die-Trill, 2000). These beliefs may be based on ideas about punishment for prior misdeeds committed by the affected individual or by another family member, about justice versus injustice, about stress, life decisions (*e.g.*, having too many or no children), other physical factors, religion, heredity, homosexuality, lifestyle, or luck (Die-Trill, 2000). Individuals have reported beliefs about the outcome of genetic testing based on variables such as having achieved a certain age without being diagnosed with cancer (*e.g.*, "I'm 50 and I figure if I haven't gotten it by now, I probably won't") and about having been diagnosed with another serious disease or illness (*e.g.*, "I'm already facing diabetes, and I just don't think both diabetes and cancer would happen to me"). Family myths and beliefs that influence one's perceived risk of mutation-positive status and cancer include being raised to believe that one would eventually develop cancer and believing that one would inherit a cancer-predisposing mutation from a parent because they were physically similar to that parent (*e.g.*, "I look like my dad, and he has the mutation, so I figure I will have it, too") (H. T. Lynch et al., 1997).

These beliefs are not based in any medical fact or healthcare provider's advice. Still, they may provide comfort to an individual or group of individuals within a family. Some family beliefs and stories may be adaptive when they "[promote] the flexible use of multiple biologic and psychosocial healing strategies" (Rolland, 2005, p. 2592). However, these beliefs may also facilitate living in denial about the actual presence of risk. These beliefs can also be problematic when they contribute to individual and family pathology by focusing on blame, shame, or guilt and therefore inhibit coping and adaptation (Rolland, 2005). Further, attributions that are internal have been associated with maladaptive coping with illness-related stressors, whereas external attributions or a belief that the causes of illness are unknown are more often associated with facilitative coping (Bearison, Sadow, Granowetter, & Winkel, 1993).

Use of stress & coping theory in psychosocial research on HBOC. Much of the psychosocial research previously undertaken to examine the experiences of *BRCA* mutation carriers has used the Stress & Coping theory (Lazarus & Folkman, 1984). A brief review of this theory and its use with this population is appropriate because of its extensiveness in the current literature; however, as the stress and coping model is not the primary focus of the current project, it should not be considered a component of the project's theoretical background. Rather, I have used a more finely-tailored theoretical foundation to explicitly examine the experiences of *young* mutation carriers.

Briefly, stress and coping theory posits that one's ability to cope with a threat is related to the internal and external resources that are available to an individual (Dagan & Gil, 2004), including social support; and psychological and behavioral coping skills. Social support may be instrumental, in the form of "advice, assistance, or information,";

or it may be emotional, including “moral support, sympathy, or understanding,” (Carver, Scheier, & Kumari Weintraub, 1989, p. 269) from close others. Although they are identified as separate types of support provision, it should be noted that seeking/receiving informational and emotional support often co-occur in practice. Receiving either type of support may have both positive and negative consequences. Seeking instrumental support can be effective when it successfully meets the informational needs of the requestor, but may be ineffective (even though the requestor may not realize this) if the information given is inaccurate, biased, or otherwise not of high quality (Carver et al., 1989). When effective, emotional support can help to reassure an individual and to alleviate the burden of a stressor, but there is also a risk that an emotionally supportive dynamic can develop into an outlet for venting one’s feelings, which may be counter-adaptive when it continues long-term and impedes adjustment (Carver et al., 1989).

In many ways, female members of HBOC families are uniquely qualified to provide both informational and emotional support to young women in various stages of the *BRCA1/2*-positive experience because they can easily empathize and can share the knowledge they have gained through their own processes. However, several distinct characteristics of HBOC families may impinge upon members’ ability to effectively provide support for loved ones experiencing *BRCA1/2*-related stressors. For example, in watching their younger female family members cope with the grief, fear, uncertainty and ambiguity that may accompany a positive mutation test result, members of older generations within a family may experience intense feelings of guilt for having passed a mutation on to a loved one, making it difficult for them to serve as sources of support. Furthermore, the presence of multiple cases of cancer within the family may place

unrealistic support demands on the family in general. At a time when there are perhaps several active cancer cases, unaffected mutation carriers may find it difficult to ask for or receive the attention they need, because the family's support energy is being focused elsewhere and/or because the individuals that would otherwise be relied on for support are themselves coping with cancer (Dagan & Gil, 2004).

Findings from studies investigating the association between partner relationships and experiences with illness are relevant to the current study because, although *BRCA1/2*-positive women are not currently ill, they are still faced with a number of unique stressors regarding their physical health. Like chronically ill patients, *BRCA1/2*-positive women may experience frustration about the unpredictable nature of their situation, as well as fears about the future (Revenson & Majerovitz, 1990). In studies related to various health concerns (*e.g.*, lupus, cancer, diabetes, heart conditions) (Fekete, Parris Stephens, Mickelson, & Druley, 2007; Trief et al., 2003), high-quality partner support has been identified as having positive effects on individuals' physical, psychological, and overall well-being during periods of increased stress caused by illness (Burman & Margolin, 1992; Fekete et al., 2007; Trief et al., 2003). Specifically, having a partner has also been consistently associated with lower levels of distress among women at high genetic risk of breast and ovarian cancer and is likely associated with the increased availability of social and emotional support for couple members (Audrain et al., 1997; Baider, Ever-Hadani, & Kaplan De-Nour, 1999). The couples with the highest levels of well-being, marital satisfaction, and illness adjustment in the face of chronic health concerns are those who can effectively reciprocate each other's support efforts (Manne et al., 2004).

With regard to the experience of living with increased genetic risk of cancer in the context of a couple relationship, very little specific research exists. However, there is evidence that couples who experience high levels of distress prior to a cancer diagnosis are more likely to experience problems during the active phase of the disease (Northouse, Templin, Mood, & Oberst, 1998), and relationship distress prior to genetic counseling predicts higher levels of distress both during and after the counseling process (Dudok de Wit, Duivenvoorden, Passchier, Niermeijer, & Tibben, 1998).

Coping with *BRCA*-related stressors. Various studies have highlighted a number of coping strategies utilized by women at high genetic risk of breast/ovarian cancer. A group of Scottish women who participated in a telephone focus group reported that they purposely shifted their focus to the present, avoided worrying about breast cancer cues, and thought positively about their situation by adopting an optimistic attitude about the future. They also reported a purposeful reassessment of their lives and priorities, taking on an attitude that they should maximize the potential for fullness in life and make their health a greater priority. They discussed the adoption of a “one-day-at-a-time” approach, which prevented them from becoming overwhelmed by thinking about the negative experiences that might occur over time; this approach was also described as a method of combating persistent worry, varying from mild to intrusive. They engaged in excessive reassurance seeking from doctors, family, etc. in times of acute anxiety, in an apparent attempt to mitigate that emotion. They adopted significant lifestyle changes such as healthier diets, increasing frequency or intensity of exercise, stopping smoking, use of natural or herbal remedies, and various forms of stress management. They strived to pass along these healthier habits to other members of their family, particularly members of the

younger generation. These behaviors were seen as giving women some sense of control over their increased risk. With regard to screening, some women in this study became increasingly vigilant about performing breast self-exams in an effort to maximize their awareness of their breast tissue and health; however, others moved in the opposite direction, purposely avoiding breast self-exams to avoid the discomfort and anxiety associated with the behavior (Appleton et al., 2000).

Denial, characterized by a refusal to acknowledge or believe that the stressor exists or acting as though it does not (Carver et al., 1989), is another psychological coping strategy, and may be effective in that it can facilitate coping if it minimizes distress (Appleton et al., 2000) and therefore allow the individual to shift his/her internal energy toward information processing and decision-making (Decruyenaere, Evers-Kiebooms, Welkenhuysen, Denayer, & Claes, 2000). However, it may also exacerbate problems in the long-term if they are ignored rather than effectively dealt with and/or resolved (Carver et al., 1989), or if denial behavior prohibits the appropriate acquisition of information or prudent screening practices (Decruyenaere et al., 2000). In the context of a *BRCA1/2*-positive women, denial may be an effective coping mechanism during one's late teens and early twenties if it allows her to live her life as "normal" during this period of relatively low risk; however, if denial behavior continues into one's thirties and forties and results in failure to participate in appropriate screening and prevention behaviors, it may certainly be considered maladaptive and risky. To a lesser extent, minimization and disengagement may result in outcomes similar to those of denial. Some individuals may attempt to cope with distress by convincing themselves that a problem is not as bad as it really is (Decruyenaere et al., 2000). It is common for the need for and

chosen method of coping to vary based one's level of engagement, in this case, the degree of cognitive and emotional involvement with one's increased risk of developing cancer (McAllister, 2003). In fact, *disengagement* may be a result of intense emotional pain experienced as a result of previous engagement (McAllister, 2003), and can be interpreted as a method of avoidance.

At the other end of the spectrum from denial and disengagement is acceptance, which may occur more frequently in response to stressors that cannot realistically be eliminated (Carver et al., 1989), such as carrying a *BRCA1/2* mutation. Acceptance may also be, in some cases, a step along the route toward active behavioral coping (Carver et al., 1989). Finally, for *BRCA*-positive women, turning to religion, may be a "vehicle for positive reinterpretation or growth, or...a tactic of active coping with a stressor," (Carver et al., 1989, p. 270). One might engage in prayer, ask others to pray about a certain topic, or seek comfort in passages from a religious document, among other religiously-based coping strategies. Given the variability in individual religious beliefs and importance of spirituality, it seems obvious that different people will utilize this coping strategy to varying degrees.

Some women describe seeking social support for instrumental reasons, such as for information, advice, or assistance in dealing with the specific demands that come along with a stressor (Carver et al., 1989). Women in HBOC families may ask health professionals for breast/ovarian cancer information, ask friends or family members to accompany them to doctor or mammography appointments, or ask family members for their opinion about *BRCA1/2*-related decisions. Seeking and possessing knowledge and

information about one's risk status and the ramifications thereof was an oft-discussed topic in Appleton's study. Specifically,

The women commonly expressed a need for specific *types of* information, particularly up-to-date, professionally approved, detailed information on a variety of topics including the clinical services available (particularly genetic testing), scientific research (concerning breast cancer treatments, current trials and genetics), preventive measures (such as diet), hormone replacement therapy, the oral contraceptive pill and stress management, (Appleton et al., 2000, p. 517).

Some of the women in that study reported that having information about their mutation helped them cope because they felt more able to make informed decisions about risk reduction and prevention, thereby decreasing anxiety; others reported that having more information actually added to their feelings of anxiety and distress; others sought a balance between having an appropriate level of knowledge to be adequately informed, and not getting so much so as to become overwhelmed (Appleton et al., 2000).

Life-Course Challenges Faced by Young *BRCA1/2*-Positive Women

Although the information provided by these studies is both interesting and informative regarding the experience of *BRCA* mutation carriers as a whole, it was collected from samples with a broad range of ages and therefore does not provide a focused perspective on the experiences of *young* carriers. Women who are aware of their positive mutation status during young adulthood face normative life course challenges that set them apart from older mutation carriers. Specifically, their positive mutation status may shape the manner in which they navigate couple relationships and family formation, and each of these may be related to women's decision-making about use of risk-management and/or risk-reduction options.

Establishing and maintaining couple relationships. Although psychosocial research in cancer is starting to explore issues related to couple relationships during and

after cancer, very few studies have examined the experiences of young adult women living with increased hereditary risk of cancer. Women's experiences in dating, courtship, and early marriage may be profoundly shaped by the presence of a mutation in *BRCA1/2*, which may force them to consider issues related to family formation and their own mortality much earlier than their mutation-negative peers, putting unique stressors on developing couple relationships. In the limited research that has been undertaken on this topic, there exists some evidence that the mutation-positive experience is a relevant consideration in terms of context for development of early couple relationships. Women seeking genetic counseling and/or medical advice in research settings in which psychosocial issues were also attended to have reported feeling reluctant to get involved in relationships or attached to potential partners for fear that they will ultimately suffer and inflict a loss on those partners (Wellisch & Lindberg, 2000). This phenomenon, termed *life truncation syndrome*, is characterized by a seeming inability to allow life to progress around common commitments of adulthood, including leaving the parents' home, formation of a permanent couple relationship, and forming one's own family (Wellisch & Lindberg, 2000) and has been observed with varying degrees of severity in a number of young, high-risk women.

Prior to developing this dissertation proposal, a qualitative pilot study was undertaken to explore the feasibility of conducting a large-scale qualitative dissertation research project focused on the experiences of young *BRCA1/2*-positive women (Hoskins et al., 2008). The results of that study provided significant insight regarding how young pre-marital couples and those who are newly married communicate regarding the health threat that accompanies a positive *BRCA1/2*-mutation status. Findings indicated that

young women in pre-marital and non-marital relationships who were aware of having a *BRCA1/2* mutation experienced significant anxiety regarding communicating their risk status to their romantic partners. Some women imagined negative relationship outcomes resulting from such communication, and these negative experiences were a reality for a few. However, the majority of respondents in the pilot study reported that disclosure of their high-risk status to their partners was a positive experience that resulted in increased emotional intimacy. Further research is needed to increase our understanding of how the presence of a *BRCA1/2* mutation continues to impact these couples over time as they move toward relationship permanence and confront other issues at that stage of the life course. In addition, knowledge about how women handle communication about their mutations with successive partners when early pre-marital relationships come to an end should be expanded through further research.

Buried deep within the larger issue of genetic discrimination is a very subtle form of discrimination whereby a *BRCA1/2* carrier “may find herself less welcome as a possible wife,” (Surbone, 2001, p. 155). This concept was confirmed by our pilot study, which demonstrated that although sharing information about *BRCA1/2* mutations has the potential to bond members of young couples more closely together, it also caused tension and anxiety for young mutation-positive women and for their partners. Young women reported that they worried about being viewed differently by a partner who finds out about their mutation, and that some spent significant time and energy thinking about when and how to broach this subject in the context of a new relationship. Women also reported that they held specific ideals regarding how their partners *should* react, and responses outside of this ideal range were experienced as negative. That is, women

expected their partners to respond to the news of the mutation with enough anxiety to demonstrate care and concern, but not so much as to increase the women's own anxiety (Hoskins et al., 2008).

Couple dynamics with regard to communication and integration of the mutation-positive experience into the couple relationship appear to be related to women's cancer risk perception and to attachment. The familial cancer experiences that underlie high levels of perceived breast and ovarian cancer risk of *BRCA1/2*-positive women may contribute to attachment difficulties that make disclosure about risk to partners difficult. In my experience working with women from HBOC families in the CGB clinic, the concepts comprising attachment theory seemed particularly salient with regard to how *BRCA1/2*-positive women integrate family cancer experiences with their current methods of coping and communicating about cancer risk.

Family formation. That reproductive choices may be affected by the presence of a *BRCA1/2* mutation is widely accepted among those who study individuals and families at high genetic risk of cancer (Coughlin, Khoury, & Steinberg, 1999). Just as discrimination at the interpersonal level may lead to a woman being perceived as a less desirable partner, so too might this type of discrimination limit her options for childbearing (Surbone, 2001). Further, one's desire to conform to societal, family, and relationship expectations regarding how and when family formation will occur may lead to stress in light of the challenges posed by positive *BRCA1/2* status. Individuals may experience a mismatch in the roles of "mutation carrier" and "parent" when the demands of one role run counter to the demands of the other. For example, a woman may have to make difficult decisions regarding risk-reduction options at a time when she feels

committed to retaining fertility. The use of oral contraceptives (OCPs) is associated with a ~50% reduction in ovarian cancer risk (McLaughlin et al., 2007) (this is discussed in more detail in the *chemoprevention* section). If chemo-prevention via the use of OCPs was her chosen method of ovarian cancer risk-reduction during the pre-childbearing years, she will experience significant new stresses when, in order to achieve a pregnancy, she terminates use of OCPs and thereby increases her vulnerability to ovarian cancer. Ceasing chemo-prevention and relying on screening for ovarian cancer (generally regarded as ineffective) during one's active childbearing years may increase cancer anxiety, in turn affecting a women's psychological and emotional well-being, as well as her relationships with her partner, children, family of origin, healthcare team, and others.

Individuals at high genetic risk of cancer report multiple bases for concern about childbearing. Concerns about the mother's health and mortality during childbearing years and during children's early lives are legitimate given the potentially very early onset of cancer among mutation carriers (Narod et al., 2002; K. R. Smith, Ellington, Chan, Croyle, & Botkin, 2004). There is also a concern regarding passing a *BRCA* mutation on to children conceived naturally (K. R. Smith et al., 2004). Finally, but perhaps of the greatest immediate importance, there are some concerns that pregnancy itself could serve as a catalyst for the development of cancer due to increased exposure to estradiol during pregnancy, which has been associated with tumor growth in *BRCA1* carriers specifically (Chetkowski, Cassidy, Ma, Kagan, & Szell, 2008).

A 2004 study investigating differences in fertility intentions among those who either did not know their mutation status, had tested positive, or had tested negative found that both women who tested positive and women who did not know their mutation status

were less likely than those who tested negative for a *BRCA* mutation to want additional biological children (K. R. Smith et al., 2004). This study “suggested that carriers are 5.5 times more likely...to have altered their family planning than noncarriers because of their test results,” (K. R. Smith et al., 2004, p. 738).

In the presence of a known genetic mutation, it is certainly possible that some individuals and couples will choose not to have biological children as a way to guarantee that they will not pass on a harmful genetic mutation, or to avoid the potential cancer risks associated with pregnancy; some of these may remain childless, while others may choose to adopt, use donated gametes (*i.e.*, sperm or egg) in place of those of the mutation-positive parent, or employ surrogacy. Others choose to proceed with natural childbearing just as they would if the mutation was not an issue, and face any potential consequences as they arise. Others may choose to intervene in some way, in an attempt to “load the genetic dice” (Harmon, 2006) in their favor, using reproductive technology to increase the odds that they will produce a mutation-free biological child. The range of family formation choices available to couples in which one partner is a *BRCA1/2* carrier includes adoption, third party reproduction (gamete donation and/or surrogacy), pre-implantation genetic diagnosis (PGD) and pre-natal diagnosis (PND). Each of these strategies brings with it a unique set of considerations, and the choices faced by young couples are challenging and complex. However, because the current study focuses on how on the relationship between family formation and risk-management decision-making, and not on the specific processes women and couples use to achieve their family formation goals, a detailed discussion of family formation options is beyond the scope of this literature review. A brief overview of these options can be found in Appendix C.

Breast and ovarian cancer risk management and reduction. There are three primary categories of cancer prevention strategies available to women at increased hereditary risk of breast and ovarian cancer: clinical screening/surveillance, chemoprevention, and risk-reducing surgery (Dowdy, Stefanek, & Hartmann, 2004). Each individual mutation carrier is faced with difficult choices in selecting which strategy or strategies to utilize at various points in her life. These decisions are made in the context of what is available and recommended to any given woman in the medical settings in which she interacts, and these settings vary widely from person to person. For example, some women may have annual visits with gynecologic oncologists or physicians who specialize in hereditary cancer risk, while others may receive their risk-related care from general practitioners and/or obstetrician/gynecologists (Ob/Gyns) who, given the relatively low frequency of *BRCA1/2* mutations in the general populations, may encounter as few as 1 *BRCA1/2* mutation-positive patient every 20 years in their careers (de Bock et al., 2001). Clearly, these professionals are differently able to responsibly address the concerns and meet the needs of *BRCA1/2* mutation carriers. This healthcare mismatch is worrisome because, for example, prior research indicates that general practitioners are more likely to recommend or endorse surveillance as a strategy to deal with increased risk than are specialists trained specifically to deal with women from HBOC families, who are more likely to instead recommend chemoprevention or risk-reducing surgery (de Bock et al., 2001; Eisen, Rebbeck, Wood, & Weber, 2000).

In making decisions regarding cancer prevention, women must take into consideration family cancer history, mutation status, perceived and actual cancer risk, the efficacy and risks associated with each prevention method, financial and insurance issues,

their own preferences and those of loved ones. This process is even more complicated in light of the inability of any strategy to provide complete protection against cancer at this time (Dowdy et al., 2004), leaving mutation carriers with residual vulnerability to cancer no matter what they choose. There is also a risk of false reassurance when women select a screening method and erroneously believe that they are “protected” from cancer, when in fact they may still be quite vulnerable (Bottorff et al., 2000). Largely for this reason, healthcare professionals have begun to refer to this set of options as “risk-reduction” options rather than “prevention” options.

Surveillance. Risk management strategies that fall under the umbrella term *surveillance* (also referred to as “screening”) include monthly breast self-examination (BSE) beginning at age 18, biannual clinical breast examination (CBE), annual mammography and breast MRI (Brekelmans et al., 2001; NCCN, 2007; Vasen et al., 2005), annual gynecological examinations, semi-annual serum CA-125 tests, and transvaginal ultrasound (TVU) (NCCN, 2007; Pavelka et al., 2007; Scheuer et al., 2002; Vasen et al., 2005). Generally, it is recommended that female *BRCA1/2* mutation carriers commence surveillance at the age of 25 or 5-10 years earlier than the youngest cancer diagnosis in their family, whichever is earlier. The performance characteristics related to each of these screening procedures range from terrible (ultrasound, CA-125 for ovarian cancer) to good (MRI for breast cancer), and all may be accompanied by anxiety prior to each test, substantial risk of false-positive test results, and anxiety until definitive results indicating an absence of concern are in hand. It has been estimated that 40-80% of *BRCA1/2*-positive women choose to utilize screening alone to manage their risk, at least for some time after they complete the genetic testing process (Lerman et al., 1996).

Although it is frequently discussed as a risk-reduction strategy by healthcare providers and mutation carriers alike, screening should instead be considered a way of *managing* risk, rather than reducing it, because it does not actually alter the likelihood that cancer will develop. It is simply a method of *detecting* cancer if it is present. Choosing screening rather than more radical risk-reducing surgeries has advantages in that the breast or ovarian tissue is preserved, as is fertility and the ability to breastfeed; many women choose this option in the hope that new diagnostic options and treatments will emerge that make surgery unnecessary (Dowdy et al., 2004).

Some researchers and practitioners have expressed worry that genetic testing for cancer risk may result in increased distress that may reduce adherence to screening (Lerman et al., 2002), and this hypothesis has been borne out in some research of untested high-risk women (Kash, Holland, Halper, & Miller, 1992). However, studies specifically examining women who have undergone testing do not support this hypothesis. One study failed to find a significant relationship between posttest scores on the State-Trait Anxiety Inventory (STAI) (Spielberger, Gorsuch, & Lushene, 1970) and Impact of Events Scale (IES) (Horowitz, Wilner, & Alvarez, 1979) and one-year posttest mammography adherence in multivariate analysis (Botkin et al., 1996); another identified a positive correlation between BSE frequency and IES scores among carriers, but not among non-carriers (Lerman et al., 2000).

Chemoprevention. The term *chemoprevention* in *BRCA*-related literature is used to describe a group of pharmacologic treatments, many of which alter the balance of the female hormones estrogen and progesterone. Both of these hormones have been implicated in breast and ovarian carcinogenesis (Eeles & Powles, 2000). The use of such

chemopreventive medications has been associated with reductions in both sporadic breast and ovarian cancer incidence (McLaughlin et al., 2007). Tamoxifen, for example, has been shown to reduce the risk of developing estrogen-receptor (ER) positive breast cancer among women at modestly increased risk by up to 69% (Fisher et al., 1998; Fisher, Powles, & Pritchard, 2000). Although data regarding the efficacy of tamoxifen in *BRCA* mutation carriers are sparse and limited by small sample size and therefore not definitive, the evidence is generally consistent with a protective effect (Dowdy et al., 2004; Eisen & Weber, 2001; Johannsson et al., 1997; Karp et al., 1997; King et al., 2001; Vogel, 2001), though more recent research has questioned this (Temin, 2009). Raloxifene is a second member of the selective estrogen receptor modulator (“SERM”) drug family, which also has demonstrated a protective effect for sporadic, ER-positive breast cancer; it has not been evaluated in mutation carriers (Chlebowski, Collyar, Somerfield, & Pfister, 1999; Temin, 2009). Ongoing trials continue to investigate the efficacy of various chemopreventive medications in mitigating breast cancer risk among for high-risk women (Palma, Ristori, Ricevuto, Giannini, & Gulino, 2006).

Potential adverse effects of chemopreventive medications comprise a major barrier to the acceptance of these agents by consumers and healthcare providers alike. These include increased incidence of endometrial cancer, deep vein thrombosis, pulmonary embolism/superficial phlebitis, stroke, hot flashes, and vaginal discharge (Fisher et al., 1998). It has become clear that medications like those just described, which are readily accepted by women as part of breast cancer treatment regimens, are viewed with much greater skepticism and wariness by healthy (albeit at-risk) women who do not

have cancer. Thus, tamoxifen – a scientific triumph in the field of cancer prevention – has proved unacceptable to most high risk women.

The use of oral contraceptive pills (OCPs) has long been associated with reduced frequency of ovarian cancer among women in the general population (Francheschi et al., 1991; Whittemore, Harris, Intryre, & Group, 1992). A consensus is now emerging that a protective effect of similar magnitude (~50% reduction) is also seen in *BRCA* mutation carriers (Bosetti et al., 2002; Francheschi et al., 1991; Lakhani et al., 2002; McLaughlin et al., 2007; Narod et al., 1998; Whittemore et al., 1992). Current chemoprevention research for ovarian cancer is focused primarily on progestational agents (Rodriguez, 2003).

The use of oral contraceptives as a method of reducing hereditary ovarian cancer risk in young women is an appealing option for several reasons. First, the treatment is generally well-tolerated and does not present significant side-effects for most women; many women of this age may be likely to use OCPs anyway as a preferred method of birth control, and so the risk-reduction may be seen as an added benefit of an already positive personal choice (Narod et al., 1998). Second, fertility can be preserved in cases where women wish to have biological children in the future but are not ready to do so yet. Third, using oral contraceptives to reduce cancer risk can be a way of “buying time” before more drastic measures, such as risk-reducing surgery, are deemed necessary, allowing women to maintain breast and ovarian tissue and function throughout early adulthood. However, it is important to note that OCP use has also been associated with *increased* risk of breast cancer among *BRCA1* mutation carriers, especially when used for five years or more and when used prior to age 30 (Kaduri et al., 1999; Narod et al., 2002; Narod et al., 1998). This relative “tradeoff” of risks is an important component of

decision-making about OCP use for *BRCA1* carriers (Grenader, Peretz, Lifchitz, & Schavit, 2005), and perceived risk of breast versus ovarian cancer (likely based on family history) may be an important factor in women's decision-making regarding this risk-reduction strategy. For *BRCA1* carriers, use of OCPs may be an attractive option for ovarian cancer risk management after RRBM. No association between OCP use and breast cancer risk is noted for *BRCA2* carriers.

Risk-reducing surgery. Of the three cancer prevention strategies, risk-reducing (or “prophylactic”) surgery offers the highest level of protection against cancer (Karlan, 2004). Risk-reducing bilateral mastectomy (RRBM) is the complete (or near-complete) removal of breast tissue in the absence of a breast cancer diagnosis. Risk-reducing salpingo-oophorectomy (RRSO) is the complete removal of ovarian and fallopian tube tissue in the absence of an ovarian cancer diagnosis. Studies of women who have chosen these prevention strategies suggest that RRBM reduces the risk of breast cancer by more than 90% in high-risk women (Esplen et al., 2004; Hartmann et al., 1999; Meijers-Heijboer et al., 2001), while RRSO reduces the risk of ovarian cancer by 85-95%; in both cases, residual risk of breast or ovarian cancer is believed to be lower than population risk. RRSO also reduces the risk of breast cancer by 50% in pre-menopausal women (Kauff et al., 2002; Kramer et al., 2005; Rebbeck et al., 1999; Rebbeck et al., 2002). Research indicates that women who elect to undergo risk-reducing surgery “benefit both medically and psychologically...by the reduction of both their objective risk and their perceived risk of developing cancer and that this benefit is the greatest among women who undergo both ovarian and breast surgeries,” (Madalinska et al., 2005, p. 6897). These procedures are associated with high direct costs, approximately \$3,100 to \$4,500

for RRBM and \$6,000 to \$13,900 for RRSO (Grann, Panageas, Whang, Antman, & Neugut, 1998), making them inaccessible to women without insurance coverage; however, reports indicate that insurance companies have routinely paid for risk-reducing surgery for women who are confirmed *BRCA1/2* mutation carriers (Eisen et al., 2000), a practice that is likely to become more common with time as more and more women opt for these procedures.

Risk-reducing bilateral mastectomy. Originally considered controversial given its performance in the absence of a cancer diagnosis and the seemingly drastic nature of the procedure (Eisen & Weber, 1999; Fentiman, 1998; Holzgreve, Beller, Niedner, & Niehaus, 1989; Klijn, Janin, Corez-Funes, & Colomer, 1997), RRBM has become an increasingly utilized method of cancer risk reduction among *BRCA* mutation carriers (Frost et al., 2000; Hartmann et al., 1999; Hatcher, Fallowfield, & A'Hern, 2001; Meijers-Heijboer et al., 2001). Research indicates that up to 51% of *BRCA1/2*-positive women consider risk-reducing mastectomy (Lerman et al., 1996; H. T. Lynch et al., 1997; Meijers-Heijboer et al., 2003; Meijers-Heijboer et al., 2000; Meiser, Butow et al., 2000; Meiser et al., 2003; van Dijk et al., 2003). Among those who consider the procedure, only about half who are eligible will actually undergo the procedure, with women who have children significantly more likely to undergo RRBM than those who do not (Meijers-Heijboer et al., 2000).

This is consistent with other research noting that many women who choose to undergo risk-reducing surgery are motivated to do so by a sense of obligation to their families (Hallowell et al., 1998), which may be a product of women's acknowledgement that choosing surgery is the most effective method of preventing cancer. Women who are

pre-menopausal but likely finished with childbearing (aged 40-54) are more likely to elect RRBM than women who are under 40 or over 55; however, perhaps surprisingly, women between the ages of 30 and 35 (a subset of the under-40 group) were more likely than any of the other age groups to elect this procedure. This finding is supported by our pilot data, which demonstrates that some mutation-positive women in their 20s and 30s move quickly toward RRBM in order to bring about peace of mind prior to entering the parenting phase of their lives (unpublished data). One group of researchers notes that “combining both predictors of age and parenthood 70% (28 of 40) of women aged below 50 years with children opted for [risk-reducing] mastectomy,” (Meijers-Heijboer et al., 2000, p. 2018). However, none of 41 confirmed carriers in a 2000 study had undergone RRBM within one year following receipt of results, and only 17% were considering the procedure (Botkin et al., 1996). Similarly, only 3% of unaffected carriers in a separate study had undergone RRBM within one year of result receipt (Lerman et al., 2000). This suggests that women may need some time to adjust to the idea of utilizing surgical risk-reduction, and explains the previously mentioned phenomenon whereby the vast majority of *BRCA* mutation carriers utilize surveillance for at least some period of time after learning about their mutation statuses.

The decision to undergo RRBM is predicted by motherhood (Meijers-Heijboer et al., 2001), higher levels of breast cancer worry (Stefanek, Hartmann, & Nelson, 2001), the belief that surgery would significantly reduce the risk of disease, higher perceived risk, having had previous breast biopsies and/or genetic testing (Hatcher, Fallowfield, & A'Hern, 2001), older age, and positive family cancer history (Lerman et al., 2000). It is also of note that rates of RRBM acceptance are higher in countries with socialized

medicine systems (*e.g.*, UK, Netherlands) than in the US (Dowdy et al., 2004), presumably because the cost barrier is eliminated for women in those countries and may be very present (or even insurmountable) for those in the US.

Satisfaction with RRBM and reconstruction is generally high among women at high risk of cancer (Borgen et al., 1998; Frost et al., 2000; Klijn et al., 1997; Meijers-Heijboer et al., 2000; M. E. Stefanek, 1995), but varies with reconstructive decisions/outcomes and by interval between surgery and measurement of satisfaction (Bebbington & Fallowfield, 1999). A 1998 study found that 5% of women had significant regrets after completion of RRBM (Borgen et al., 1998). These may be associated with negative outcomes such as pain during recovery, necrosis, hematoma/seroma, encapsulation, wrinkling, pain, asymmetry, and diminished breast sensation, which occur in 10 to 59% of patients (Eeles, Cole, Taylor, Lunt, & Baum, 1996; Eisen et al., 2000; Gabriel et al., 1997; Hopwood et al., 2000). Rates of complication may be as high as 30% over five years for women who chose implant reconstruction (Gabriel et al., 1997).

In general, women who undergo RRBM generally experience positive psychological outcomes, as was reported in 94% of a sample of 609 women with family histories of breast cancer (Frost et al., 2000). Breast cancer anxiety is generally significantly diminished, and a majority of women report either positive changes or no changes with regard to emotional stability, stress, self-esteem, sexual relationships, femininity, and appearance. These responses were not associated with age at surgery, length of follow-up, level of actual family cancer risk, or type of mastectomy performed (*i.e.*, total or subcutaneous) (Frost et al., 2000). Overall satisfaction with RRBM is

associated with satisfaction with post-surgical physical appearance, lower levels of stress, and the decision *not* to have reconstruction after RRBM.

Possible negative emotional/psychological outcomes of RRBM include loss of femininity and sexuality (Eeles et al., 1996; Frost et al., 2000; Hopwood et al., 2000; Meijers-Heijboer et al., 2000); difficulties with body image including perceptions of decreased sexual and physical attractiveness, and decreased satisfaction with body (especially among those who elect not to have reconstruction) (Carver et al., 1998; Frost et al., 2000; Hopwood et al., 2000; Meijers-Heijboer et al., 2000); loss of self-esteem (Frost et al., 2000; Hopwood et al., 2000); unexpected difficulty accepting breast loss (Hopwood et al., 2000); difficulty adjusting to the appearance, feeling and/or relationship impact of reconstructed breasts (Carver et al., 1998; Hopwood et al., 2000); psychosomatic symptoms related to implants (Hopwood et al., 2000); avoidant behavior with respect to reconstructed breasts (i.e., avoiding intimacy or exposure) (Hopwood et al., 2000); negative changes in sexual relationships (Frost et al., 2000; Hopwood et al., 2000); depression as a result of surgical complications or relationship issues following surgery (Hopwood et al., 2000); adverse effects in one's level of emotional stability; and increased stress (Frost et al., 2000). However, most research suggests that over time, the procedure has no permanent detrimental effects on body image or sexuality (Borgen et al., 1998; Frost et al., 2000; Stefanek, Helzlsouer, Wilcox, & Houn, 1995). That said, there can be no doubt that the process of contemplating the possibility of RRBM, and then living with the consequences of that decision (regardless of what those might be), can be the source of an enormous amount of stress, anxiety, and interpersonal conflict.

Although a great deal of research has examined individual outcomes of risk-reducing mastectomy and the process of decision-making, as outlined previously, there is a relative absence of research that examines these issues from the couple perspective, or wholly considers RRBM decisions/outcomes in the context of the couple relationship. One small study investigated partners' perceptions of the outcomes of surgery, finding high levels of overall agreement between patients' perceptions and partners' perceptions; there were also some cases in which partners highlighted issues in adjustment to surgical outcome that were not identified by the patients themselves (Brereton et al., 1999). Other research has posited that women are reluctant to confront potential sexual problems pre-operatively, and that this may lead to more problems in the post-operative phase (Dudok de Wit, Tibben, Frets et al., 1997), such as reluctance to let a partner touch or look at reconstructed breasts (Hopwood et al., 2000), which may present difficulties within other aspects of the couple relationship. Most research on RRBM in *BRCA1/2* mutation carriers is conducted with women who are approaching surgery in the months leading up to the procedure; little existing research examines the thoughts and feelings of young women regarding risk-reducing surgery over time as they move through the mutation-positive experience, and this is especially true for young women. This absence leaves a significant gap in the research. We do not know, for example, about the experiences of women who are aware of their increased hereditary risk of cancer and/or undergo RRBM prior to the formation of a permanent couple relationship. What is their experience in explaining this decision and its implications to a new partner? When these women experience negative consequences with regard to their own sexuality and feelings of attractiveness as a result of an RRBM, how is this related to their desire and ability to form and/or maintain couple

relationships? To what extent might some women delay RRBM because they are not yet in a couple relationship, perhaps feeling that they will be less able to attract a partner without their natural breasts? Pilot data indicate that these are important considerations (unpublished data), and they have not yet been captured in the existing research

Risk-reducing salpingo-oophorectomy. Risk-reducing removal of the ovaries and fallopian tubes is a preferred ovarian cancer risk management strategy for many female *BRCA1/2* carriers for several reasons. First, as previously discussed, there is no effective screening strategy for the early detection of ovarian cancer. Second, the majority of patients present with advanced disease while only a minority are cured, giving many carriers the (largely correct) impression that an ovarian cancer diagnosis is likely to mean death. Finally, RRSO leaves no visible change in bodily appearance (Palma et al., 2006). A growing number of health care providers strongly recommend the procedure for this population (Eisen & Weber, 2001). Sources of stress related to this decision include:

- Symptoms which prove (in retrospect) to have been the harbingers of cancer development are vague, non-specific and common (i.e., easily confused with menstrual cramping or bloating);
- Screening with CA-125 and transvaginal ultrasound is fraught with apprehension, worry and a high rate of false positive test results;
- The need to make a final and irreversible decision about bringing child-bearing potential to an end, perhaps at an earlier point in time that it might otherwise be contemplated;
- Concerns that the loss of one's ovaries will bring a loss of femininity, particularly for premenopausal women; and

- Fears regarding the potential long-term complications of premature menopause, particularly with regard to cardiovascular and bone health.

In general, patient acceptance of RRSO is high (Pavelka et al., 2007). Studies taking place in different countries have yielded varying results regarding the attractiveness of RRSO to high-risk women as well as their reasons for choosing this procedure. Various studies have reported that 33-76% of women at high genetic risk of breast/ovarian cancer consider RRSO (Hallowell et al., 1998; Lerman et al., 1996; Lynch et al., 1997), and a subset of those will ultimately elect to have the procedure.

Researchers have demonstrated that RRSO is effective in reducing anxiety about and perceived risk of ovarian cancer for women at high genetic risk (Elit, Espfen, Butler, & Narod, 2001); this level of anxiety reduction is generally thought to be high enough to outweigh the potential adverse effects of the procedure (Tiller et al., 2002). RRSO is generally recommended for *BRCA1/2* mutation carriers who are over 35 years of age and have completed childbearing (Burke et al., 1997; Dowdy et al., 2004; Pavelka et al., 2007); this age is selected because it “represent[s] an acceptable tradeoff between cancer risk reduction and the preservation of hormonal function,” (Dowdy et al., 2004, p. 1115). Women who have not yet completed childbearing or are unsure about their desire for children or more children are not ideal candidates for RRSO. Since risk of ovarian cancer does not increase significantly until the late thirties for *BRCA1* carriers (Ford, Easton, Bishop, Narod, & Goldgar, 1994) and the late fifties for *BRCA2* carriers (M. C. King et al., 2003), many women are able to delay the procedure until after childbearing is completed (Pavelka et al., 2007). However, the beneficial effect of RRSO is greater the earlier it is performed, with the greatest risk-reducing effect observed among pre-

menopausal women (Armstrong, Schwartz, Randall, Rubin, & Weber, 2004; Kramer et al., 2005).

The risk-reducing benefits of RRSO include a near total elimination of risk of ovarian cancer (although residual risk of uterine and peritoneal cancer remains) and about a 60% reduction in breast cancer incidence if RRSO is completed by 35 (Rebbeck et al., 2002), with breast cancer risk reduction benefits decreasing with age thereafter (Pavelka et al., 2007). With more and more women delaying childbearing into their thirties and forties (Arnett, 2004), *BRCA1/2* carriers who delay RRSO in order to have children may be exposing themselves to significant levels of ovarian cancer risk and may lose some of the protective effect of the surgery (Kauff & Barakat, 2004).

With regard to acceptance of RRSO, there is significant overlap with the predictors of RRBM. Rates of acceptance of RRSO among known *BRCA1/2* mutation carriers may be as high as 64% (Meijers-Heijboer et al., 2000). In most studied samples, a majority of women aged >35 years who have completed childbearing elect to undergo RRSO (Kauff et al., 2002; Meijers-Heijboer et al., 2000). Cancer worry, high perceived risk of cancer, a belief that surgery will prevent ovarian cancer, and being aged 40 to 54 are strong predictors of the decision to undergo RRSO (Fry et al., 2001; Meijers-Heijboer et al., 2000; Miller, Fang, Manne, Engstrom, & Daly, 1999); reduction of cancer worry seems to be the single most commonly cited motivation for undergoing RRSO (Fry, Rush, Busby-Earle, & Cull, 2001).

In the Dutch study discussed previously, 60% of unaffected *BRCA1/2* carriers elected to have RRSO, many of them at the same time as their RRBM and most within nine months of discovering their positive mutation status (Meijers-Heijboer et al., 2000).

Those most likely to elect RRSO were parents between the ages of 40 and 54. It should be noted that the low rate of acceptance of RRSO among women aged 55+ is likely due to the fact that the breast cancer-risk-reducing benefits of RRSO decrease significantly post-menopause, and undergoing the procedure after the age of 55 is unlikely to reduce breast cancer risk considerably (Kramer et al., 2005). Also, the short time between receipt of test results and undergoing surgery should be considered in the context of the family cancer histories for the women studied; many of them had been under regular surveillance for several years due to family history and had had ample time to consider risk-reducing surgery as a prevention option prior to learning of their actual mutation status (Meijers-Heijboer et al., 2000).

A recent study of 359 high-risk women (120 of whom were confirmed *BRCA1/2* carriers) investigated the predictors of decision to undergo RRSO and found that older age, being married, lower level of education, and being postmenopausal were all associated with choosing RRSO. The authors note that this latter finding may be attributable to the decreased incidence of negative post-surgical outcomes for women who have already experienced natural menopause, even though the benefits of RRSO decrease after menopause (Madalinska et al., 2007). This study also noted that “women who opted for [RRSO] perceived their health as significantly worse...and reported significantly higher levels of worries...and intrusive thoughts...about ovarian cancer” than women who chose screening (p. 302). They were also less likely to perceive ovarian cancer as curable. Other studies of women at high familial risk of ovarian cancer (including *BRCA1/2* mutation-carriers) have noted that older age (Fang et al., 2003; Tiller et al., 2002; Watson et al., 2004), greater perceived risk of ovarian cancer (Fang et al.,

2003; Schwartz et al., 2003), higher perceived benefits of RRSO (Fang et al., 2003), higher levels of cancer anxiety (Meiser et al., 1999), parity (Watson et al., 2004), family history of ovarian cancer (Schwartz et al., 2003), and early breast tumor stage (Meijers-Heijboer et al., 2000) are associated with electing to undergo RRSO.

An Australian study demonstrated that desire to undergo RRSO was associated with high ovarian cancer anxiety, but was not associated with high objective risk (Meiser et al., 1999). In Britain, a study of women who had recently undergone RRSO categorized decision-making factors into pros and cons regarding the surgery. Pros included fulfilling obligations to family members and reducing ovarian cancer fear/anxiety; cons included not being able to fulfill social obligations, residual risk of cancer, upsetting the natural balance of the body, surgical complications, surgical menopause, and potential effects on body image/sexuality and relationships (Hallowell et al., 1998). Another study investigated which factors were most important for women considering RRSO; the top three were reducing risk of ovarian cancer, reducing cancer worry, and age (Fry, Rush et al., 2001). Women who chose surgery over screening placed higher importance on “reducing cancer worry, age, worries about effectiveness of ovarian screening, reducing risk of ovarian cancer, and loss of ‘periods,’” (Fry, Rush et al., 2001, p. 581). Premenopausal women placed higher importance on age and need for hormone replacement therapy than did their postmenopausal counterparts. Finally, a recent study examined 88 US women with a median age of 42, and was unique in that it examined RRSO decision-making several years after genetic testing, rather than in the 6-12 months immediately following disclosure as is common in previous studies. Results indicated that predictors of RRSO included older age at genetic testing, having children, non-

Hispanic white race, having already had a mastectomy (either as breast cancer treatment or as a risk-reduction strategy), and having a family history of ovarian cancer (Bradbury et al., 2008)

Regarding rates of acceptance of RRSO, one study found that only 13% of a sample of 29 confirmed carriers underwent RRSO within one year of receiving their mutation test results (Lerman et al., 2000), while a study of RRSO uptake in women who had known about their mutation status for several years reported uptake of 70% (Bradbury et al., 2008). However, it is important to note that most of the research on acceptance of and motivation for RRSO has been done using women in their late thirties, forties, and fifties as participants. We know far less about the experiences of young women with regard to decision-making about and planning for RRSO, and how these might impact other dimensions of young adulthood (*e.g.*, expediting couple and/or family formation in order to proceed with RRSO by the recommended age). Pilot data indicate that these factors are, in fact, quite salient for young mutation carriers (unpublished data).

Several studies have investigated post-surgical changes to quality of life, and have found that several improvements do result from RRSO. Beneficial effects have been noted with regard to cancer-specific distress and perceived cancer risk; however, they also note that decreases in quality of life are notable in terms of sexual functioning and vasomotor symptoms (*i.e.*, “hot flashes”) (Elit et al., 2001; Robson et al., 2003; Tiller et al., 2002). In a study comparing women who opt for gynecological screening to manage their increased ovarian cancer risk with women who choose RRSO, the RRSO group reported significantly lower overall quality of life and reported that the surgery did not relieve their cancer-specific distress, nor did it worsen sexual functioning (Fry, Busby-

Earle, Rush, & Cull, 2001). A second study comparing these groups in more detail found that both reported similar overall quality of life (quite high) and intrusive thoughts. However, the RRSO group reported lower levels of cancer worry, including worry about ovarian cancer for self, worry about family members, and worries affecting mood and/or functioning. They reported that their concern about developing both breast and ovarian cancer had decreased substantially as a result of surgery. They also reported more sexual discomfort, lower levels of pleasure and satisfaction during sexual intercourse, and more endocrine symptoms than the screening group (Madalinska et al., 2005); this is consistent with more recent research in which nearly a quarter of women who elected to undergo RRSO reported a worsened sex life (McArthur et al., 2007). These findings are important because post-surgical sexual symptomatology is one of the best predictors of satisfaction with RRSO (Robson et al., 2003).

With regard to psychological outcomes, there are consistent reports of reduction in ovarian cancer anxiety in the years following surgery (Meiser, Tiller et al., 2000; Tiller et al., 2002). However, there are also reports of other, negative sequelae to PO, including greater interference with work functioning, interference with social activities, and both physical and emotional symptoms than among comparable groups of women who choose surveillance, especially those who are pre-menopausal at the time of decision-making (Fry, Busby-Earle et al., 2001).

The majority of women who undergo RRSO are satisfied with their decision (Tiller et al., 2002). In one recent study comparing women who chose RRSO and those who chose surveillance, fully 97% of women in the RRSO group reported that they were satisfied with their decision, compared with 82% of women in the screening group and

consistent with other, similar research (McArthur et al., 2007; Meiser et al., 2000; Swisher, Babb, Whelan, Mutch, & Rader, 2001). Many women who have undergone RRSO report high levels of post-surgical satisfaction, that they would choose to do so again, and that they would recommend the procedure to a friend in a similar situation (Madalinska et al., 2005; McArthur et al., 2007; Swisher et al., 2001).

Decision-making. As previously stated, mutation-positive women are faced with difficult choices in selecting a method or methods of cancer prevention across the lifespan, which is further complicated by the failure of any strategy to completely eliminate risk (Dowdy et al., 2004). Surveillance is an appealing option for young women who are not willing to sacrifice their fertility and/or ability to breastfeed, but leaves them maximally vulnerable to cancer development; use of this strategy is especially risky for women in families with a history of very early-onset cancer. Chemoprevention is likely to be used by women who wish to delay both surgical intervention and childbearing, and can be considered a sort of “middle ground” method of risk reduction. However, women who are actively engaged in family formation cannot participate in chemoprevention. Chemoprevention is differently efficacious for women with various mutations, and has differential impact on breast and ovarian cancer risk, creating a sometimes confusing tangle of considerations for its use. Finally, surgical risk reduction via RRBM or RRSO, while the most effective method of risk reduction, is also the most drastic, complicated (physically and, perhaps, financially), and final. Choosing to undergo a risk reducing surgery requires that a woman is willing to give up her ability to breastfeed or to bear (more) children, and because many young adult women are not ready to make this

sacrifice, risk-reducing surgery is not seen as an option, requiring extended reliance on surveillance and/or chemoprevention.

Research suggests that women who have participated in genetic counseling are more likely to adhere to screening and risk-management recommendations, and that counseling participation is associated with use of risk-reducing surgery (Scheuer et al., 2002). However, genetic counseling is not provided for all women who undergo testing for *BRCA1/2*, and even for those who receive it, the content and quality of services is not always consistent from provider to provider (Wang, Gonzalez, Milliron, Strecher, & Merajver, 2005). Further, as previously discussed, women who receive their *BRCA1/2*-related medical care from specialists trained to address their specific needs, rather than from general practitioners or Ob/Gyns who may be less familiar with the options, risks, and recommendations for mutation carriers, may be better informed and equipped to make decisions over time that best meet their needs and maximally mitigate their breast/ovarian cancer risk (de Bock et al., 2001).

In sum, young adult women who are aware that they carry a mutation in *BRCA1* or *BRCA2* face challenges that make them unique from both non-carrier peers and older carriers. Young adult mutation carriers with high levels of perceived risk may find themselves reluctant to become involved in couple relationships due to fear of inflicting emotional and psychological distress on a loved one. Those who are in relationships must navigate the complexities of couple relationship development and communication with regard to their health concerns, and may face discrimination from partners and potential partners due to their high-risk status. In accomplishing goals related to family formation, these women may face complex decisions about *how* to go about having children,

including whether or not to have biological children, whether or not to attempt to reduce risk of passing on one's mutation, and how to manage their own cancer risk during their active childbearing years. Finally, decisions about risk-management and risk-reduction can be especially challenging for young women, as the most effective strategies for reducing risk (i.e., surgeries) are often in conflict with young women's ideals about physical attractiveness and sexuality, and other strategies (i.e., chemoprevention) may run counter to goals for family formation.

Clearly, the complicated and sometimes conflicting considerations and research findings regarding methods of risk management and/or reduction create a difficult dilemma for female *BRCA1/2* mutation carriers. This situation is confusing enough for women who are approaching menopause and who have completed childbearing; facing these decisions during young adulthood adds a considerably more complicated layer to the decision-making process, because these women are more likely to have to balance all of this information with their own desires, plans, and relationship issues regarding couple and family formation. To better understand this experience, we should examine it from a theoretical perspective. To do this, a combination of the Biopsychosocial Perspective and Life Course Theory will be utilized.

CHAPTER 3: THEORETICAL APPROACHES TO UNDERSTANDING THE *BRCA1/2*-POSITIVE EXPERIENCE

As with most phenomena studied in the social sciences, there are many frameworks that might be helpful in understanding the processes by which young women move through the BRCA mutation positive experience. Because this research is positioned at the nexus of the family science and medical genetic fields, and even broader array of theories are applicable. A full list and description of applicable theories would overwhelm this project; in the interest of clarity and brevity, I select concepts from two primary theories and two secondary or supplemental theories. From the medical genetics field, the Biopsychosocial perspective, examined through the lens of attachment theory and anticipatory loss, will allow for exploration of the range of contexts in which individuals experience health-related stressors and how biology relates to the individual and the larger system in which she is situated. From the field of family science, Life Course Theory and a feminist perspective will be used to examine and understand process and meaning. Each of these theories was selected and prioritized because of my perception of their applicability to the issue at hand.

The Biopsychosocial Perspective

The biopsychosocial model of health and illness considers the interaction between individual, family, and illness development. It was first proposed by George Engel in the 1970s, and grew out of his belief that medical and psychological issues must be considered in tandem in order to fully understand the human experience of disease. Engel believed that biochemical defects/abnormalities were only one factor in the cause of

disease, and that thoughts, emotions, decision-making, and relationships also play a significant role in disease development, coping, and recovery. He argued that

a medical model must also take into account the patient, the social context in which he lives, and the complementary system devised by society to deal with the disruptive events of the illness, that is, the physician role and the health care system,” (Engel, 1977, p. 132).

A biopsychosocial model is also thought to more fully absorb patient perceptions of themselves as “sick” or “well,” as these self-evaluations are not always based purely on biochemical functioning; in other words, the biopsychosocial model expands upon the biomedical model in that it allows for the understanding and addressing of somatic conditions by the physician or other healthcare professional (Engel, 1977). This expansion allows physicians, mental health providers, and other healthcare professionals who adopt a biopsychosocial view to more readily attend to general patient “stress” and not just to the physical causes and manifestations of illness.

Engel outlined the levels of the “hierarchy of natural systems” that should be considered as part of a holistic biopsychosocial perspective. Each system level acts both as an organized whole and as a component of a larger system. From narrowest to broadest, they are: cells, tissues, organs, nervous system, **person**, two-person, family, community, culture-subculture, society-nation, and biosphere. He noted that the person can be conceptualized as the bridge of two smaller hierarchies; the person is both the largest unit of the organismic hierarchy, and the smallest unit of the social hierarchy. Each level influences and is influenced by all other levels, without exception (Engel, 1980). In this way, Engel’s biopsychosocial model is similar to Bronfenbrenner’s Ecological Theory (Bretherton, 2004); both consider the interactions between different, hierarchically-arranged levels of analysis. The biopsychosocial perspective can be

thought of as the “public health” version of Bronfenbrenner’s model because it expands upon the Ecological perspective by including relevant medical issues and context.

Today, more than thirty years after Engel first proposed this new lens through which to view human health and well-being, the biopsychosocial model has been used in research and practice for a wide variety of health issues and is the guiding model of the field of health psychology (Anderson, 1998; Suls & Rothman, 2004). Its focus on multiple causality has received particular attention in psychological research and literature, affirming the model’s contention that multiple interacting causes and contributing factors align to create dysphoric and disease states in humans, including genetic, environmental, intrapsychic, and social factors. In other words, “biological, psychological and social processes are integrally and interactively involved in physical health and illness,” (Suls & Rothman, 2004, p. 119). For example, consider an overweight, depressed patient being seen for diabetes and arthritis by her/his primary care physician. It is likely that the patient’s weight problems contribute to both the diabetes and the arthritis. All three may also be related to genetic susceptibility. The weight problems may also be exacerbated by the patient’s depression, and the depression is likely exacerbated by the weight problems, the arthritis, and the diabetes. The patient’s ability to cope with these conditions depends largely on her/his individual traits, as well as the relationships in which s/he is embedded (*e.g.*, a family who is willing to support the patient in eating healthily will have a positive impact on disease outcome, whereas an unsupportive or emotionally absent family who eats unhealthily in the patient’s presence will have a negative impact) (Borrell-Carrio, Suchman, & Epstein, 2004). Along with this comes recognition of structural causality, or the belief that interventions suggested to

address health-related problems should be meted out hierarchically, because they have differential power to effect change. For example, for an individual with high blood pressure, cutting back on fatty foods may have a greater impact than daily meditation to reduce stress, (Benjamin, Flynn, Hallett, Ellis, & Booth, 2008) but not as great an impact as taking a blood pressure medication.

Application of biopsychosocial perspective to young *BRCA1/2* carriers. The biopsychosocial perspective provides a convenient lens through which to examine and understand the *BRCA1/2* mutation-positive experience. It is inarguable that the mutation-positive experience begins at the most basic level of analysis within Engel's Hierarchy of Natural Systems (1980), in that the cells of the human biological system carry the genetic mutation on chromosome 17 (*BRCA1*) or 13 (*BRCA2*). This mutation is present within each and every cell of the body and inhibits cells from performing their natural growth regulation processes (more information about this can be found in Appendix A). This cellular change has significant implications for the tissue and organ levels of the hierarchy, in that women at increased risk of cancer are typically encouraged to perform breast self exams and to undergo frequent screening, even if they are using chemopreventive strategies. Further, breast or ovarian tumor development may occur and bring about changes to the ability of the tissues and organs to function correctly, perhaps affecting other tissue and organ systems and necessitating treatment (*e.g.*, radiation, chemotherapy) or partial or full removal (*i.e.*, lumpectomy, mastectomy, oophorectomy, hysterectomy). Women who opt for risk-reducing tissue/organ removal during young adulthood may acutely notice an absence of tissue and organ function if it affects, for example, their ability to bear or breastfeed a child (unpublished data) or feelings about

their bodies and sexuality (Eeles, Cole, Taylor, Lunt, & Baum, 1996; Frost et al., 2000; Hallowell et al., 1998; Hopwood et al., 2000; Meijers-Heijboer et al., 2000). At the nervous system level, locating an abnormality in one's breast tissue during a breast self-exam may lead to greatly increased anxiety for an individual at very high risk of cancer. Undergoing a surgical procedure will result in physical pain, perhaps creating negative emotional and psychological associations with the chromosomal mutation. All of these phenomena happen within the person, who in turn acts within and upon the various social and environmental contexts in which she exists.

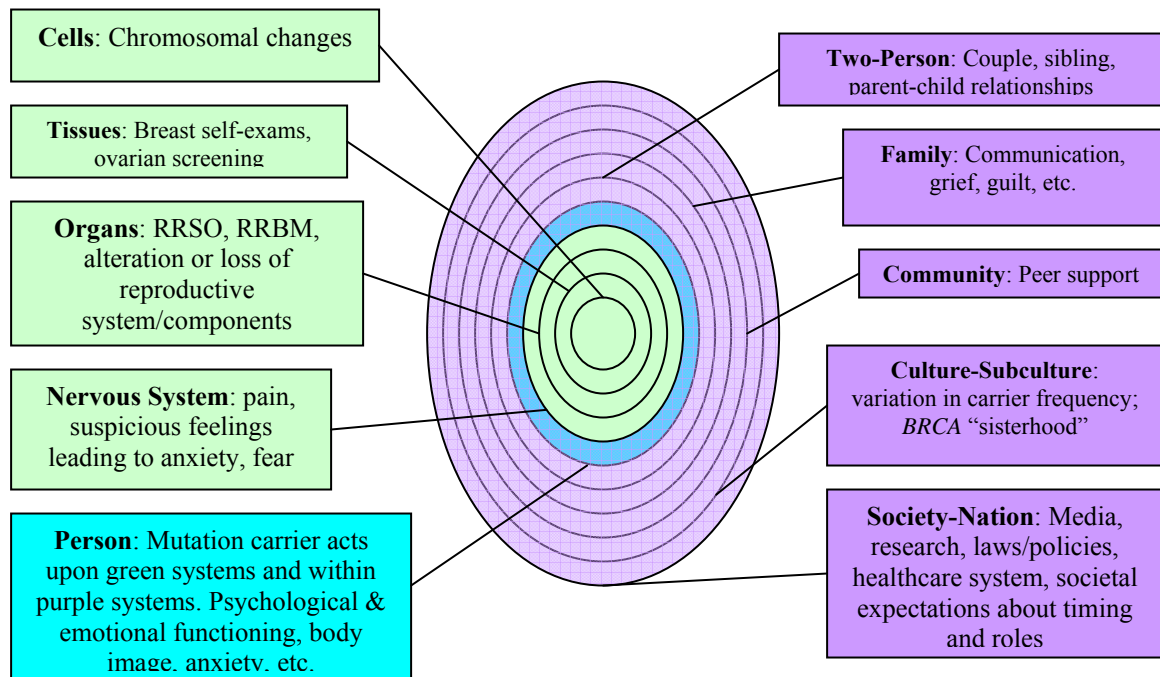
The manner in which a woman responds to these phenomena is related to her own psychological and emotional functioning, such as underlying levels of anxiety and optimism/pessimism. The person level will also be affected by various screening and treatment procedures across the mutation-positive experience; for example, undergoing a mastectomy (either for treatment or risk-reduction) will likely lead to changes in body image and sexuality, which may impact a woman's overall sense of well-being and self-perception, as discussed previously. This may have implications for the couple dyad, a major two-person system in which the individual patient is embedded. Other two-person systems of importance include the doctor-patient dyad and, where applicable, mother-daughter, father-daughter, sister-sister, and other family relationships that may affect and be affected by the mutation-positive experience.

At the family level, relationships within both the family of origin and family of choice are affected. The family of origin may experience issues related to grief, guilt, and communication about the transmission of the mutation from generation to generation and its result on the health of individual family members. Within the family of choice, a

mutation-positive individual may confront issues related to procreation, communicating with children about the mutation and its implications, or feelings of urgency regarding the formation of a family in what seems like a limited timeframe given medical recommendations. The community level of the hierarchy may be related to a woman's ability (or perceived inability) to find similar others with whom to share her experiences, fears, and successes, such as through online support networks (*e.g.* FORCE, Bright Pink). Culturally, there exist issues related to the feasibility of detecting a mutation in *BRCA1/2* or the ability of the medical community to interpret the meaning of a present mutation that depend on one's racial/ethnic background (*e.g.*, Ashkenazi background increases risk of carriership, mutations identified in the African American population may be more difficult to interpret). Additionally, a subculture of *BRCA*-positive young women has developed, in which newly-identified carriers are welcomed into a sisterhood of sorts and guided along the path toward understanding oneself as a carrier, including learning the *BRCA* lingo and how to identify resources. At the societal level, young women's experiences as mutation-positive are certainly related to popular and media representations of breast/ovarian cancer, intense research focus on the disease, and widespread beliefs about the frequency and severity of the cancers. Further, the health information, advice, and care they receive is largely a product of the system of laws and policies in the country in which they live; American women may have different options and face different barriers associated with a given risk-reduction option than, for example, women in a country with a universal healthcare policy. Finally, societal and cultural expectations about the role of women in young adulthood and the "right" timing for major events such marriage and childbearing may conflict with carriers perceived

needs related to risk-management, and managing these different expectations can become a source of stress for women in this population.

Figure 1: Biopsychosocial perspective on BRCA-positive experience



It is important to consider, when contemplating the influence of family cancer history on any one individual, the simultaneous contexts of illness, individual, and family development. Rolland notes that

cancer will influence the development of the affected individual and various family members in distinct ways, depending on several factors, including the age of onset of the illness, the core commitments in the affected individual and each family member's life at that time, and the stage of the family life cycle (2005, p. 2588).

Notably, it is not just affected individuals who are impacted by the presence of a cancer diagnoses in a family; siblings, parents, children, cousins, and other family members are often affected as well, and the experience of growing up in a family in which breast

and/or ovarian cancer is a common occurrence is likely to impact young women significantly.

Attachment theory and anticipatory loss. The current study necessitates a particularly careful examination of various interpersonal relationships, most notably the couple (two-person) levels of the biopsychosocial model. (The family level is also important to understanding the experience of young mutation carriers, but a full analysis of that set of relationships is outside of the scope of the current study). Adopting an attachment perspective can aid in understanding these relationship processes, which define young adulthood for many women.

A basic tenet of attachment theory is that the experiences we have in early relationships with parents and other primary attachment figures play a significant role in shaping certain qualities of adult relationships. The extent to which relationships with early attachment figures are secure or insecure, and how separations and losses are handled in those relationships, “contribute to shaping reactions to subsequent real or threatened separation from or loss of important others, to whom individuals seek attachment in response to threat (Bowlby, 1969)” (Hoskins et al., 2008, p. 298). Further, “secure...relational processes steady the life course through containment of distress generated by threats,” (Weihs & Reiss, 2000, p. 16), which is easily applicable to women living with increased genetic risk of breast/ovarian cancer, as they seek and acutely notice the absence of supportive attachment relationships in coping with the stressors presented by their mutations. In a clinical context, “[a]ttachment theory...provides a framework for understanding patients and their different ways of dealing with emotions, the way they tell their story, think about themselves and their dilemma, [and] their expectations and the

nature of the relationships that they establish with others,” (Evans, 2006, p. 60), supporting the use of this perspective in qualitative research.

The use of Attachment Theory as a lens through which to examine the experiences of young couples in negotiating the presence of *BRCA1/2* mutations makes the current study unique, as no other such studies currently exist. Predisposition to breast and ovarian cancer brought about by the presence of a *BRCA1/2* mutation represents an ambiguous threat of loss within couple relationships and families. Carriers and those with whom they have close relationships do not know whether, when, and where cancer will develop, and live with uncertainty regarding the future of the life course. Weihs & Reiss (2000) note that “the unpredictable nature of the cancer experience [and, by extension, the pre-cancer period] is a threat to family function,” (p. 21), and the long-term success within a couple or family unit in coping with the threat presented by cancer may be largely dependent on the security of attachment within the relational unit.

My prior research supports the attachment-related ideas put forth by other psychosocial cancer researchers and suggests that cancer risk perception may be one avenue through which attachment shapes the couple and family experiences of young *BRCA1/2* mutation carriers. Cancer risk perception is important to understanding the impact of positive *BRCA* mutation status. Significant variation in cancer risk perception has been observed among individuals who have tested positive for *BRCA1/2* mutations and other cancer predisposition syndromes. Those with very strong family cancer histories tend to overestimate risk, even after genetic counseling, a process which seeks to align perceived and actual risk (McInerney-Leo et al., 2006) and which may reveal that risk is lower than expected (Lerman et al., 1995). Lower perceived partner support has

been associated with increased cancer distress (of which perceived risk is a component) in a study of individuals in HBOC and hereditary nonpolyposis colorectal cancer (HNPCC) families (van Oostrom et al., 2007).

Cancer risk perception is directly and profoundly influenced by exposure to the cancer-related experiences of family members (Hallowell, 1999; Hopwood, 2000). Illness or death due to cancer may disrupt relationships with primary attachment figures, with implications for couple attachment in adulthood (Evans, 2006; Hopwood, 2000). Specifically, Hopwood hypothesizes that “[attachment] theory could also help to explain variations...in ability to communicate about risk: feeling free to talk openly about the loss or risk would be associated with secure attachment/realistic risk; whereas being preoccupied and communicating negatively would be associated with insecure attachment/ overestimating risk,” (2000, p. 388). Alternatively, results from our pilot investigation suggest that “those with higher perceived risk often [demonstrate] effective partner communication and [describe] healthy partner attachment,” (Hoskins et al., 2008, p. 312). An apparent balance is at play here, whereby individuals who have experienced a testing of their attachment relationships due to cancer, but not a loss, are more likely to report high perceived risk *and* healthy attachment than either those who have lost a love one to cancer (an attachment loss) or those who have not experienced a cancer-related attachment threat (*e.g.*, a female who inherited a mutation from an unaffected father and did not lose any female paternal relatives to cancer). Understanding the interplay between perceived breast and ovarian cancer risk and partner relationships is important because research has demonstrated that availability of partner support is important in formulating accurate estimations of risk (Hopwood, 2000), and partners serve as important supporters

during major life stresses, including breast cancer (Shields, Travis, & Rousseau, 2000; Wilson & Oswald, 2005).

Defined as “the experience of living with possible, probable, or inevitable future loss,” (Rolland, 2006, p. 140), anticipatory loss is certainly relevant to *BRCA1/2*+ women and those with whom they have close relationships. Anticipatory loss is characterized by feelings of uncertainty and fear, which may be managed through effective coping and adaptation (Rolland, 2006). For *BRCA*+ women, such stressors must be considered against the backdrop of family life-cycle and development (McDaniel, Rolland, Feetham, & Miller, 2006). The life course “arises from [the family/couple’s] particular history and it is guided toward the future by shared values and goals...[including] the maintenance of health,” (Weihs & Reiss, 2000, p. 19). For those seeking partners and forming permanent relationships, their life situation may trigger thoughts of threats to permanence, including illness or death (Weihs & Reiss, 2000). Women who are aware that they carry a *BRCA1/2* mutation are faced with a second, major non-normative stressor related to effectively communicating information about cancer risk to new partners, perhaps in the context of feeling “[vulnerable] to rejection as potentially defective,” (Rolland, 2006, p. 164).

Our prior research has shed light on the question of whether and how young *BRCA1/2*-positive women experience anxiety regarding partners’ reactions to mutation disclosure. They experience concern over partners’ willingness to alter individual/family life cycle plans in response to women’s heightened sense of urgency to progress through stages to mitigate the risk of cancer and pursue risk-reducing surgery. They worry that partners will be unable to respond with appropriate understanding and empathy when they share information about their increased cancer risk (Hoskins et al., 2008). Other

couple relationship dynamics may also be present. Intra-family communication patterns regarding genetic cancer susceptibility may be established as soon as awareness of genetic risk exists (Rolland & Williams, 2006). Therefore, it may be challenging for new partners to learn to navigate successfully within established family systems and communication patterns during the formation of new relationships with *BRCA1/2+* women. In permanent relationships, partners must continually navigate the challenges, demands, and stressors inherent in high-risk status as they move through the life course and make decisions ranging from jobs and health insurance to geographical location to how many children to have and when.

In many ways, living with a *BRCA1/2*-mutation is an illness experience. Although young adult breast and ovarian cancer pre-vivors often may not face an immediate health threat, they must cope daily with the threat of future illness, and therefore with some of the psychosocial and relationship implications of illness. Prior research indicates that

illness may threaten a woman's self-worth and jeopardize her ability to form intimate relationships. A young woman may be pulled *in* toward family when she is trying to differentiate, and exposed to issues surrounding body image and sexuality that may impede relationship development. When an illness experience begins during courtship, couples may realign the balance of agency and communion, allowing mutation-positive individuals to rely on partners for emotional and psychological support at a time when both might expect the dynamic to be equitable. Disclosure of a *BRCA1/2* mutation is uniquely ambiguous in that it prompts consideration of the *possibility* of eventual health problems for women and future children. However, parallels exist in that both situations force young couples to consider whether and how they can cooperatively manage health issues across the life-course (Hoskins et al., 2008).

The biopsychosocial perspective, examined through the lenses of attachment theory and anticipatory loss, is exceptionally useful for explaining the range of contexts in which individuals experience health-related stressors. The theory explains how biology relates to the individual and to the larger systems in which the individual is situated. However,

this theory does not allow us to consider process or meaning. It tells us that biology is related to other system levels, but not *how* it is related. The theory does not explain how individuals adapt to their context, to the challenges presented by their biological state, and how they make changes over time in response to shifting demands. It also falls short in accounting for meaning. The question of how individuals make meaning of the circumstances in which they find themselves, and how that meaning-making impacts emotions, attitudes, and decisions, still remains. To compensate for this and to add to the theoretical credibility of the current study, let us now turn to the life course perspective.

Life Course Perspective

The life course perspective adds to the theoretical strengths of the biopsychosocial perspective by including consideration of process and meaning. The term “life course perspective” embraces a range of different frameworks, including Life Span Development (used primarily by psychologists) and Family Developmental Theory (utilized by family science professionals) (Rodgers & White, 2004). Both of these describe the common or typical phases that most individuals and/or families move through as they age and grow. They assert that there are developmental stages relatively common to the human experience, and that significant events occur within a relatively small range of ages (Settersten, 2003) and mark transitions from one phase to the next. Generally, the six phases of the family life course include: 1) leaving parents’ home, 2) joining/creating of a new family through marriage or partnership, 3) raising young children, 4) raising adolescents, 5) launching children and moving on, 6) retirement and later life (Carter & McGoldrick, 1999). Over time, families must negotiate the spectrum of emotional processes and boundaries (both internal and external) necessary to

successfully complete each phase; this task often requires significant change in transitioning from one phase to the next. For example, raising young children may require a high level of cohesion with relatively little contact outside the family, as opposed to raising adolescents, a phase generally characterized by increased autonomy for all family members and looser boundaries between the family and the outside world (Rolland, 2005).

Life Course Theory is fundamentally relational in that it takes into account the many ways in which social interactions with others shape the experience of an individual's life over time (Bengston & Allen, 2004). The theory has, at its base, several "paradigmatic principles" that are critical to its understanding. The first is that development occurs continually across the lifespan, not just during one's early years or in spurts around significant transitions (Elder, Johnson, & Crosnoe, 2003). Therefore, human development must be conceptualized in the long-term. Not only does a life course perspective then accommodate the myriad changes and experiences that happen over the course of human life, it also accounts for the significant effects of social and environmental change that can and do occur during the human lifespan and which bring about important changes to context.

A second principle is that of *agency*, which states that "individuals construct their own life course through the choices and actions they take within the opportunities and constraints of history and social circumstance," (Elder et al., 2003, p. 11). In other words, at all stages of the life course, individuals make decisions given the opportunities and constraints with which they are presented that, over time, have significant bearing on the paths that their lives take.

A discussion of agency must be contextualized within the sociological literature from which the concept originated. Sociological theory holds that agency, considered from a life course perspective, describes “actions [that] occur with a broader sense of our futures involved, and these orientations are important for shaping individuals’ adaptations to situations,” (Hitlin & Elder, 2007, p. 182). This form of agency can also be described as “the self-reflective belief about one’s capacity to achieve life course goals,” (Hitlin & Elder, 2007, p. 182) and has social consequences; that is, a greater perception of agency is related to greater perseverance in the case of situational or structural obstacles (or constraints). Hitlin and Elder contend that although all individuals possess existential agency to make decisions willfully in situations, but the same is not true for this more nuanced form of agency that describes this ability to make decisions with a future orientation. Often, these occur at significant turning points in life that may or may not be clearly identified as such at the time. Life course agency is drawn upon in the navigation of context and structure, wherein expectations held at various levels of one’s social context (i.e., partner, family, culture) impose a set of behaviors that one “should” accomplish, which are not always congruent with what one wishes or is able to pursue.

Third, the principle of time and place asserts that historical phenomena exert considerable influence on the life course for any given individual (Elder et al., 2003). Individuals born at similar points in history will be exposed to many of the same historical phenomenon, but will react to these in unique and individual ways, resulting in variation of the path through the life course from person to person. Timing, the fourth principle, states that “the developmental antecedents and consequences of life transitions, events, and behavioral patterns vary according to their timing in a person’s life,” (Elder et

al., 2003, p. 12). That is, the same event will impact on individuals differently depending on their age and developmental status at the time that the event occurs. For example, children may be very differently affected by the divorce of their parents when the divorce occurs at age three than they would be if the divorce occurred at age 13. A three-year-old is likely to react with sadness, to have difficulty understanding what is happening, and to be quite resilient in the short- and long-term; in contrast, a 13-year-old may react with anger, more easily understand intellectually what is happening but still have difficulty coping with it emotionally, and may suffer from inappropriate boundaries with one or both parents as each attempts to align with the adolescent against the other parent by relating to the adolescent as an adult (Menning, 2008).

Finally, the principle of linked lives states that because we are interdependent, social relationships with others are the context by which humans share the socio-historical influences of the world in which they live (Elder et al., 2003). For example, children who grow up in poverty suffer not only from a lack of access to resources, but also may suffer from the negative psychological and emotional states of their parent(s), *e.g.*, depression. Further, the addition and subtraction of relationships across the life course, called “turning points,” can bring about either changes or stability to behavior, depending on the quality of their impact (Elder et al., 2003). Surrounding oneself with productive and positive others is likely to lead to productivity and positivity, while associations with others that are counter to these may decrease one’s likelihood of personal progress.

Integral to understanding family phenomena from a life-course perspective is the concept of on-time and off-time transitions. Cultural norms and traditions, along with

those that develop within families, often create dynamics whereby individuals are expected to accomplish specific tasks in a given order, sometimes with formal or informal “deadlines”. Historically, many young adults have and were expected to complete their education before getting married, and they are expected to marry by their late twenties or early thirties (Arnett, 2004). These norms are becoming more flexible given greater variability in recent decades, as young adults delay marriage in order to pursue higher education, experience a period of independence, and become established in a stable career (Arnett, 2004). However, many still feel social pressure to marry by a certain age, and this is especially true for women, who

...believe they face a biological deadline—if they wish to have children...they want to have them no later than their early thirties, because the risk of infertility and prenatal development problems rises substantially by the late thirties. But part of the pressure is also social and cultural—they fear that by the time they pass age 30 they will have missed their chance to marry because men their age will prefer younger women. The term ‘old maid’ may be used rarely today, but the stigma it represents still lingers (Arnett, 2004, p. 105).

The perception that one is making a significant transition (such as from being single to being married) “off-time” can cause stress (Settersten Jr., 2003). Life course theorists generally support the notion that, from the individual psychological perspective, having age norms for certain behaviors allow individuals to form their own “mental maps” of the life course and use those to make long-term plans and preparations, thereby fulfilling needs for predictability and order (Hagestad & Neugarten, 1985). From a sociological perspective, age norms for behaviors and transitions are a method of maintaining social order. Societal expectations for individual progress across the life course and movement from stage to stage create a set of roles that individuals feel compelled to fulfill at certain points in their lives. Internal conflict may exist when one feels that s/he is not fulfilling

societal expectations appropriately, for example, by delaying marriage or childbearing in order to pursue further education. In other words, choosing a non-normative role over a normative one can cause stress.

Application of life course perspective to the BRCA1/2 experience. Many elements of the *BRCA1/2* mutation-positive experience present significant challenges to both individual and family development. Living with increased hereditary risk of cancer may be considered a chronic health condition; although pre-symptomatic individuals are not facing illness challenges *per se*, they are still subject to a non-normative level of medical attention in the form of testing and screening, worry, decision-making, etc. These issues are likely to vary in applicability and intensity depending on one's location in the individual and family life cycle and proximity of transitions (Rolland, 2005). For example, a mutation-positive woman in her early thirties may be transitioning from a period in which she was comfortably managing her risk via chemoprevention (*e.g.*, through the use of oral contraceptives), to one in which she chooses to end chemoprevention in order to attempt to become pregnant. During this time, she may be relying solely on increased surveillance as a strategy to manage her cancer risk. This life-cycle change may, therefore, be associated with a psychologically and emotionally significant change in her experience as a mutation carrier, even though it would not necessarily be associated with development of disease. Similarly, a mutation carrier who has completed or decided to forego childbearing may choose to transition from surveillance or chemoprevention to risk-reducing surgery as a risk management strategy; this family life-cycle transition would then likely be associated with a reduction in cancer

worry as a result of the surgery, but also with an immediate increase in stress, given the physical, emotional, and financial challenges associated with undergoing surgery.

Life course concepts of *time and place* and *time* are relevant to today's young *BRCA1/2* mutation carriers for several reasons. The mapping of the human genome and identification of the *BRCA1* and *BRCA2* mutations was a significant historical event with major implications for members of hereditary breast cancer families. Prior to the mid-1990s, members of families in which many individuals had experienced breast and/or ovarian cancer may have had a sense that there was a hereditary component to their experience, but did not have any method of confirming that belief or ascertaining their individual risk. Today's young female members of HBOC families are at an advantage in that they can choose to undergo genetic testing and therefore make relatively informed decisions with regard to life choices and risk management (although some uncertainty in these contexts remains, as discussed previously); this opportunity is afforded to them by virtue of the fact that they are young adults at this point in history. In addition, with regard to *timing*, we must also consider the differential impact that knowledge of positive *BRCA1/2* mutation status will have on the lives of young women, simply because it occurs at this demographically dense period of the lifespan, when so many other important transitions are happening and roles are being fulfilled. Women who undergo testing in their twenties will have a very different experience of genetic testing and learning their results than will women in their forties, simply because this phenomenon is timed differently within the life course. Further, even within the young adulthood phase of the life cycle, issues of timing affect the context in which individuals cope with and make decisions about their risk. Women who learn that they are mutation positive before

they have completed their education or identified a permanent partner are faced with a different set of challenges than are present for women who learn of their mutations after they have married and had children.

Another important way in which life course concepts are related to the issue of cancer development in *BRCA1/2*-mutation carriers relates to some of the relevant risk factors for the disease. It is well-established that breast cancer risk is related to cumulative exposure to active ovarian function and therefore risk increases with younger age at menarche, older age at first birth, lower parity, and later age at menopause (J. Lynch & Davey Smith, 2005). Although some of these factors are simple biological issues that have little to do with behaviors, factors such as older age at first birth and lower parity are intricately tied to life course decision making, and how those choices are made by any individual can be understood through the lens of life course agency. Women of any mutation status who delay childbearing, or who make that life course transition “off-time” with regard to historical expectations about appropriate roles for women across different ages, actually put themselves at higher risk of breast cancer. This level of risk is further increased for those who carry a mutation in *BRCA1/2*, and carriers face a different challenge in navigating the constraints imposed by their mutations, acting with agency to balance often competing demands. Similarly, lower parity is related to life course decision-making, as women who commence childbearing at a later age would be likely to have fewer children overall given the fewer available years in which the ability and desire to bear children overlap. Women who choose to remain childless or form a family by means other than biological childbearing are making decisions with implications for their objective cancer risk, and often doing so after careful consideration

and thoughtful decisions about their needs and desires related to personal/family and risk-management goals. Both groups are increasing their breast cancer risk due to increased exposure to active ovarian function prior to or in the absence of first birth.

Synthesis of Theories

A synthesis of life course and biopsychosocial theories provides the ideal theoretical lens through which to understand and explain the unique challenges faced by women who are aware of their positive *BRCA1/2* mutation status during young adulthood. Responsible qualitative methodology calls for consideration of process, context, and meaning in understanding how a given phenomenon affects members of a relevant population. In the current study, the biopsychosocial perspective provides a lens through which to understand context, while life course theory allows for understanding of individuals' unique processes and the manner in which they make meaning of their experiences. The two theories marry nicely in their conceptualization of multiple system levels that influence individual behavior. A woman in the potentially lengthy, chronic "pre-vivor" phase of the *BRCA1/2* mutation-positive experience, making decisions in light of knowledge of her mutation, is reacting to a significant phenomenon that is present at the cellular level, which influences the tissue, organ, and nervous system levels in her body, as discussed previously. In part, she must make life decisions concerning relationships, family formation, and risk management in response to the limitations and risks imposed on her by these intra-individual biological constraints. Her choices are made within the context of the various two-person, family, cultural, and societal environments in which she exists, and may be significantly impacted by the broader events that occur at the societal level during her lifetime (*e.g.*, availability of certain

treatments, or medical opinion regarding the efficacy of available risk-reduction strategies). Choices are also influenced by the manner in which a mutation-positive woman and those with whom she has close relationships have navigated the crisis phase of the *BRCA1/2* experience (discussed next), or the extent to which they have successfully integrated their awareness of the mutation with their identities and conceptualization of their lives going forward (Rolland & Williams, 2006).

The biopsychosocial model of illness development can “provide a better link between the biological and psychosocial worlds, clarifying the relationship between chronic illness...and the family experience,” (Rolland & Williams, 2006). A key concept of this model is the *time phase of illness*, which provides insight regarding individual and family processes in the context of living with genetic susceptibility to cancer.

Understanding the time phase of a health condition allows patients, families, and healthcare providers to recognize and plan for the changing demands of a health issue, and to recognize the unique challenges relevant to each phase of the illness experience. The three phases are the crisis phase, the chronic phase, and the terminal phase. During the crisis phase, affected individuals and members of the family/couple systems in which they are embedded are challenged with creating meaning for their health condition that preserves a sense of mastery. Often, they must grieve the loss of their pre-illness identity, reconciling their illness status with the entirety of their lives going forward. They must develop flexibility in order to be able to respond to the stresses of uncertainty and possible loss (Rolland & Williams, 2006), work to become accustomed to a new understanding of normality, and learn to live with illness-related treatments, symptoms, tests, etc. This phase is clearly applicable to young *BRCA1/2*-positive women in the

period immediately following genetic testing, during which they may struggle to create meaning with regard to their mutation and come to a personal understanding of *why* the mutation has affected them, and find a way to feel powerful and capable of handling the challenges associated with the mutation. They may deal with anger, disappointment, fear, loss, and other difficult emotions during this phase. For young women especially, the crisis phase may be characterized by a significant shifting of life plans, including long-held ideals about when major events such as marriage and childbearing might occur. The current study can further our understanding of this dynamic because it includes many participants who are moving through the crisis phase, or have recently completed it.

The chronic phase of illness is characterized by learning to live with the day-to-day challenges of an illness condition and preserving or redefining one's developmental goals and those of the larger system within the individual and family developmental goals given the limitations imposed by the illness (Rolland & Williams, 2006). This phase is relevant for many of the women recruited for this study. It should also be noted that for *BRCA1/2*-positive women, the "chronic" phase of this health condition may continue indefinitely; in fact, for those who never develop cancer, the chronic phase may last the rest of their lives. Therefore, the stresses, challenges, and strategies for success applicable to this phase may become quite "normal" and be viewed as part of everyday life by women in this population.

The terminal phase of illness is related to issues of death and dying and provides an opportunity for acknowledging an imminent loss and saying goodbye (Rolland & Williams, 2006). Though this phase is not likely relevant for *BRCA1/2* mutation carriers during young adulthood, it may be seen by some as looming on the horizon given

mutation positive women's recognition of a higher risk of cancer-related mortality and a potentially shortened lifespan. In sum, the concept of the time phase of illness (or, in this case, the time phase of a health-related pre-illness condition) speaks to the challenge facing young women in reconciling their personal and family goals and desires with the constraints placed on them by the presence of a *BRCA1/2* mutation (e.g., completing childbearing by a certain age in order to move forward with risk-reducing surgery, or balancing family formation with chemoprevention).

With a focus on women's lives and experiences, I draw on a feminist approach, which can further serve to link the two major theories. Gordon (1979) defined feminist theory as "an analysis of women's subordination for the purpose of figuring out how to change it," (p. 107). This focus necessitates attention to how gender and gender relations (as well as class, race, etc.) shape social experience, and how the social construction of differences and power relations between genders (and other groups) impact women's lives. Feminist theory purports that gender is a socially constructed system, in which inequitable expectations of men and women shape how individuals operate within their relationships and broader social settings. Many variations of feminist theory exist, but all concur that women's experiences, values, and activities are meaningful, important and deserve to be acknowledged (Acker, Barry, & Esseveld, 1983). Feminists argue that unequal distribution of power between genders promotes feelings of powerlessness in women, in which they may feel that something is intrinsically wrong with them rather than recognizing the cultural flaw (Osmond & Thorne, 1993).

This clearly has application in the context of young female *BRCA* mutation carriers and in considering the impact of mutation-positive status on women's

experiences in families and relationships. Women's angst at deciding whether or not to disfigure their bodies to prevent cancer is largely related to their fear that they will no longer fit the cultural ideal of beauty, and specifically that a man (or men in general) will find them less attractive, challenging women's socially constructed roles as partners and their perceptions of themselves as sexually desirable. They also worry about how altering their fertility or ability to use their breasts to nourish their children will conflict with their roles as mothers. Finally, *BRCA1/2* mutation carriers may experience an exaggerated conflict over work/family balance when they are told that they must consider accelerating their plans for family formation in order to move forward more expeditiously with risk-reducing surgery, likely requiring them to make sacrifices to their educational and/or career plans and goals. A feeling of powerlessness may be the result of believing that one's choices regarding couple relationships, family, and career are being limited by the presence of a mutation and the changes to one's life trajectory that it necessitates.

An important feature of research guided by feminist theory is the inclusion of women's subjective knowledge of their own lives. Capturing this requires researchers to incorporate the many voices of women, affording them the opportunity to articulate and interpret their own life experiences (Lugones & Spelman, 1999; D. Smith, 1987).

Respecting women's unique voices expands our understanding of their actual experiences, rather than relying on distorted and false constructions set forth by the male monopoly over accounts of women's lives (Baber & Allen, 1992; Lugones & Spelman, 1999). This qualitative study provides women from this understudied population with just such an opportunity, and gives the medical, mental health, and family science communities important insight about their strengths, opportunities, and challenges.

Feminist researchers have examined women's decisions around health, work, and family, how those choices are made in the context of the cultural and structural expectations placed on women, and the impact of structural inequalities and differential access to power. Han and Moen (1999) used life course theory to describe how conflicting contexts of work and family intersect, and illustrate both men's and women's life trajectories are differently prioritized and altered within families when competition between individual and family objectives necessitates some compromise, revealing that women's career objectives more often take a backseat to what their male partners wish to accomplish, while women often find themselves managing priorities related to home and children. Marks and Lambert (1998) examined the impact of marital continuity and change on men and women in young adulthood versus middle age, and found that relationship dissolution and continuing never-married status had a more significantly negative impact on young women than on similarly aged men, presumably due to the cultural pressure women feel to partner and bear children before their "biological clock" expires. Other, more recent work has focused on physical and mental health, and how both structure and physiology shape women's choices and create gender differences in health (Bird & Rieker, 2008; Weber, 2006).

Beyond these social expectations and values, the use of feminist theory as a lens through which to examine the *BRCA*-positive experience for these young women requires a consideration of how allocation of power and resources in institutions compete with women themselves for authorship over their lives. Young women in HBOC families comprise a wide range of life experiences, and this study describes a small subset of them. The highly educated, almost exclusively white, upper-middle-class participants in

the current study are unlikely to emphasize or even see how power works on their behalf. Because they have been tested for the mutation, we know they have access to information and to medical care. By virtue of the fact that they have learned about this study, and because many of them have completed or are contemplating very proactive approaches to managing their risk, we know that they are agentive. But a group of participants with less access to power might perceive structure quite differently, and recognize how institutions, or assumptions about class/race, have constrained their choices and agency and, perhaps, contributed to less positive experiences as mutation carriers.

The feminist approach marries nicely with the life course concept of agency, in that *BRCA1/2* positive women are actively and thoughtfully making personal decisions based on the information and choices available to them in their circumstance; they are not simply being acted upon by some outside force with no individual control over the path they take through the mutation-positive experience. Further, as described by Becker (1997), young mutation-positive women amend and navigate existing cultural ideologies in their own lives, sometimes finding ways and reasons to resist these structural “shoulds” and “musts.” They are using agency in their everyday lives and finding the power to create continuity after disruption. Becker states that “people’s initial concerns after a disruption are about the loss of personal power and how to regain that power, that control over their lives,” (p. 201). The data presented in Chapters Five through Nine will make clear that mutation-positive women acutely feel this lack of power, and will illustrate the multitude of ways that is dealt with by different individuals, such as by finding like others and taking action to alter context. Like those in Becker’s study, these participants use agency to “create order out of disruption” (p. 203)

This study can aid in understanding how young women who carry mutations in *BRCA1/2* act with agency to create unique life paths given the opportunities and constraints presented to them by their status as mutation-positive – a clear link between the biopsychosocial perspective and life course theory. Their biological circumstance presents a set of significant and difficult challenges to their ability to successfully fulfill societal expectations about the roles of “partner,” “mother,” and “woman” at certain times in their lives. They must make difficult decisions when, for example, they wish to delay marriage and/or childbearing in order to experience a period of independence and autonomy or to focus on education or career development, but their doctors advise them to complete childbearing by the age of 35 in order to pursue risk-reducing surgery at the most advantageous age. The life paths of these women, then, are shaped not just by societal and familial expectations, not just by individual preferences and desires, but by the unique biological challenges and constraints presented by their mutations. Learning of one’s mutation demands consideration of how one might alter previously held beliefs and plans about the life path, roles, and timing of transitions in order to take these new challenges into account. What was previously considered “on-time” in terms of transition from one role or life stage to another may be suddenly thrown into “off-time” status by the knowledge of a mutation when, for example, delaying childbearing until one’s late thirties no longer seems like a viable or healthy option. The *timing* of the presence of the mutation in one’s life creates a maelstrom of issues and challenges at the individual, dyadic, and family levels.

This unique interface between biopsychosocial and life course perspectives is on the leading edge of public health and behavioral genetic research. When biological

perspectives on life course theory have been utilized previously, researchers have maintained that complex interactions between biological and social forces define ranges of likely behaviors (Shanahan, Hofer, & Shanahan, 2003). In recent years, life course theorists have begun to consider biological influences on behavior across the life course, including that of genetics. However, most of this research and study has focused on “behavioral genetics,” or how specific genotypes (genetic makeup) influence behavioral phenotypes (the products of genes) (Shanahan et al., 2003). For example, behavioral genetics may contribute to our understanding of how levels of warmth and empathy between parent and child are created. Some children may be genetically predisposed to a more difficult temperament, which may or may not be similar to that of their primary caretakers. In response to this difficult temperament, a parent may have more or less difficulty successfully bonding with the child, which is at least to some extent determined by his or her own genetic predisposition to temperament. The product of these interactions is the quality of the parent-child bond which, although partially determined by genes, is also the product of individual decision-making and reaction to elements of the relational system.

Recent research on the integration of behavioral genetics and the life course recognizes that “the links between genotypes and phenotypes are often heavily conditioned by social location and personal experiences,” (Shanahan et al., 2003, p. 611); that is, both genetics *and* individual histories of experiences, contexts and relationships are key in determining the set of behaviors that will be demonstrated by a person. Currently a major focus in the study of genetic epidemiology is on gene-environment interactions; life course theory can contribute to this field of research because of its utility

in examining shared and non-shared environmental influences over the life course (*e.g.*, two siblings who share 50% of their genetic make-up and some proportion of their life experience). So, life course theory can help explain why two sisters with the same mutation in *BRCA1/2* and the same family cancer history may have different outcomes with regard to their personal experience of cancer. It is possible that, for example, although they both observed and were involved to some extent with their mother's battle with breast cancer, sisters' different ages at the time of the diagnosis can create a differential psychological impact and a different personal conceptualization of cancer. A younger sister who watches her mother experience breast cancer during early puberty, at a time when her own breasts are just forming and she is in the process of becoming accustomed to breasts being part of her body and identity, may be more significantly and negatively impacted by her mother's cancer (Wellisch & Lindberg, 2000) than would a woman whose mother had cancer when she was 22, had been finished with puberty for several years, and was likely away at college and somewhat removed from the family's cancer experience. This may result in the younger sister taking more proactive steps to reduce her risk (*e.g.*, undergoing a risk-reducing surgery at a very young age) than the older sister, and thereby successfully avoiding cancer while the older sister does not. These concepts relate to factors of attachment theory and perceived cancer risk development over the life course, whereby a daughter's attachment relationship with her cancer-affected mother may, in some ways, set the stage for her own manner of dealing with personal cancer risk during young adulthood and beyond (Hoskins et al., 2008).

Research Questions

It is inarguable that women who are aware of their positive *BRCA1/2* mutation status during young adulthood, although they are dealing with normative life tasks, face a unique set of challenges that are significantly different from the experiences of both non-carrier peers and older female mutation-carriers. As thoroughly reviewed here, previous research has investigated how women in the immediate pre-menopausal years, who are typically finished with childbearing and for whom relationship status is relatively consistent, make decisions with regard to risk reduction. Only a small subset of this research has been psychosocial, and most has focused on women's decisions about and experiences with genetic testing (Lerman et al., 1994; Lerman et al., 1997; Lodder et al., 1999), the psychological impact of genetic testing (Dagan, 2005; Lynch et al., 1997; van Oostrom et al., 2007), dynamics of perceived versus actual risk (d'Agincourt-Canning, 2005; Kelly, Senter, Leenthal, Ozakinci, & Porter, 2008; Werner-Lin, 2007), and decision-making about and coping with risk-reduction (Madalinska et al., 2005; Schwartz, 2005; Tercyak et al., 2007). However, partly because of the relative recency of the widespread availability of genetic testing for *BRCA1/2* and the rapidly increasing population of young women with confirmed pre-vivor status, there has been very little research on the lived experiences of young adult *BRCA1/2* mutation carriers. Although there may certainly be some overlap with the experiences of older mutation carriers, it is important to specifically investigate this younger subset of the *BRCA1/2*-positive population in order to understand how they face their unique challenges, so that they may be better served by the medical and mental health communities.

Women in their twenties and thirties face challenges related to the transition to adulthood, including differentiation from family of origin, couple and family formation,

and career development. In the current study, I explored how young women with *BRCA1/2* mutations cope with these normative challenges and navigate the constraints imposed by their cancer risk. In other words, how do women who have been identified as *BRCA1/2* mutation carriers navigate the experience of young adulthood and, specifically, how can we understand the ways in which *BRCA1/2* mutations and breast/ovarian cancer risk perception shape and are shaped by young women's interactions in partner relationships, their decision-making about reproduction and/or family formation, and their handling of risk-reduction strategies and decisions?

CHAPTER 4: DESIGN AND METHODOLOGY

Overall Strategy and Rationale

Rooted in the principles of symbolic interactionism, qualitative research seeks to richly describe the complex ways in which people in various contexts think, behave, and create meaning (Ambert, Adler, Adler, & Detzner, 1995; K. J. Daly, 2007; Patton, 2002). Unlike the positivist approach of quantitative research, which seeks to identify statistically significant relationships among predetermined variables, qualitative research uses rigorous methodologies to explore *how* and *why* particular phenomena affect people's lives – data that are not always accessible from a simple survey or questionnaire (Creswell, 1998). Using a flexible, emergent design, qualitative researchers adapt their designs and inquiries through the process of data collection as their understandings of particular phenomena deepen or change. Adaptations may include pursuing new variables of interest as they emerge, including more or fewer participants based on saturation levels, or modifying interview questions to better explore a particular concept (Patton, 2002). Unlike quantitative data, which represent subjects and variables numerically, qualitative data use words and images to create thick, detailed descriptions of people's perspective and experiences (K. J. Daly, 2007; Patton, 2002). To do this, qualitative researchers attempt to understand and empathize with participants' experiences to the degree that they are able to capture the meanings and emotions of the studied phenomenon (Patton, 2002).

The current study was conducted using qualitative methodology for three primary reasons. First, qualitative methods and data complement the fundamental quantitative methods of psychosocial research, as well as the principles of family research. The

knowledge gained from this study about the experiences of young *BRCA1/2*-positive women can be used to create change at the individual, family, and cultural levels (Patton, 2002). In addition, qualitative research contributes to a deeper understanding of the fundamentals of family life, in terms of the diverse processes by which families create meaning and subjectively interpret their relationships and social environments (K. J. Daly, 2007; Gilgun, Daly, & Handel, 1992).

Second, Patton (2002) advises that qualitative research should be undertaken by those who believe it is the right decision personally. As a licensed marriage and family therapist who works with families with hereditary cancer syndromes in both therapeutic and research settings, I have heard young mutation carriers emphatically lament their perception that the healthcare providers with whom they interact do not sufficiently understand the unique challenges they face and wish for additional recognition of the perceived impossibility of their choices. I am deeply committed to understanding *how* individuals navigate the mutation-positive experience, and *why* they are affected in different ways psychologically, emotionally, and within their family and other close relationships; and to extending this broad understanding to other professionals with whom women in these populations interact. As a researcher and a clinician, I demonstrate this commitment through the process of hearing clients' and patients' stories, helping them to extract meaning from their experiences, and creating an environment that conveys respect and understanding for each person involved. As Patton (2002) notes, there are several principles of qualitative research that also match my personal style of quality therapy, which include: (1) recognizing and respecting individual differences; (2) collaborating with people about how they want to change rather than imposing my own

beliefs about what they should do; (3) being open to the perspectives and experiences of others; (4) allowing emotional experiences to transpire in our work together by providing an environment where others feel comfortable expressing their closely held thoughts and feelings; and (5) remaining nonjudgmental, accepting, and supportive so as to empower people to effect change in their own lives. The qualitative emphasis on my abilities to be empathic both within an interview and a therapy session brings together the research and clinical aspects of the proposed study. As a result, this study is uniquely positioned to contribute to both bodies of knowledge.

Third, there are no available data sets that contain quantitative information about the experiences of young *BRCA1/2*-positive women in navigating the tasks of young adulthood in the context of their mutation-positive status. The reason for this is that we do not yet know what the relevant issues are for women in this population, because no other researcher has yet undertaken a high-quality, exploratory qualitative investigation to allow these women to tell their stories and bring their challenges and successes to light. Therefore, a quantitative investigation into how different factors may be related is not possible. Qualitative research will serve the purpose of providing preliminary data about this topic, upon which future quantitative studies could be based, and in which variables identified as being of potential interest could be evaluated more formally.

Modified grounded theory is the qualitative approach that was used in the current study. Pure grounded theory, developed in 1967 by Glaser and Strauss, emphasizes the process of researchers suspending preconceived ideas about a specific topic or population, and allowing phenomena and theory to emerge from the data rather than strictly interpreting the data within the parameters of a pre-determined theory (K. J. Daly,

2007; Strauss & Corbin, 1998). Through the simultaneous use of both inductive and deductive analytic processes, grounded theorists listen for clues about the salience of key issues, which then provide direction as to the types of questions that should be asked to truly develop a grounded theory (K. J. Daly, 2007; Patton, 2002). Theoretical findings that emerge from qualitative data are subject to change, just as participants' experiences and stories are expected to change over time (K. J. Daly, 2007).

A more recent adaptation of pure grounded theory is often referred to as *modified grounded theory*, which seeks to unite the concepts of a pre-existing theory with emergent theoretical concepts steeped in qualitative data. This process begins with an explicit theoretical perspective, which in this study is the combination of the biopsychosocial perspective and life course theory. This perspective is to direct the initial creation of the interview questions and interpretation of findings (Patton, 2002). Theoretical concepts from each of these perspectives (e.g., on-time/off-time progression through the life-cycle; partner, family, and societal expectations) serve as *sensitizing concepts*, which are ideas that suggest a direction along which to develop interview questions and organize and analyze the data (K. J. Daly, 2007). However, while questions were developed with these concepts in mind, they do not specifically ask about women's perceptions of being on-time/off-time, or their perceptions of others' expectations regarding their choices. Rather, open-ended questions are used to ask study participants to describe their previous and current plans for progression through normative developmental stages, or to describe various aspects of their relationships with partners, family members, etc. These questions were developed in such a way that the sensitizing concepts would emerge if they are salient to the participant, but other related or unrelated

concepts may unexpectedly develop that contribute to a more holistic theoretical understanding of the topic and population being considered. As a result, modified grounded theory allows the researcher to develop a substantive theory, which is a theoretical explanation unique to a specific empirical area of inquiry, rather than a formal theory that offers explanations at a broader level (K. J. Daly, 2007). Modified grounded theory also allows researchers to investigate if and how the formal theories initially used to guide the development of a study are, in fact, useful in understanding the sample and issue being researched.

Participant Selection and Recruiting

Women aged 35 and younger who have tested positive for a deleterious mutation in *BRCA1/2* were recruited to participate in semi-structured, open-ended telephone interviews. Criteria for participation included the ability to speak and understand English fluently; completion of genetic testing for *BRCA1* and/or *BRCA2* and receipt of positive results; and having experienced in the past or contemplating at some point in the future at least one of the following: (1) creation of a permanent couple relationship; (2) family formation through biological childbearing or any other method; or (3) risk-management via clinical surveillance, chemo-prevention, or mastectomy/oophorectomy. Inclusion criteria were determined via pre-interview screening telephone calls (see Appendix E).

Site and Sample

Participants were recruited through three primary sources. Fourteen were current or past participants in the Breast Imaging (BI) study (NCI protocol 01-C-0009). Of these, one was also a member of a family involved in the Hereditary Breast and Ovarian Cancer (HBOC) family study (NCI protocol 02-C-0212). Both the BI and HBOC studies are

currently being conducted by the Clinical Genetics Branch (CGB), DCEG/NCI. The former is a pilot study of screening breast MRI and ductal lavage in 200 women from *BRCA* mutation-positive families, who are evaluated at the NIH clinical center annually for four years. Women enrolled in these CGB studies are ideal interview participants for the current research because they have developed a long-standing relationship with CGB researchers and because many of them have previously met with me to complete a novel psychosocial tool – the Colored Eco-Genetic Relationship Map (CEGRMs) – which assesses the social and relationship functioning of individuals within their social networks, including both family and friendship relationships (Kenen & Peters, 2001; Peters et al., 2006). Further, six of the fourteen women recruited through existing CGB clinical studies had previously participated in the pilot investigation undertaken to prepare for the current study, in which they took part in 45-120 minute interviews about communication with partners relative to their mutation-positive status (Hoskins et al., 2008). This previous research participation likely helped these women to feel comfortable completing a telephone interview with me, as they already knew me from a similar context and we have established a functional and friendly relationship.

The HBOC study is a multidisciplinary etiologic study of 65 hereditary breast/ovarian cancer families, half of whom have known mutations in *BRCA1* or *BRCA2* and who have been under prospective evaluation since the late 1960s. Women enrolled in the HBOC study are ideal participants for the same reasons noted previously with regard to women in the BI study, although they have not had direct contact with me. These families have a long-standing relationship with CGB, having participated in multiple studies over the years, and are open to continued participation in future studies.

As a CGB staff member, I have easy access to both of these groups and was able to present myself to them within my role as a CGB investigator, a familiar dynamic to them. Because of the unexpectedly strong response received from the FORCE community, it was decided that we would contact only the BI participants within the 18-35 age limit, rather than contacting both BI and HBOC groups. Therefore, each mutation-positive BI participant aged 35 or younger as of January 1, 2009 (n=29) was contacted by telephone and invited to participate in the study. Fifteen were willing to participate and met all eligibility requirements (as measured by the telephone screening interview; see Appendix E). Subsequently, one participant withdrew from the study for personal reasons (after the screening interview but before the qualitative interview). BI participants who also participated in the pilot study (Hoskins et al., 2008) and who wish to be interviewed for the current study (n=6) were all be eligible to participate, since the topics being studied in the current project are a significant expansion from the previous.

In addition, 24 participants were recruited from the membership of Facing Our Risk of Cancer Empowered (FORCE), an online support and patient advocacy group which serves the needs of individuals at increased genetic risk of breast and ovarian cancer. Through my research and therapy work in the hereditary cancer field, I have established a relationship with Dr. Sue Friedman, the founder of FORCE. Dr. Friedman generously offered to allow me to use FORCE's message boards, e-mail contact list, and Sue's personal relationships with young mutation-positive women to recruit additional participants. A notice about the study was posted on the web page dedicated to research opportunities for women in HBOC families, as well as on the online discussion board used specifically by young pre-vivors. A link to study information was posted in the new

visitor survey that greets each visitor to the FORCE website. Additionally, Dr. Friedman sent e-mail “blasts” to young women on the mailing list with information about the study and how to contact me if they wish to participate; these e-mails were sent periodically until recruitment goals were reached (i.e., when theoretical saturation was achieved). The FORCE study notice can be found in Appendix D.

Table 1: Recruitment Sources

Source	Description	# Participants
CGB Breast Imaging (BI) Study	Subset of BI participants who meet eligibility criteria; participated in 1-4 annual screening visits to NIH clinical center, including interviews focusing on behavioral/psychosocial issues related to BRCA	14
FORCE	Women who have voluntarily submitted contact information to FORCE and meet eligibility criteria. Invited to participate via announcements on the FORCE message boards and e-mail “blasts.”	24
Snowball Sampling	Women known to other participants and meet eligibility criteria. Interviewees were asked to inform BRCA+ acquaintances/ relatives of study and share researcher’s contact information.	2

Finally, two participants were recruited through snowball sampling, in which participants identified through the BI study or FORCE were asked to facilitate recruitment by recommending other women they know who may be eligible and willing to participate (Marshall & Rossman, 1999). In both cases, a participant referred her sister;

contact with women identified through this method was not initiated by the researcher. This strategy was utilized for its potential to yield new and important information as a result of recruiting multiple members of the same family or support network, which allowed for the examination of the different ways in which individuals within families interpret system interactions, family cancer history, traditions, etc.

True to the nature of qualitative research, the total number of interviews that were conducted was unknown at the beginning of the project. Unlike quantitative research, which bases its decisions related to study sample size upon estimated detectable effect size and estimated prevalence of an outcome in a target population, qualitative research relies on the process of “saturation,” in which interviewing ceases when stories and theoretical concepts become redundant to the researcher. Daly (2007) states that saturation is achieved when the researcher is no longer “surprised” by what she learns from individual participants during interviews, because it is familiar to what has been heard previously. While an exact number of participants could not be pre-determined, I anticipated conducting 30-40 telephone interviews. According to Daly (2007), 20-25 interviews is often a general point at which theoretical saturation is reached; however, because I utilized three distinct recruitment sources, allowing for a greater number of interviews ensured that I was able to fully capture all relevant themes and stories from members of all groups. The need for more interviews was evaluated throughout the data collection process.

The number of responses received from members of the FORCE community to postings and e-mails about the study was unexpectedly high (n=82). Because future analyses and follow-up studies are planned as extensions to the current study, we chose to

maximize data collection and continue to conduct interviews beyond theoretical saturation. However, because theoretical saturation was indeed achieved after completion of approximately 40 interviews, 40 interviews (out of a total of 60 conducted) were included in this analysis. In order to keep the distribution of participants as even as possible, we included all participants recruited from the BI study, all participants recruited through snowball sampling, and 24 of 44 participants (54.5%) of participants recruited through FORCE. Participants who were related to others who completed interviews were automatically included in the current sample in order to maximize the ability to examine members of the same families. Data from the remaining 20 participants recruited through FORCE will be analyzed at a later date. In the remainder of the current document, all references to dissertation participants pertain to the 40 individuals included in the current analysis, rather than the entire group of 60. A summary of participant demographics may be found in Table 2.

The final group of 40 participants ranged in age from 21.0 to 35.3 years at the time of the interview; the average age at interview was 29.8 years. All 40 participants self-identified as white/Caucasian, and five additionally identified themselves as Jewish. Participants had known about their positive *BRCA1/2*-mutation status for an average of 37 months (range: 1 to 97 months). The average age at which participants learned of their mutations was 26.8 (range: 19.6 to 34.8). Twenty-eight women were *BRCA1* mutation carriers, and 12 were *BRCA2* carriers; among the 40 participants, 24 unique mutations were represented (15 in *BRCA1* and nine in *BRCA2*), including all three Ashkenazi Jewish founder mutations (n = 16) and the Icelandic founder mutation (n = 1). All five participants who self-identified as Jewish carried Ashkenazi Jewish founder mutations;

Table 2: Participant Demographics

Characteristic Avg (Range)	BI (n=14)	FORCE + Snowball (n=26)	Total (N=40)
Age at Interview	31.1 (26-34)	29.3 (21-35)	29.9 (21-35)
Age at Disclosure	25.5 (21-31)	27.4 (19-34)	26.8 (19-34)
Months since disclosure	67.2 (33-97)	21.1 (1-74)	37.3 (1-97)
Characteristic n (%)	BI (n=14)	FORCE + Snowball (n=26)	Total (N=40)
Mutation: # (%)			
<i>BRCA1</i>	11 (78.6)	14 (53.8)	25 (62.5)
<i>BRCA2</i>	3 (21.4)	12 (46.2)	15 (37.5)
Ashkenazi mutation	9 (64.3)	7 (26.9)	16 (40.0)
Relationship Status at Disclosure			
Single	4 (28.6)	6 (23.1)	10 (25.0)
Serious relationship	8 (57.1)	5 (19.2)	13 (32.5)
Engaged	1 (7.1)	2 (7.7)	3 (7.5)
Married	1 (7.1)	13 (50.0)	14 (35.0)
Relationship Status at Interview			
Single	3 (21.4)	4 (15.4)	7 (17.5)
Serious Relationship	1 (7.1)	7 (26.9)	8 (20.0)
Engaged	1 (7.1)	2 (7.7)	3 (7.5)
Married	9 (64.3)	13 (50.0)	22 (55.0)
Family Formation Status at Interview			
Have kids	4 (28.6)	12 (46.2)	16 (40.0)
Want (more) kids	9 (64.3)	19 (73.1)	28 (70.0)
Pregnant	4 (28.6)	0 (0)	4 (10.0)
Risk-Reducing Surgery Completed at Interview			
Mastectomy	5 (35.7)	11 (42.3)	16 (40.0)
Oophorectomy	2 (14.3)	4 (15.4)	6 (15.0)
*family finished	2 (40.0)	4 (57.1)	6 (54.5)

the remaining 11 Ashkenazi founder mutations were carried by women who did not make explicit mention of an Ashkenazi Jewish background, and may or may not have been aware that their mutation was a founder mutation. Eight participants were related to another participant in the study; there were three pairs of sisters (one pair was recruited through the BI study; there were two pairs in which one sister was recruited through FORCE and then brought her sister into the study through snowball sampling) and one pair of first cousins among the participants (both individuals recruited through FORCE).

With regard to relationship status, there was significant variation among participants. At the time they learned that they carried mutations in *BRCA*, ten participants were single (not in a relationship), 13 were in serious/long-term relationships, three were engaged, and 14 were married (one of these was in the process of getting divorced). At the time of their interviews, nine participants were single (not in a relationship), six were in serious/long-term relationships, three were engaged, and 22 were married. Of those who were in relationships of any type at both times ($n = 28$), 24 were with the same partner, and four were with different partners when interviewed than they were with when they learned of their mutations. Eight women were single when they learned of their mutations and remained single at the time of the interview, although some of these women had a relationship in the intervening months. Three women were single when they learned of their mutations, but had entered relationships that were ongoing at the time of the interview (one was in a significant relationship, one was engaged, and one had become married).

Table 3: Participant Characteristics/Descriptions

Pseudonym & Source	Status at Disclosure	Status at Interview	Months Aware	Children	Surgery	Related
Acacia β	22, LTR	30, married	94	1 * -	(M) (O)	
Annie Γ	31, single	32, LTR	13	0 +	[M] (O)	
Audrey Γ	23, married	28, married	65	2 -	M (O)	
Beth Γ	30, engaged	30, engaged	3	1 +	(M)	
Charlotte Γ	24, LTR	26, single	21	0 +	(M)	© Kate
Chris Γ	32, married	33, married	17	1 -	M O	
Dawn Γ	27, married	27, married	4	2 -	[M] O	
Elaine Γ	34, married	34, married	2	3 -	(M) (O)	
Ellie Γ	23, single	27, LTR	44	0 +	[M] (O)	
Grace β	22, LTR	31, married	97	0 * +	M (O)	
Isabelle Γ	22, single	22, single	2	0 +	(M) (O)	§ Lilly
Jane Γ	26, married	30, married	26	1 +	(O)	
Julia Γ	24, LTR	24, LTR	4	0 ?	(M) (O)	§ Nichelle
Kate Γ	32, married ^D	35, LTR ^P	36	2 +	M (O)	© Charlotte
Kristy β	24, LTR	29, single	63	0 +	M (O)	
Leigh Γ	33, married	35, married	22	3 -	[M] [O]	
Libby Γ	31, married	32, married	11	0 +	[M] (O)	
Lilly Θ	24, single	25, single	2	0 +	(M)	§ Isabelle
Lynn Γ	19, single	24, LTR	56	0 +	M (O)	
Maelie β	28, LTR	33, married	66	3 -	M O	§ Trixie
Marie Γ	28, married	28, married	1	1 +	(M) (O)	
Marjory Γ	24, LTR	30, LTR ^P	74	0 +	[M] (O)	
MaryAnn Γ	26, engaged	26, engaged	2	0 +	(M) (O)	
Melanie β	26, LTR	31, married	57	0 * +	(M) (O)	
Monique β	21, single	26, single	61	0 ?	(M)	
Nichelle Θ	20, single	21, single	2	0 +	(O)	§ Julia
Noelle Γ	24, LTR	26, LTR	22	0 +	(M) (O)	
Pauline Γ	30, married	30, married	3	0 +	[M] (O)	
Rachel Γ	30, married	33, married	42	2 -	M O	

Reina β	25, single	29, married	54	0 +	(O)	
Rose Γ	29, married	30, married	6	0 +	M (O)	
Ruby β	31, married	34, married	36	2 -	(O)	
Rylan β	25, single	34, LTR	50	0 +	M (O)	
Sadie β	28, engaged	33, married	60	0 -	(M) O	
Serena β	23, single	30, single	96	0 +		
Shannon Γ	29, married	31, married	26	2 ?	[M] (O)	
Sophie β	23, LTR	31, married ^P	88	0 +	(O)	
Trixie β	24, LTR	27, LTR ^P	33	0 -	M (O)	§ Maelie
Valerie Γ	25, LTR	29, married	44	0 -	M (O)	
Wanda β	24, LTR	32, married	86	1 * ?	(O)	

Note. (1) Recruitment sources are noted by symbols following each pseudonym, whereby β = Breast Imaging Study, Γ = FORCE, and Θ = Snowball. (2) LTR = Long-term relationship. (3) Status at Disclosure: ^D = Divorcing (4) Status at Interview: ^P = Partner at interview is not the same as partner at disclosure. (5) Months Aware = # months between genetic testing results and interview. (6) Children: For each participant, the number of current children is listed, followed by either: - = no more children desired; + = wants more children; ? = undecided about additional children; * = pregnant at interview. (7) M = mastectomy completed; (M) = considering mastectomy; [M] = mastectomy planned; O = oophorectomy completed; (O) = considering oophorectomy; [O] oophorectomy planned. (8) Related: § = sisters; © = first cousins

With regard to family formation status, women varied both in terms of how many children they currently had, and in terms of whether and how many more children they were planning to have. Sixteen participants had children at the time of the interview, and 24 did not. In response to the question, “at this time, are you considering or planning to have more children in the future?” ten participants responded “no;” two responded “probably not;” three said “maybe,” (all three of these already had at least one child); and 25 responded “yes,” with one of these currently experiencing fertility difficulties that may challenge her plans. Four participants did not currently have children and reported that they would not or probably would not have children in the future. Eight women reported having completed childbearing, i.e., they already had at least one child and were

not planning to have any more. Four participants were pregnant at the time of the interview; two were expecting their first children and two were expecting their second.

Several potentially important differences in group demographics should be noted. For the purposes of discussion, participants recruited through snowball sampling are considered together with those recruited through FORCE because a) both snowball recruits were referred by FORCE recruits, and b) this allows for a clinical vs. non-clinical comparison (i.e., a comparison of participants previously seen at the NIH Clinical Center vs. those who were not). Participants from the BI group tend to be older than those recruited through FORCE/Snowball. One-way analysis of variance (ANOVA) revealed that this difference in age is not statistically significant. However, there is a statistically significant difference between the two groups in terms of the number of months elapsed between mutation test disclosure and interview ($\alpha = 0.00$). This makes sense when considered in the context of how BI study involvement fits into the life course of young BRCA mutation carriers. Because the BI study is a four-year process that all of the participants had either completed or were nearing completion, and because the vast majority of BI participants are aware of their mutation status at the beginning of the study, participants recruited from this source necessarily have known about their mutation longer. The range in time from disclosure to interview for women recruited through FORCE or snowball sampling included some who had known for as little as two months; this is simply not possible among women recruited through BI. Additionally, several women recruited through BI are members of families who have been involved in research studies at NIH for many years, and therefore have known about the presence of a *BRCA* mutation in the family (not necessarily in individual participants) for some time.

Generally, the FORCE/Snowball group was comprised of women whose families had learned more recently about the presence of a mutation in *BRCA1* or *BRCA2*.

The proportion of participants whose mutation is one of the three Ashkenazi founder mutations is notably different in comparing the two groups. There are several possible explanations for this. One is that because the three Ashkenazi founder mutations are easier to test for than testing for all possible mutations in *BRCA1* and *BRCA2*, members of families with Ashkenazi founder mutations may more easily access and afford testing. Additionally, because the increased prevalence of *BRCA* mutations is fairly well acknowledged in the Ashkenazi Jewish community, it is likely that individual families are more aware of the possibility that a mutation exists in the context of a family history of breast and ovarian cancer, and therefore more likely to seek genetic testing. Finally, because the NIH clinical center, where the BI study takes place, is located in a part of the country with a higher-than-average population of individuals with an Ashkenazi Jewish background, it is possible that those recruited through BI were more likely to have an Ashkenazi Jewish mutation than those recruited through FORCE, who could be from anywhere in the United States.

The number of women who had elected to have risk-reducing surgery is notable, especially given the age of the participants. The number of participants who had undergone oophorectomy was considered in two ways: as a portion of the respective groups and total, and as a portion of the subset of each group who believed that they would not have any more children, since women who desire (additional) biological children do not view oophorectomy as an option. The fact that women in the BI group were somewhat less likely to elect surgical risk-reduction of either type may reflect their

involvement in an intensive screening study and a commensurate feeling that they were being protected via surveillance and therefore did not need to consider surgery yet.

Table 3 provides an overview of pertinent individual and relationship characteristics for each participant. Narrative descriptions of each participant and her relational context can be found in Appendix H.

Data Collection and Management

After receiving IRB approval from the University of Maryland and the National Cancer Institute, recruitment efforts commenced. Prior to beginning the interviews, each participant was required to read and sign an informed consent form (see Appendix F). Consent forms were either e-mailed or mailed in hard copy to participants (based upon participants' preferences), and the interviews did not take place until forms were signed and returned in hard copy. To comply with state and federal laws regarding taping of telephone conversations, all participants were informed that the phone interviews would be audio-recorded, and provided both written consent on the informed consent form and verbal consent on the telephone prior to the recorded interview taking place. Furthermore, participants were informed that they are allowed to ask questions about the study at any point before, during, or after the interview, and that they may withdraw from the study at any time without penalty and with no effect on their ability to participate in other current or future studies through CGB.

Telephone interview data were collected in 40 one-on-one, semi-structured, open-ended interviews from February 23 to June 18, 2009. The average length of the interviews was 74.6 minutes (range: 30 to 160) and varied depending on participants' talkativeness and how many of the issues of interest each chose to speak about and at

what length. In total, 2,984 minutes of recorded telephone interviews were transcribed and analyzed for the current study. An additional 275 minutes of recorded data were folded into the current study from the six BI participants who also participated in the earlier pilot study discussed previously; therefore, a total of 3,259 minutes of interview data were analyzed for the current study. Since the participants were recruited from a wide variety of geographic locations (participants represented 20 states and the District of Columbia), all interviews were conducted over the phone. Phone interviews have several advantages over in-person interviews, including no required travel of the interviewer and participant, recruitment from a larger geographic area, and the potential for participants to feel more open in sharing their personal stories since they are not face-to-face with the interviewer (K. J. Daly, 2007). Additionally, conducting interviews by telephone allowed me to digitally record the interviews onto an NIH computer, which later facilitated the transcription of the interviews and minimized time between data collection and analyses. The interviews were digitally recorded and process notes were taken before, during, and after each interview so as to capture any thoughts or reactions I may have had related to the content and process of the interviews, as well as to document key phrases and major points expressed by the participants. All recorded interviews were transcribed by a team of hired transcribers located on the West Coast. I personally proofread all transcriptions by simultaneously listening to the interview and reading the written version.

Qualitative methodology and modified grounded theory allows for the revision of recruitment strategies during the data collection process based on emergent themes. As previously noted, *purposive sampling* was used to recruit participants from three distinct sources: CGB's BI study populations, FORCE membership, and through snowball

sampling. This varied method of sampling provided greater diversity in the sample as a whole, as it brought forth a wider range of prior experience and attitudes regarding planned behavior within the group of participants recruited through FORCE and/or snowball sampling, since they are not all current or past participants in a longitudinal screening study, as the BI participants are. Because of their participation in CGB studies, women in the BI group share several common experiences: (1) they have received similar and consistent messages regarding recommendations for fertility, risk-management, and risk-reduction from CGB staff; (2) they all received state-of-the-science screening during the period in which they actively participate in the BI study, including annual or more frequent clinical breast examination, mammography, MRI, transvaginal ultrasound, and CA-125 serum screening; (3) they were all motivated about screening enough that they chose to participate in this relatively intensive research study. However, women recruited through FORCE or snowball sampling varied more widely on each of these dimensions (i.e., messages received from their own healthcare providers; participation, or lack thereof, in various types of screening; level of monitoring with regard to their risk; length of family knowledge about the presence of hereditary cancer risk).

Because qualitative research rooted in grounded theory allows for the emergence of whatever themes the participants introduce, and because it was unknown at the start of the study whether the variables listed here would prove to be important in the women's stories, *theoretical sampling* was utilized, which means that some participants were recruited (or included in the current study from the larger pool of data collected) based on a particular idea or theme (Patton, 2002). In this case, theoretical sampling occurred in the selection of 24 of the 44 participants recruited through FORCE. Because age and

relationship status at the time of the interview proved to be important components of women's thinking about risk-related choices, an effort was made to ensure diversity of sampling by selecting women with unique features on these dimensions (*e.g.*, very young women, women who were engaged). The outcome of this was a more appropriate representation of the full range of experiences than might have been present if 24 participants recruited through FORCE had been randomly selected.

Interview protocol development. An original, standardized interview guide was generated specifically for the current study. Prior to beginning the interview, participants were informed of the purpose of this study and how the data will be used. They were informed that the study seeks to understand how women who have been identified as *BRCA1/2* mutation carriers navigate the experience of young adulthood and, specifically, how the mutation affects their interactions in partner relationships, their decision-making about reproduction and/or family formation, and how they have handled or anticipate handling decisions about risk-reduction strategies. The data are not being used to prove a specific theory or hypothesis about how women are affected by their mutation-positive status. Rather, the data are being used to better understand individuals, couples, and families in which a mutation is present, and possibly to inform couple and family therapy treatment or other psychosocial services available to young mutation carriers. In an effort to build rapport and be transparent with each participant, I shared with them that I am a licensed marriage and family therapist who works with couples and families in a medical family therapy context, as well as with individuals in the context of their participation in the BI and HBOC studies at NCI.

The interview consisted of open-ended questions that asked about the participants and their experiences before, during, and after they underwent genetic testing for *BRCA1/2* (see Appendix G for complete interview protocol). The interview protocol consisted of six sections:

- Section A: demographic information and participants' personal cancer histories.
- Section B: history of cancer in the participants' families; questions in this section were developed based on systems-based sensitizing concepts of Attachment and family communication.
- Section C: experience with genetic testing.
- Section D: issues related to couple relationships, including experiences with disclosure of positive mutation status to past, current, and/or future partners.
- Section E: intersection of participants' experiences as mutation-positive and their plans regarding family formation.
- Section F: questions regarding the past, current, and/or future use of cancer risk-management and risk-reducing strategies.

Questions in sections C through E were developed based on sensitizing concepts of attachment theory, on-time/off-time transitions over the life course, and the hierarchically interrelated nature of biopsychosocial systems, both within and outside of the individual.

Interviewing skills. As many books on qualitative research methodology have explored, the skillfulness of the interviewer is an important component of research that should not be dismissed or minimized. Daly (2007) lists seven fundamental skills of interviewing that I endeavored to utilize, which include beginning the relationship, being attentive to the participant, staying in the present, maintaining naïveté, holding to the

interview protocol, monitoring personal engagement with the participant, and maximizing the collaborative potential of the interview. To begin and build the relationship, I shared a bit of information about myself and my role as a couple and family therapist who works with individuals and families facing medical (and specifically genetic) issues. While my interview questions were developed around sensitizing concepts of biopsychosocial, life-course, and family systems theories, I remained open to emergent concepts and naïve to the uniqueness of each participant's story. Because I have not personally faced the challenges of living as a young *BRCA1/2* mutation carrier or supported a friend or family member through that experience, I am already considered an "outsider" to the mutation-positive experience; this allows me to maintain an *etic* perspective and effectively analyze the nuances of the data. This position also affords me the curiosity to understand what the experience of being a young mutation carrier is like and to understand that through each participant's lens, rather than imposing my own experiences and/or assumptions. This is in sharp contrast to the *emic* perspective, which allows the researcher to intimately share in the life and activities of the participants and their environments (Patton, 2002).

To ensure that participants perceived that their contributions are helpful and important, I responded to their stories from a stance of *empathic neutrality*, a position of understanding and caring that is free of judgment, while also providing support and recognition responses (e.g., thank you, this has been very informative so far) throughout the interview (Patton, 2002).

Privacy and ethical issues. All efforts were made to protect the privacy of the participants and their families, as well as to maintain confidentiality of identifiable

information. Prior to the start of data collection, an alphanumeric code was assigned to each participant's data in lieu of her name, and all paperwork containing participants' names and other identifying information was kept separate from the raw data. All data and paperwork associated with this study are kept in a locked file cabinet in my office in the Clinical Genetics Branch, to which only I have access. Electronic data is stored on my personal drive on the CGB computer server, which is password protected and can only be accessed by me when logged into the secure NIH computer network. Additionally, each participant chose her own pseudonym to be used in writing and presenting the results.

As Patton (2002) notes, interviewers need an ethical framework for dealing with issues that may unexpectedly arise before, during, or after an interview. Since this study explores women's thoughts and emotions regarding their experiences as mutation carriers, there was the potential for emotional hardship if the interview questions touched on sensitive or painful topics. Therefore, it was essential that I be prepared to handle situations in which participants sought therapeutic advice from me during the interview (given their understanding that I am a therapist). In the event that a participant expressed discontent as a result of her participation in the study or sought help from me for issues either related or unrelated to her mutation status, I was prepared to refer the participant to an online therapy directory where she could locate a mental health professional qualified to assist her in coping with those difficult emotions. In fact, several participants requested information about how to locate a qualified therapist in their area for assistance dealing with personal or relationship issues related to their mutation positive status. Furthermore, if a participant disclosed any information about past or present abuse of a child or disabled adult, expressed suicidal ideations, or expresses intent to harm someone else, I

was prepared to break confidentiality in order to notify appropriate authorities.

Participants were made aware of these policies at the time they provide informed consent.

No participants disclosed any such information, and therefore no reporting was required.

Data Analysis

Qualitative data are subject to both content and thematic analyses at all stages of the research, from the time of the first interview to the discussion of findings and implications. According to Daly (2007), there are four stages of analysis in grounded theory, which are: (1) open coding and the creation of concepts; (2) creating categories; (3) making linkages in the data; and (4) creating the theoretical story line. In an effort to organize the data in a way that would allow me to move smoothly through these four stages of analysis, I entered all of my raw data (i.e., transcribed interviews, transcribed focus group sessions) into the qualitative data management software NVivo, version 8 (QSR, 2008). This program allowed me to organize the data and develop and modify an electronic codebook throughout the ongoing analysis.

The first stage of the analysis, *open coding* and the creation of concepts, involves breaking down the data into manageable segments through line-by-line analysis of each transcription. Open coding is a liberal process of openly applying labels and codes (naming) to data segments that are meaningful in some way (Strauss & Corbin, 1998). During this process, the researcher sifts through the data searching for salient words, phrases, sentences, concepts, and ideas that are repeatedly used by participants. For example, multiple participants discussed their feelings of urgency about completing childbearing before a cancer diagnosis or RRSO ended their fertility; these interview segments were coded under the common specific concept “Urgency to complete family.”

As the researcher continues open coding of several transcripts, it is likely that similarities in participants' stories and the related codes will emerge, thus creating a concept around that labeled group of data. Concepts are labels associated with the salient words, phrases, and sentences, and they serve as the building blocks of theory (K. J. Daly, 2007). It is at this point in the analysis that the researcher may rely on sensitizing concepts taken from the theories to guide the development of codes and concepts (Gilgun et al., 1992; Ragin, 1994).

The second stage of analysis, creating categories, refers to the ongoing process of open coding and bringing together related concepts under a higher level of abstraction (K. J. Daly, 2007). Concepts within a category may be similar or dissimilar, but somehow are related. For example, in addition to speaking about their urgency to complete family formation goals, many participants reported having thought about whether and how their desired family size might be impacted by their mutation statuses. While each of these segments would be coded separately during open coding, this second stage of analysis would cluster these various codes into one larger concept labeled "Timing of Family Formation," because they both contain data about how participants conceptualize managing the timing of childbearing in the context of managing their mutation.

Comparative analysis, in which concepts are compared in an effort to construct categories, serves as the primary way in which the data are organized and synthesized into meaningful groups during stage two of analysis (Patton, 2002). Daly (2007) warns researchers that this is the stage where the analysis is most complex due to the potentially overwhelming number of categories and related codes that can emerge. However, this

complexity speaks to the wide range of theoretical possibilities, and so it is important to allow these numerous categories to emerge.

The third stage of analysis, making linkages in the data, is often referred to as *axial coding*, which is a reassembly of the data after completing the open coding process (K. J. Daly, 2007; LaRossa, 2005). This stage of analysis involves looking at emergent relationships within and between the categories created in stage two. As a result, several categories that are somewhat similar (have the same core “axis”) may be collapsed into larger, more abstract concepts with new properties and dimensions. For example, during stage two of coding, the categories “timing of family formation,” “breastfeeding,” and “risk perception” all emerged as important concepts. In an attempt to examine the relationship between these concepts, a higher-order concept emerged that described women’s strategies for and decisions about controlling childbearing-related issues in concert with risk-management decisions. Women who have opposing desires or perceived needs on these issues often feel as though they must sacrifice one priority or the other. This new, higher-order concept was then examined among other higher-order concepts. As the categories became more defined and unique to the data, *theoretical saturation* of each category was achieved, such that no new information emerged from the interviews to deepen the meaning of each category (Patton, 2002).

The fourth and final stage of analysis, creating the theoretical story line, involves integrating and refining the theory through the analysis of participants’ stories (K. J. Daly, 2007). Using *selective coding*, data were interpreted and carefully selected to help tell a “story” that integrates the categories created through open and axial coding. At this point, I sought to weave together the data with the theories, a process known as

abduction, to determine the core categories that will serve as the “narrative spine” for the story (K. J. Daly, 2007). As expected, the story created as a product of this research progresses somewhat chronologically through the lifecourse, beginning with their perceptions of cancer risk before and after genetic testing, then addressing issues related to couple formation, family formation, and risk-reduction in that order. However, I remained open to the various ways, aside from chronologically, in which the narrative about women’s experiences as members of high-risk families and as mutation carriers, could be storied, and in fact it is the case that not all women move through mutation-related issues in this order. For example, some women had already established couple relationships when they were tested for the mutation, and others had already completed their families at the time of testing.

It is important to note that movement through these stages of coding and analysis is not necessarily linear or consecutive. Strauss and Corbin (1990) highlight the significance and utility of researchers moving back and forth between stages as data collection and analysis simultaneously occur so as to modify and polish codes and categories in an effort to capture the authentic story emerging from the data. In practice, movement between the stages of analysis continued up to and throughout the writing process, whereby higher levels of abstraction were reached as new insights about women’s processes were discovered in the course of sifting through the data with different foci. In fact, many important insights about the abstract meaning of the data were made *after* having written drafts of each substantive chapter, which allowed me to step back and think about the story that was emerging and how different elements of that story were related to one another.

Trustworthiness and Authenticity of Qualitative Data

The trustworthiness and authenticity of qualitative research are determined by a variety of methodological and analytical components throughout the research process. Much as quantitative research focuses on validity, reliability, and objectivity of the analyses, the trustworthiness of qualitative research is measured by its credibility, transferability, dependability, and confirmability (Lincoln & Guba, 1985). To ensure that this qualitative study is trustworthy and authentic, a variety of techniques were used.

Credibility. The *credibility* of a qualitative study is determined by how well the project is designed and executed in accordance with relevant qualitative methodologies, as well as how accurately the participants' stories are portrayed (K. J. Daly, 2007; Lincoln & Guba, 1985). Credibility has been compared with the internal validity of quantitative studies, which refers to the ability to show a cause-and-effect relationship between the independent and dependent variables. According to Daly (2007), a study's credibility is strengthened by the theoretical frameworks guiding specific lines of inquiry, the ways in which participants are selected, how data are generated, and the extent to which the researcher is involved in the field. The credibility of procedures and outcomes of the current study are strengthened by the use of theoretical sampling, peer review and debriefing, triangulation, and member checking.

Theoretical sampling refers to the process of sampling based on specific information and ideas that are being sought (Patton, 2002). Since the goal of qualitative research is to show how theoretical concepts are related to a specific population, and not to generalize findings to a broader population, using a selected group of women sampled for specific emergent themes strengthens the researcher's ability to accomplish this goal. Ensuring that the study population was as diverse as possible (i.e., recruiting women

across the spectrum of ages, recruiting participants who were related to one another) also helped to increase the credibility of the study.

Peer review and debriefing is an important technique that allows the researcher to receive constructive criticism and feedback about her/his qualitative work in an environment that is supportive and non-threatening (Lincoln & Guba, 1985). Requesting and processing colleagues' thoughts, suggestions, and questions about one's qualitative work are similar to the quantitative concept of inter-rater reliability. In the context of my training in the Department of Family Science at the University of Maryland, I have formed collegial relationships with other doctoral students who are also currently, previously, or soon-to-be engaged in qualitative research. These colleagues, dissertation chair Kevin Roy, and dissertation committee member and CGB mentor Mark H. Greene, were consulted throughout the study provided opportunities to ask questions, receive feedback, and explore topical and methodological areas of qualitative research that are new or challenging. These individuals read some of the interview transcripts and reviewed the categories and emerging theoretical framework that were developed and provided feedback regarding how well they represent the data and relate to each other. Drs. Roy and Greene were also involved in the creation and evolution of the narrative spine of the final document.

Triangulation involves utilizing a variety of data collection techniques and drawing from a variety of participants with the purpose of substantiating findings and gaining a more holistic perspective of the emergent concepts (K. J. Daly, 2007; Patton, 2002). The four types of triangulation in qualitative research are data, investigator, theoretical, and methodological. For the purposes of this study, both data and theoretical

triangulation were utilized. Data triangulation refers to the use of multiple data sources throughout the study. As previously noted, study participants were recruited from three distinct sources: the BI study, the FORCE website, and via snowball sampling. In addition, the data themselves consist not only of the transcribed interviews but also of transcripts from focus groups conducted with young mutation-positive women at the FORCE 2009 Annual Conference. Further, the notes I took before, during, and after each interview to capture my thoughts, questions, and reactions to the interview process, as well as to note important points made by the participants, were referenced during the construction of the narrative produced from the data. These notes are considered an important source of supplemental data because they include qualitative information that is not necessarily captured during the actual interview (and subsequent transcription). Thus, these memos were used to triangulate the interview data and help me reach a level of saturation, meaning that no new themes emerge from the whole data set (interviews, focus groups, and memos).

I also used theoretical triangulation, which refers to the use of multiple theoretical perspectives to analyze the data from a modified grounded theory approach. As previously discussed, the combination of the biopsychosocial perspective and life-course theory, taking into consideration attachment theory and a feminist perspective, provide a more holistic lens through which participants' stories were analyzed than would exist had only one of these theoretical perspectives been considered.

Member checking allows the participants to review the researcher's analyses and interpretations of the data and to provide their reactions and suggestions for improvement (Patton, 2002). In this study, participants were asked if they would like to receive a copy

of some portion analyses and provide feedback; the participants who wished to do so were given an opportunity to provide suggestions and ask questions prior to the final defense of this study so that the findings and discussions could be constructed partially in light of their feedback. Twelve individuals from among the 40 participants were sent single chapters to read and asked to reflect on how accurately the chapter captured their experience, as well as whether they believed that the chapter conveyed a comprehensive description of the relevant phenomena. All twelve individuals responded positively and reported a high degree of enthusiasm about the manner in which their own and others' experiences were woven together to comprehensively describe the issues they had faced as young *BRCA* mutation carriers.

In addition, member checking was accomplished through the use of the FORCE focus group of young *BRCA1/2*-mutation carriers who attend the 2009 Annual Joining FORCEs Conference in Orlando, FL. Two groups of approximately 15 *BRCA*-positive women (some of whom were also study participants) were convened at the conference, and these women were asked to provide feedback about preliminary findings and themes, as well as to participate in a discussion of what they felt were the most relevant mutation-related themes in their own lives at the time of the conference. This provided an opportunity to check these focus group findings against the findings identified through the analysis phase of the study.

Transferability. *Transferability*, or fittingness, refers to the degree to which qualitative findings can be generalized to another situation or setting under similar, but not identical, conditions (Patton, 2002). This naturalistic term is often paralleled with the quantitative concept "external validity," the extent to which the results can be generalized

to the larger population. According to Lincoln and Guba (1985), the degree to which findings may be applied to other contexts is heavily reliant on the researcher's field notes which should be full of *thick description* (Patton, 2002). Descriptions that are thick, deep, and rich take the reader into the setting being described, the experiences of the researcher while in that setting, and the outcomes of that experience. Patton (2002) notes the importance of separating description from interpretation, in which descriptions should not explain or answer "why," but should simply portray the experience. In the current study, attempts were made to create thick, rich descriptions of the interview experiences, so that readers will be able to decide how well these findings may transfer to other contexts.

Dependability. *Dependability* is the qualitative equivalent of the quantitative research term, reliability, which refers to how well a study and its findings can be replicated. Since the hallmark of qualitative research design is flexibility and allowing findings to emerge within the context of interactions between researchers and participants, ensuring replicability is nearly impossible and not necessarily a goal of qualitative research. Thus, Lincoln and Guba (1985) developed the concept of *inquiry auditing*, in which the process and product of the qualitative research is tracked and examined for consistency by a third party. The researcher is responsible for creating an audit trail, which may include recorded materials, interview transcripts, interview guides, lists of interviewees, lists of created categories, field notes, and written guidelines of research procedures (Schwandt, 1997). To ensure dependability in the proposed study, a written record of all products and processes related to the research was kept, and ongoing consultations with both the dissertation chair, Dr. Kevin Roy, and colleagues with experience conducting qualitative research were afforded the opportunity to "audit" the

materials (e.g., interview transcripts, personal memoirs) in comparison with the findings to ensure that the process and product of the research are sound and consistent.

Confirmability. *Confirmability* is concerned with how well the qualitative findings and interpretations are steeped in the data, and “not merely figments of the inquirer’s imagination” (Schwandt, 1997, p. 164). Similar to the quantitative concept of objectivity, interpretations of qualitative data should be as neutral as possible in spite of researchers’ values inherently guiding studies. To avoid the dichotomous argument of objective vs. subjective, Patton (2002) encourages qualitative researchers to strive for “empathic neutrality” (p. 50), in which the researcher may be empathic toward the participant but remains neutral toward the findings. This tenet of qualitative methodology is consistent with the adoption of a biopsychosocial perspective, the use of which in a clinical setting calls one to interact with patients from a position of “empathic curiosity.” This means that the clinician, or in this case, researcher, should be open to hearing unexpected things from a patient even if one believes s/he knows what to expect from an exchange (Borrell-Carrio et al., 2004). Adopting a stance of empathic curiosity/neutrality during a qualitative interview allows me to maintain openness to hearing the nuances of each individual’s experience as a unique phenomenon, rather than attempting to categorize it according to what I might already believe that I understand about an experience or group.

Similar to the manner in which dependability can be accounted for in a quantitative study, Lincoln and Guba (1985) suggest that qualitative researchers also seek a *confirmability audit*, in which a third party attests to the neutrality of the research interpretations, based on an audit trail consisting of raw data, process and analysis notes

and memos, personal notes, and preliminary developmental information. This is different from an inquiry audit, in that this audit trail seeks to demonstrate the researcher's neutrality in reporting the findings, as opposed to the other audit trail which seeks to track the analysis of data for overall consistency. In an effort to verify the neutrality of interpretations for the current study, a confirmability audit trail was developed and maintained, which consists of raw data; theoretical memos (notes about the formulation and evolution of theory); field notes with thick, rich descriptions; and a code book documenting the coding scheme as it is created and refined. All of these sources of information have been made available to both Dr. Roy and Dr. Greene so that findings and interpretations can be verified and confirmed.

It is important to note that this project is the initial attempt to extrapolate many of the important concepts that exist in this data, and it is impossible to fully describe and discuss all of the many important issues for this population in one document. Ongoing analyses are planned and will provide additional opportunities to evaluate the data, along with similar additional data, with the ultimate goal of developing a comprehensive understanding of the experiences of young *BRCA1/2* mutation carriers as they relate to navigation of young adulthood.

CHAPTER 5: RESULTS—EXPLANATION OF THEORETICAL MODEL

Ellie's Story

Ellie, a 27-year-old Ashkenazi Jewish carrier of a *BRCA1* founder mutation who had known about her mutation for 3.5 years, is the oldest child of two mental health professionals. Confident and colorful, she was one of the most enthusiastic participants and had much to share about her experience in a family touched by *BRCA*. Ellie was aware of breast cancer as a part of her family legacy, but an even more powerful narrative in her family was that of holocaust survivorship. Several of her grandparents had lived through that period and were some of the only survivors from their families. Knowing that family story had many powerful influences on Ellie and other members of her family, and one of the results was that information about who did or did not get cancer was unavailable because so many people in her grandparents' and great-grandparents' generations did not survive the holocaust. The many missing pieces of family history made it difficult for Ellie and her family to develop a comprehensive understanding of their cancer risk.

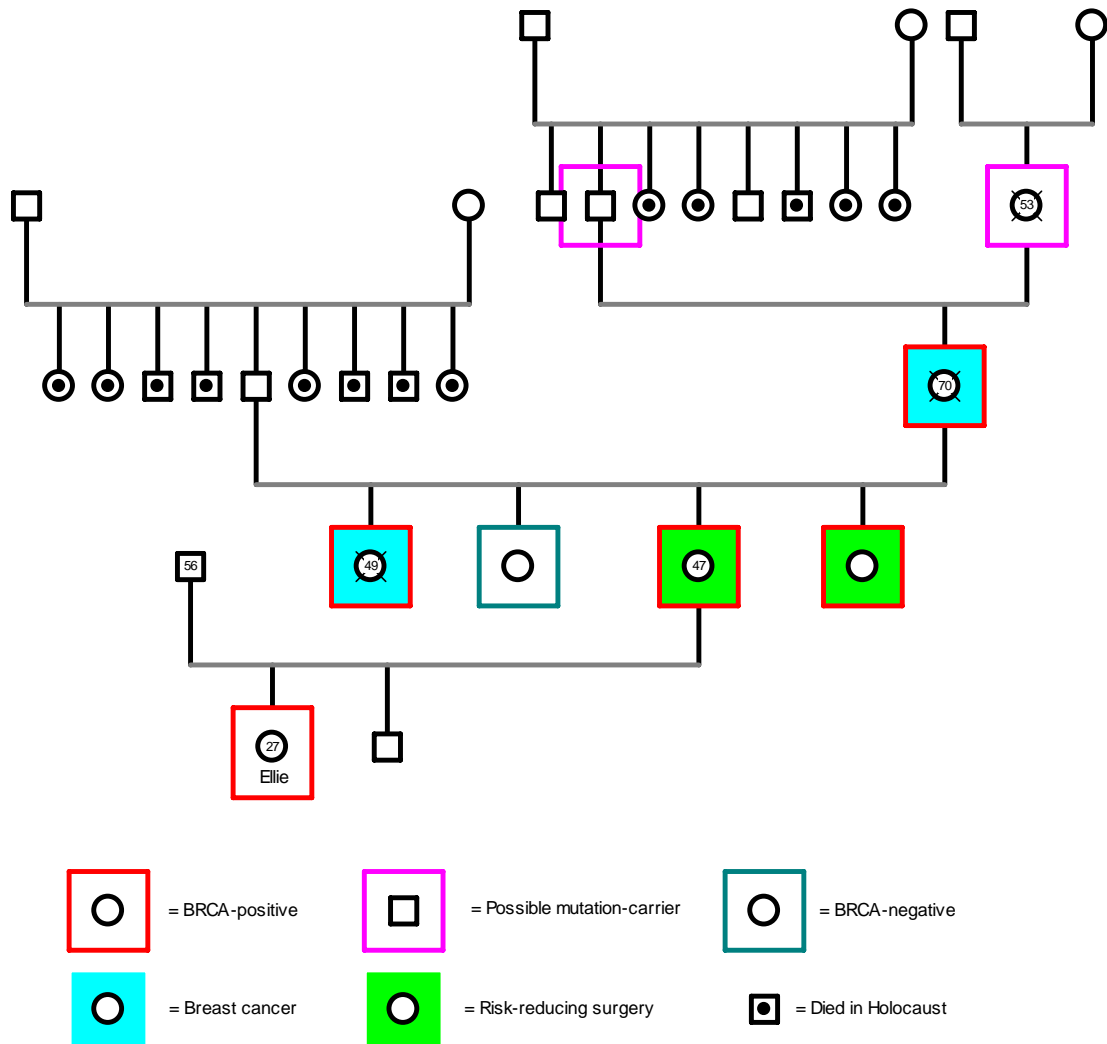
Ellie's first experience with breast cancer occurred at age eleven when her best friend's mother, in her forties at the time, was diagnosed. That experience was notable for Ellie because it was an example of someone being diagnosed at a relatively young age. Sixteen years later, that woman was still living, but her cancer had metastasized and her prognosis was poor. Ellie's next exposure to cancer occurred when she was 17 and both her maternal aunt and maternal grandmother were diagnosed around the same time. These diagnoses occurred in the late 1990s, only a few years after the *BRCA1* and *BRCA2* mutations were identified on the human genome and several years before a mutation was

identified in Ellie's family. Because they did not know that her aunt was a mutation carrier, doctors treated her with a lumpectomy and short course of chemo; the cancer quickly metastasized to her bones and brain and she passed away at the age of 49. Ellie's grandmother's cancer, however, was caught early and was less aggressive; doctors were able to bring her into remission and shortly after, she passed away in a car accident; this left an additional gap in the family cancer history because Ellie and her family could not know whether her grandmother's cancer might have come back if she had lived. The story of how breast cancer entered the lives of Ellie and her family members is striking because of its suddenness and the fact that two individuals from different generations were impacted almost simultaneously. These two cancers seemed to come "out of nowhere" because nobody in the family was aware of any other cases of breast cancer in previous generations; however, it was unknown to them whether this would have been true if members of previous generations had lived normal life spans. This lack of information further constrained Ellie and her family members ability to fully understand how cancer might have impacted the family if other circumstances had not precluded the natural medical history of members of previous generations.

Another important piece of Ellie's family narrative with respect to *BRCA* was the sense that the mutation had been identified in part because of luck: when Ellie's mother visited her regular Ob/Gyn during the time that her sister and mother were both battling cancer, he had "put two and two together and said to her, 'OK, young, pre-menopausal cancer [in] a woman who's Ashkenazi Jewish, aggressive and doesn't respond to chemo... you need to go get genetic testing.'" This was thought by the family to be lucky because it occurred so early in the development of knowledge about *BRCA*, when not all

doctors were yet aware of the mutations and their capacity to impact families and specific ethnic groups. The family encouraged Ellie's aunt to participate in the testing, and the mutation was identified shortly before she passed away. Ellie's mother and her two other sisters were all tested, and two of them (including Ellie's mother) were positive; her grandmother was never tested, but was presumed to be a carrier. However, the mutation also could have come from Ellie's maternal grandfather.

Figure 2: Ellie's Cancer Genogram



From there, the family narrative took a turn toward self-empowerment. Ellie's mother underwent both RRBM and RRSO at the age of 39, and her other mutation-positive aunt had RRBM several years later at about the same age. Both had remained cancer-free in the years since their surgeries. Ellie was armed with this knowledge as she approached the period in her own life in which she would begin to make risk-management decisions.

When Ellie was 19, her mother took her to a genetic counselor so that Ellie could learn about her risk of carrying the mutation and the implications for her own health if she was mutation-positive. Ellie recalled being exposed to conversations about *BRCA* and cancer at home, but being able to escape from thinking about it when she was away at college. Asked whether she felt pressured to get tested, Ellie replied,

It wasn't really an option. See, the way my family works, it's not like we're pressured, but she would say, "You're too young and you should wait, but when you get to a certain age you really should get tested."...If I [had] said, "You know what mom, I really don't want to get tested," my parents would have said, "Okay, that's your choice," but the expectation was that I would get tested.

The time when Ellie felt ready to get tested coincided with the end of graduate school and her decision to move away from the city where her parents lived; she was tested at age 24 and quickly made the decision to rely on screening for her risk-management at that time. She moved to a major west coast city and had trouble finding a place to get screened appropriately until a fortuitous coincidence occurred:

I was working at a middle school my first year...and it was about a month after I moved, and the Vice Principal was dealing with ovarian cancer and recovered breast cancer, and I knew she was Ashkenazi Jewish, so it kind of came up that she had the [mutation]. So, I talked to her, and she was just so thrilled to have me, but sad for me at the same time. She [said], "I know this study that they're doing at [hospital]," and she's the one that connected me to this study that I've been in.

Ellie felt fortunate to have been receiving high-level cancer screening from a program specifically designed for women who carry *BRCA* mutations, and confident that she was acting in the most effective way possible at that time to insure that she would not find herself with a late-stage cancer diagnosis and meet the same fate as her aunt.

Shortly after her cross-country move and entry into the cancer screening study, Ellie's *BRCA* mutation really began to shape her couple relationships. In the first several years after learning that she was *BRCA*-positive, she disclosed her mutation liberally and early to several dating partners; she did not think of any of these relationships as potentially permanent, so telling them about her mutation was not difficult. However, when she met Mike, she immediately had a strong sense that he was "the one." She chose to wait considerably longer to inform him about her mutation:

I didn't know it would work out for sure, but I just knew he met all the criteria and I really liked him. It wasn't like I was trying to trap him and I was waiting for the right time to tell him after he loved me, like, "Oh, I'll get him to love me and then I'll tell him." It was almost more like, "Okay, let's find out if he's the right one, and then I'll tell him because he doesn't need to know that information if he's not the right one." I approached it differently than the other guys I'd dated.

At the same time that this relationship was developing, Ellie was beginning to think about RRBM. She recalled how participating in screening for several years brought a changing sense of its appropriateness:

[At first] I felt like, okay, I know that my risks are higher, but I'm not going to deal with it for another ten years, I thought, so it's nothing I'm even going to think about. I'm just going to do my surveillance. But little did I know that doing surveillance every few months really weighs on you, and it's not like just a regular doctor exam. It's a real heavy-duty thing, and every time you go in for surveillance, you start to realize that, oh, I'm not just going in to check on things, I'm going in to see if I got cancer. You realize, this isn't me preventing cancer; this is me just finding it.

Ellie began to feel that her plan to delay risk-reducing surgeries until age 35 and after childbearing was no longer comfortable for her because she stopped feeling confident that she could do so and remain cancer-free, marking a significant transition in her approach to risk-management. This percolating decision to undertake RRBM provided the impetus she needed to begin to seriously consider disclosing her mutation to Mike:

I had to go in for surveillance and we were talking about what we were doing that day on the phone. I said, “Oh, I’m just going to this gynecologist and getting a check-up.” After that happened, I felt like I was lying to him, and I felt like I had to tell him soon because it was a pretty big thing. And I hadn’t decided yet when I was doing my surgery, but when I met him I was 25, and this was after I’d turned 26. So I was like, okay, I’m getting into my upper 20’s, gotta get more serious about this.

Shortly after this, Ellie and Mike went on a trip together, where Mike told Ellie that he loved her for the first time. Feeling sure that she loved him too, Ellie made the decision to tell Mike about her mutation:

One day he came over and I’d been talking about it with my mom, and figuring out how I was going to the FORCE conference that year, and setting up a profile on the FORCE website, and I was like, “this is just too big of a thing not to mention. [So I said], “I need to talk to you.” He thought I was going to break up with him, and I said, “No, no, no; it’s not about me and you.” ... He wasn’t expecting it, so he had no idea what I was bringing up, and I just basically started from the beginning. I said, “You know I told you I had an aunt that passed away,” and I told him how it connected to me and what I was doing now. He was just like, “Wow, it’s really heavy,” and he was kind of upset and he just said, “I still think you’re perfect.”

Ellie felt strongly that one of the primary reasons she got such a positive response from Mike was the way that she presented information to him. She believed that her ability to deliver her message directly, gently, and willingly – boldly describing her feelings about her risk and her decisions about managing it – accounted for the positive relationship outcome. Because she was closely connected to other *BRCA* positive women through

both online and in-person support groups, she had discussed this with several other young carriers:

People have asked me, “How are you so good about it? How do you approach it?” Because I’ve known girls who are in the same situation and haven’t told boyfriends yet, or aren’t in a relationship, and want to know how I go about it. I just approach it so confidently, like “This is what my situation is, it’s kind of sucky, it’s not the best situation, but it’s kind of making me stronger, and I’m taking my future seriously and I’m going to take care of it now.” I think a lot of men see that as a woman who is being really strong, and then they feel secure because they’re like, “Wow, you’re taking care of something now so you won’t get sick in the future,” where some women would have their head in the sand and ignore it, or they don’t even know they’re in my position and they’ll get sick. So if I said to my boyfriend, “I have this terrible gene, and I have such a high risk of cancer, and my kids might get cancer,” then of course the guy is going to run for the hills. He’d be like, “Holy crap!” But I don’t approach it that way.

Since he found out about her mutation, Mike had been a consistent source of emotional and instrumental support for Ellie. Mike’s ability to help Ellie think about her risk and participate in decision-making about risk-management created opportunities for closeness in the relationship. For several months Ellie considered the timing of her RRBM, but continued to participate in the screening study. Her mother suggested moving the surgery up by a few years and doing it at 32 instead of 35 as her physicians had recommended, but Ellie wondered,

“Why are we picking arbitrary numbers? Why does 32 have any more significance than 31? ... How do I know what age to pick to do this?” And then I just realized, I should just do it if I’m ready, and I started feeling like I’m going to be ready pretty soon because I don’t want to do this roller coaster ride anymore.

Ellie’s realization that decisions about managing her cancer risk were *up to her* created a situation in which her choice of surgery was much more comfortable. It was her personal choice – not a prescription from doctors or family – that mattered most.

Another important component of Ellie’s thinking about when major life events would happen was related to deciding when family formation would begin. In part, this

worry came from an early interaction with a healthcare provider who counseled her about her risk, emphasizing that she didn't have many options:

I just want to get one kid under my belt so I feel like I've started. It sounds kind of ridiculous, like that's not how life should work, but I do feel that time pressure and I want to have one kid out of me by the time I'm 30. It's just an idea I have in my head that I should do that. I think it came from knowing my risk. And also when I was 25 and in my study, my breast surgeon... said to me, and it's always stuck with me... "You know, your risk is still low, but to be honest, if you were married and had 3 kids, I would tell you to get the surgery now." It's just hearing that, it was like, "Oh, wow, maybe I should get on that." It wasn't like he was the sole person that convinced me to want to do that, but statements like that, when I let them sink in, I'm like, "Wow, I really don't have a lot of time to wait."

As their relationship became more serious and talk of marriage became frequent, Mike was one of the first people to effectively reflect what Ellie was thinking. He suggested that they schedule her RRBM for the upcoming summer during her break from work, before they were married and wanted to begin a family, so that the pressure to have the surgery would not be a factor during the years that they were focused on childbearing.

Ellie decided

that [was] a really good point, because a lot of people want to save their breasts until after they have a couple kids so that they can breastfeed and enjoy that, but ... the more people I talk to and the more I talk to my mom about it, it was like I would rather have my kids and bottle feed them than breastfeed them and not be there to see them grow, and die.

With Mike's full support, Ellie had scheduled her RRBM for several months after her interview and was looking forward to being able to put breast surveillance behind her and find some relief from her worry about breast cancer; she anticipated a drastic reduction in breast cancer risk perception after surgery, although she noted that she would still worry about her ovarian cancer risk and about residual risk after RRBM because she was choosing to have a nipple-sparing mastectomy. However, she noted that "if I'm really uncomfortable and I feel stressed about it in years to come, I can always go back and take

them off, but if I feel better about it, then great.” Ellie’s ability to take control of her risk-management plans and make significant changes to what she thought she would do to manage her risk further demonstrate her capacity to make and implement choices. Her story also illustrates how Ellie and Mike’s close couple bond provided them with the necessary resources to move past the perceived difficulties in starting a family in the face of a cancer risk; being able to eliminate breast cancer risk prior to family formation will allow Ellie and Mike to experience the early years of her children’s lives nearly free from worry that breast cancer will impose itself.

Though Ellie had no doubt about whether she and Mike would continue their relationship, she did describe how they had negotiated an acceleration of their relationship to accommodate her plans for risk-management. Having resolved the question of when RRBM would occur, Ellie remained aware that RRSO would also need to be completed at some point. Because she and Mike definitely wanted to have biological children, RRSO had to occur after childbearing was complete.

That’s been a compromise, because when it first came out about *BRCA*, I told him because of that, but also because I feel ready, I don’t want to have kids when I’m 34 or 35 – I have to start sooner. So, the plan is we’re going to get married when I’m 28 and he’s 30, which would mean we’re supposed to get engaged pretty soon. So I told him if we get married when I’m 28, I’m OK with having a kid at 29 or 30 if I can get pregnant right away. But the time we get married, we’ll have been together 2 ½ years, which is not so long, but if we have a baby by the time we’re together 3 ½ years, I don’t think that’s ridiculous at all. I don’t think that’s a crazy expectation, I mean, in Los Angeles, saying you’re having kids before 30 is kind of an anomaly. Most people get married later here and I don’t hear of people having kids until they’re like 35, but that isn’t going to jive with me.

Their ability to agree together on a plan for achieving major milestones of young adulthood became one of the major keys to the success of their relationship; had this not happened successfully, Ellie might feel much less secure about her ability to achieve both

her risk-management and family formation goals in the context of her relationship with Mike. Ellie's experience also illustrates how young women who carry *BRCA1/2* mutations may adjust their plans in a way that does not meet with the expectations of their peers; living in a large west-coast city where couples often wait until well into their thirties to get married and have children, Ellie is choosing to move toward these milestones more quickly than she might if she did not feel as though waiting would be unwise given her health considerations.

Thinking about RRSO in the future, Ellie shared her thoughts about why giving up her ovaries might be more difficult than giving up her breasts:

Part of me worries that I might get ovarian cancer earlier than typical, and I get nervous. But another part of me really doesn't want to get my ovaries out. I'm actually more worried about the ovarian stuff because I don't care about a fake set of boobs, but not having ovaries is a big deal to me because of the hormones.

It is clear that Ellie realizes that her risk of ovarian cancer in the near future is still relatively low, but that she needs to be aware of the risk moving forward. Ellie sees ovarian cancer risk reduction as a more complicated issue than breast cancer risk reduction because of the multilayered effects of RRSO (*i.e.*, hormonal changes that can lead to increased risk of heart disease, loss of bone density, sexual side effects). She felt relatively comfortable about her ovarian cancer risk in the near-term both because she was still quite young and because "I have been on the birth control pills for a long time, that was my part in protecting my ovaries for now¹." Planning an RRSO around age 40 she noted that "there's still a part of me that is worried that I won't do it soon enough ... And there's the other part of me that doesn't want to do it that soon because of the

¹ Oral contraceptives reduce the risk of developing ovarian cancer in both the sporadic and the hereditary setting. The longer the medication is taken, the greater the protective effect. The protective effect lasts for 20 to 30 years after use of oral contraceptives has ceased. (Collaborative Group on Epidemiological Studies of Ovarian Cancer, 2008)

shallow reasons.” Some women hesitate when they contemplate risk-reducing surgeries and worry that the negative effects may be very difficult to handle. This may sway some to postpone surgery, exposing them to longer periods of risk.

One of the most prominent ways in which Ellie’s story is unique is her focus on generativity, or creating something that will have a positive impact on others and will outlive oneself. Having decided to pursue RRBM, Ellie was in the process of putting together the necessary supports and talented collaborators to create a film documenting her surgery experience and filling what she saw as a gap in the information available to young *BRCA* mutation carriers. She described her thinking in making the decision to share her story:

If I do it and it’s just by myself and I don’t make a film, it’s just like my own little personal sad story that only I know about and my friends know about. But, if I did this on a larger scale where people could be inspired by it, and see that I came out happy and positive, and not feeling lost, and not feeling indecisive, but feeling really empowered, then it’s not just me doing my own little sad story. . . . the surgery will be more meaningful for me if people can share and learn from it. I’ve been so focused on it, and the really great thing about doing this so far is that it’s made me feel so much more normal about doing it. Like this isn’t such a tragedy; some people can benefit from this. I feel so much better about it that way.

Being involved in making a film had also provided an opportunity for Ellie and Mike to connect and work together. They were busy filming “confessionals” in which they captured discussions related to *BRCA* and surgery and the impact on their relationship for use in the film. Mike had also contributed his legal expertise to helping Ellie make sure that contracts and other legal issues were being used appropriately.

Melanie’s Story

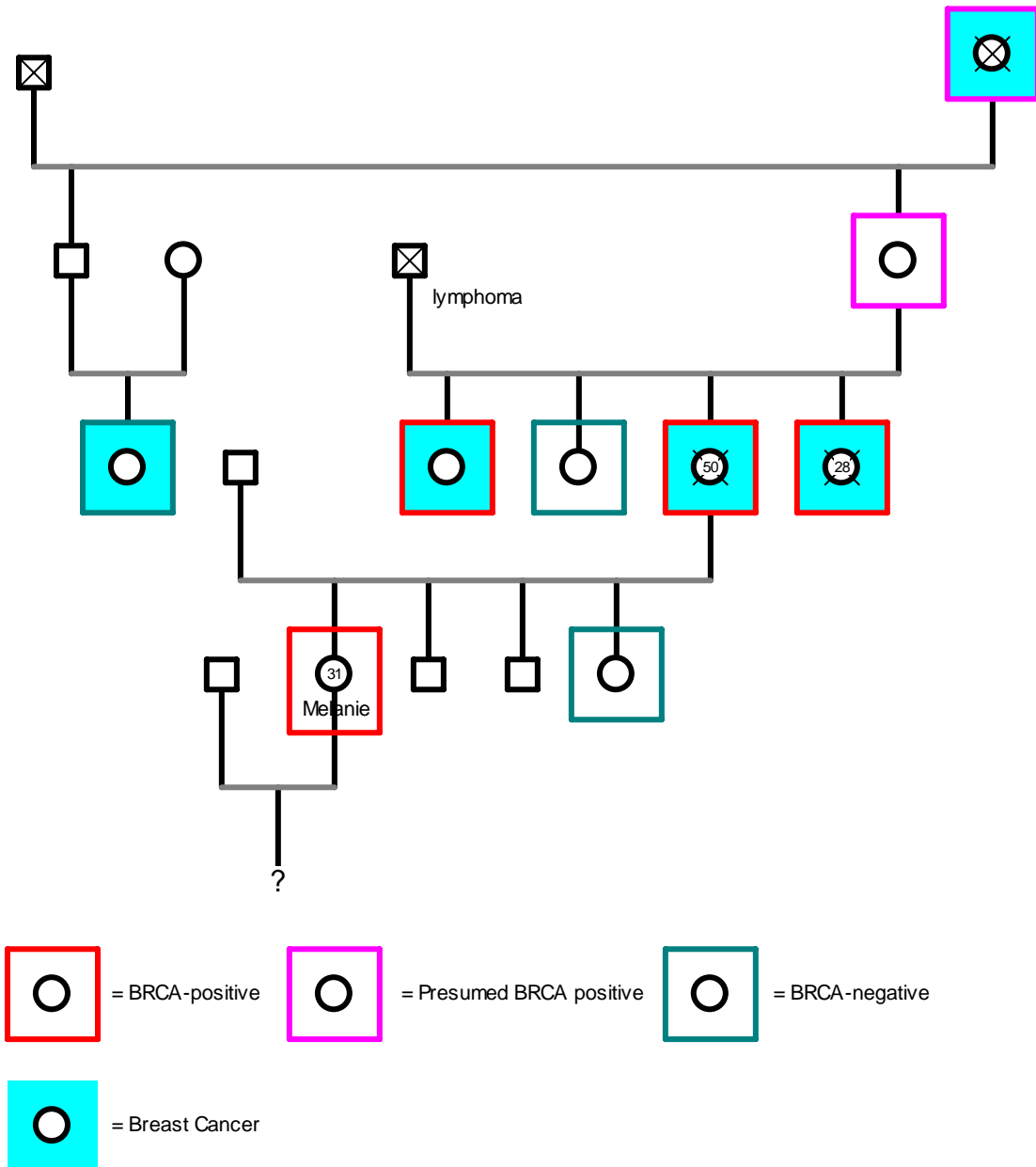
Melanie’s story provides a useful comparison to Ellie’s for several reasons. Specifically, both women inherited their *BRCA* mutations from their mothers, but

Melanie's understanding of how *BRCA* had impacted health outcomes in her family was much more comprehensive because her family did not have a legacy of death from unnatural causes like Ellie's. Both had mutation-positive mothers; Ellie's was still living and had never had cancer, and Melanie's had passed away after having breast cancer twice. Ellie's family narrative included individuals who had chosen risk-reducing surgery, while nobody in Melanie's family had yet done so. Ellie had learned about her *BRCA* mutation prior to meeting her partner, and Melanie was already in a serious relationship (which has since become permanent) when she learned that she was a carrier. Finally, the two were at slightly different developmental stages within the young adulthood phase of the life cycle, and were making different choices in managing their cancer risk.

As depicted in Figure 3, Melanie, 31, was the oldest of four children and was pregnant with her first baby when interviewed. Her family narrative included several examples of women who had been negatively impacted by breast cancer; the first occurred when Melanie was fourteen years old, and two of her mother's sisters (the oldest and the youngest) were diagnosed with breast cancer at about the same time. In fact, the younger aunt had gone to be checked by her physician precisely because the older aunt was in treatment for breast cancer, and an advanced, metastasized cancer was detected in the younger of the two sisters. Melanie's older aunt survived and was still living when Melanie was interviewed, but the younger aunt passed away at age 28. This created two stories of illness progression in the family: in one, breast cancer could be detected early and did not result in death; in the other, breast cancer struck early and aggressively and could be fatal.

Four years later, when Melanie was a freshman at a University on the other side of the country from where her family lived, she learned that her mother had been diagnosed with breast cancer. Melanie was informed of this after her mom's treatment had progressed and Melanie's parents felt confident that she would survive. This occurred in 1996, very shortly after the *BRCA1* and *BRCA2* mutations were identified,

Figure 3: Melanie's Cancer Genogram



and Melanie's mother made a passing comment about genetic testing; however, it was not pursued at that time. Five years later, when Melanie's mother was diagnosed with breast cancer for a second time, Melanie learned that her breast-cancer-surviving aunt had tested positive for a *BRCA* mutation. Melanie's mother was never tested because she passed away very shortly after her second diagnosis. Melanie had a strong sense that because there were so many cases of breast cancer in her family, it "wasn't just random." She worked with her mutation-positive aunt to construct a family history and found a University study that would provide genetic testing free of charge while she was in graduate school. In making the decision to get tested, Melanie weighed the opinions of both her aunt and her mother. Her aunt told her, "Yes, I've had cancer twice, and yes, I have this [mutation], but I'm still here and there are things you can do and the screening is getting better." Conversely, she remembered her mother "saying years ago, 'Okay, great, they have this test, but what do you do, do you run and get a mastectomy when you don't know if you need it?'" To Melanie, her mom's message clearly implied that she did not see the value in genetic testing for someone Melanie's age. Ultimately, Melanie decided that she wanted to know whether or not she carried the mutation, and learned that she was *BRCA* positive at age 26. She was able to make this choice independently and because she believed it was the right decision for her, even though she knew that her mother might have disapproved. As further evidence of this, Melanie informed only her boyfriend and her aunt that she was being tested, choosing not to share this information with her father or siblings until several months after receiving her results.

Melanie and Connor (who was her boyfriend when she was tested and her husband when she was interviewed) had to confront the realities of cancer together even

before Melanie was aware that she was *BRCA*-positive. The two met about six months after Melanie's mother had passed away, and he provided steady support for her as she coped with that significant loss. When she was deciding whether to get tested, he supported her by doing research about *BRCA* so that she could better understand what a positive mutation test would mean, and by "reassuring me that even if it was positive, it wasn't the end of the world." Since they learned that she is positive, Connor has supported Melanie by accompanying her to all of her screening appointments at the NIH (she was a BI participant) and helping her be diligent with her screening schedule. Having finished her four years in the BI study at age 29, Melanie recalled feeling nervous about how she would complete her screening and worrying about whether the cost of doing so would be covered by insurance. Her ability to feel secure about being protected from cancer was threatened by a change in access to medical care, magnified by the fact that Melanie had passed the age at which her aunt had died from breast cancer.

Another illustration of the closeness between Melanie and her partner was present in Connor's participation as an equal and engaged partner to Melanie in discussing how they would navigate family formation in light of her mutation status. Melanie recalled being frequently reminded by the healthcare provider with whom she interacted in the BI study that moving forward with family formation was urgent:

After we were married, we knew that prophylactic surgery, both the ovaries and the breasts, are definitely an option for me. Having kids is obviously affected by that, so we knew we needed to get pregnant. The people at NIH said, "What are you waiting for, hurry up and start having kids." So we knew that sooner was better than later.

These strong messages about the urgency of family formation are an example of how new and unexpected boundaries can emerge for mutation carriers. Melanie's plans for surgical

risk-reduction imposed a shortened timeline on her plans to have children, and this message was made crystal clear by the healthcare providers with whom she interacted. The potentially negative results of this were tempered by Melanie and Connor's easy agreement to have their children sooner than they might have otherwise done, to complete their family before Melanie reached age 35 and wanted to undergo RRSO.

Because she was pregnant when interviewed, issues of family formation were at the forefront of Melanie's thinking, and she discussed different ways that the presence of her mutation would arise as family formation progressed. Melanie and Connor felt relatively confident that they might want three children, and fitting all three of them in the four years before she turned 35 would require that they have children in close succession. Additionally, she would have to carefully time subsequent pregnancies to ensure proper breast and ovarian screenings could occur when she was neither pregnant nor nursing. Rather than being able to proceed naturally through the process of childbearing, Melanie was acutely aware of the extra steps she would have to take to ensure her ongoing health:

I was able to have a mammogram and MRI right before I got pregnant, which worked out really nicely. But after that, I went and met with doctors ... just to go over, "okay, I'm pregnant, what does this mean moving forward?" And the whole topic of breastfeeding came up, and what my plan is for that and what that means for screening for after that. What we've kind of decided is that I'm going to breastfeed ... for three months, because they told me that you have to stop breastfeeding for six months before you can have a screening, because I guess you'll light up [the MRI] because of the milk ducts and everything. So, they wouldn't be able to tell if there was something there. So, if we want to have kids close, I'll have to breastfeed, stop for six months, have a screening, and then try and get pregnant again. And because ... it didn't happen right away for us the first time, we need to give ourselves some time.

Melanie's story beautifully illustrates how the need to continue screening, combined with the same concerns with achieving pregnancy that are present in the general population, can feel confining for young *BRCA* mutation carriers.

Another important contrast between Ellie and Melanie is in their opinions about risk-reducing surgeries, specifically comparing RRBM to RRSO. While Ellie found it relatively easy to consider giving up her breasts but struggled to reconcile the loss of her ovaries, Melanie's feelings were just the opposite. She planned to undergo RRSO as soon as she was finished with childbearing,

...because if I'm done with kids I don't need them anymore, so that one is easier to me than mastectomy. And also, just in what I've read and heard, removing them can slightly reduce your risk of breast cancer². Also, not producing the estrogen, that sort of thing, that would be kind of a first step, and the less radical of the two.

Melanie's belief that mastectomy would be more difficult than oophorectomy helps to explain why having RRBM prior to pregnancy, as Ellie was planning to do, was not an attractive option. These differences in gut-level reactions to the two surgical procedures are likely an important component of risk-management decision-making for many mutation carriers, and have a significant impact on whether and when women are open to surgical risk-reduction. These visceral negative evaluations of the surgeries become roadblocks to effective risk management.

In the years since Melanie had learned she was *BRCA*-positive, she recalled feeling bad about imposing this ongoing burden on Connor. Their ability to handle this experience together is another demonstration of the powerful emotional bond they shared

I feel bad that I have it, but he just shoos that right away. ... now we're having a baby, I'll say, "I really need to get life insurance, I'm the one that something

² In fact, removing one's ovaries while still premenopausal actually reduces the risk of breast cancer substantially, i.e., by 50 to 60%. (Rebbeck, Kauff, & Domchek, 2009)

might happen to,” and he’s like, “stop, I don’t want to hear that, be quiet.” He just shoos it off. He’s not holding it against me, obviously. . . . We’re very, very close anyway, and this is just one thing that I’ve never felt like I couldn’t talk to him about or make it a part of our plan for our life or anything. I don’t have to be shy or worry about it by myself. It’s definitely something we, from the beginning, have taken on together. . . . everything I’ve gone through with it, it’s been *us* going through it.

This couple seems to have found the ideal way to share burden and convey support, which has allowed them to make choices with agency to feel that they will be successful at avoiding a cancer diagnosis.

Like Ellie, Melanie was generative, which she had achieved through the choices she made in her professional life. Having been part of a national service organization at the time she was tested, Melanie made a career move to a breast cancer research and advocacy non-profit shortly after she received test results. Asked how her job and her mutation status came together, she shared:

I know that I’m doing everything I can do to help. Really, [after] losing my mom, I don’t know what I’d be doing professionally if I didn’t have this to fight for. It’s given me a lot of focus, which is kind of a blessing in disguise. . . . it’s nice having a job that’s not just a job, that it’s something I really care about, and that helps in my actual work because I do a lot of work with volunteers, and they know that it’s not just a job for me to do, I’m in it for the same reasons they are. . . . When you find out [about your mutation], it’s like, “Oh, God, when is my time going to happen?” And with this, you’re not just sitting around waiting for it to happen. Whether it’s doing the walks and raising money, or people do a million different things, and I’m lucky enough that my nine-to-five is that sort of thing.

Melanie’s level of positive activity with regard to breast cancer and *BRCA* was one of the ways that she felt a sense of agency, and she was able to take this to the next level by positively impacting the lives of others as well as her own.

Agency, Constraint, & Communion

As Ellie and Melanie demonstrate, the process by which young female *BRCA1/2* mutation carriers make ongoing decisions about breast and ovarian cancer risk-

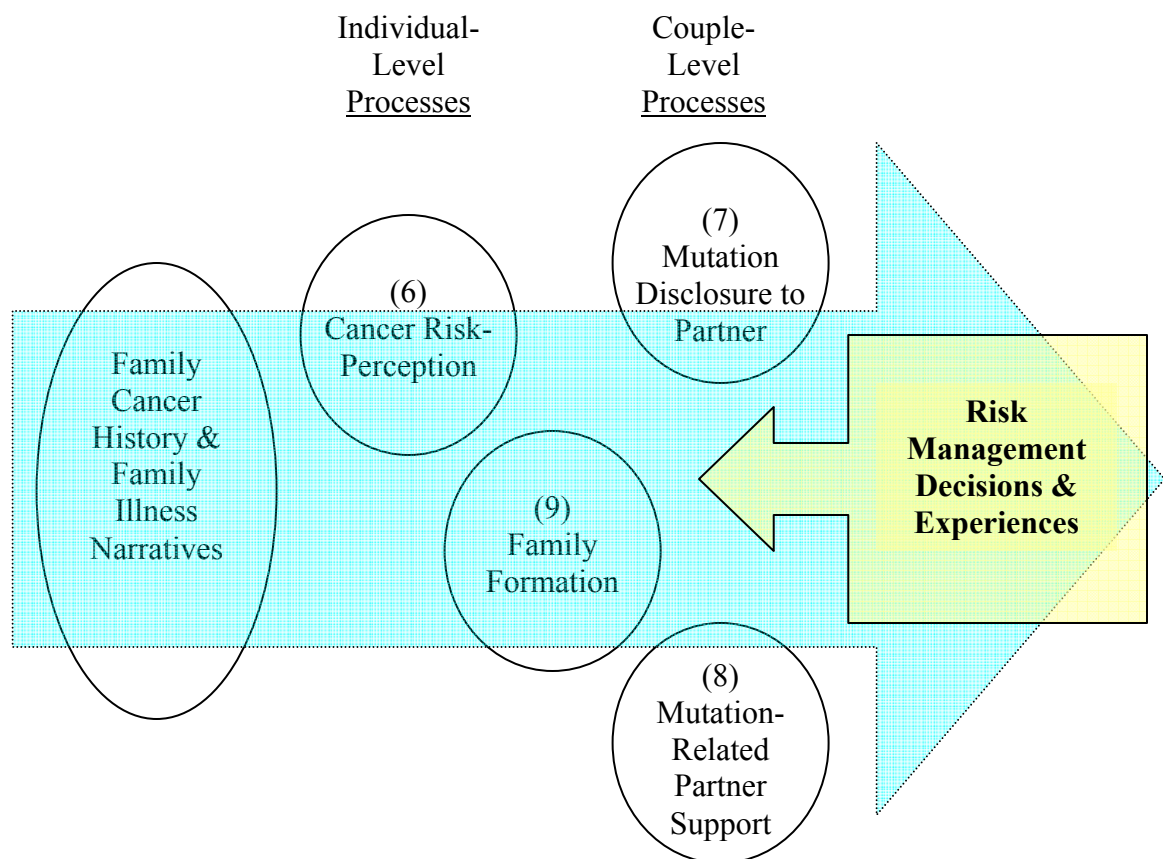
management is complex and multi-faceted, influenced by and influencing a number of other important life events and decisions. Understanding this process is facilitated by considering the concepts of agency, constraint, and communion, and how each of these come into play for mutation carriers. *Agency* describes a sense of self-efficacy (McDaniel et al., 2006). This may be understood as the sense that one has the capacity to act independently and make choices freely, or to enact change upon the world and one's environment. One might require a sense of agency in order to take action on a significant or difficult issue, or feel a sense of agency after action has successfully been taken. As previously stated, life course agency is drawn upon as individuals commit to a life path that will allow them to become who they wish to be and achieve what they wish to achieve. This type of agency describes "actions [that] occur with a broader sense of our futures involved, and these orientations are important for shaping individuals' adaptations to situations," (Hitlin & Elder, 2007, p. 182). Acting with agency is a mechanism by which young female mutation carriers adapt to their health status and achieve a sense of mastery over its stressors. *Constraint* is the opposing force – the sense that one is not fully at liberty to determine that path (or the steps that comprise it) for oneself, often because contextual or structural forces have imposed some limitations. Expectations on *BRCA* mutation carriers to be mothers, daughters, and partners convey a normative timing of progress through young adulthood, which may run counter to their preferences for health/wellbeing. On the other hand, the decisions they make about risk management can shape the timing of certain relationship family events. Finally, *communion* describes a sense of family or relationship cohesion (McDaniel et al., 2006). Communion is comprised of support, sharing of burden, and a sense of open communication.

Successfully counterbalancing feelings of constraint requires both agency and communion; relying too heavily on either (“*unmitigated agency*” or “*unmitigated communion*”) can produce negative psychosocial outcomes (Hegelson, 1994).

Components of the Model

Findings from the current study were used to develop a model to contribute to a theoretical understanding of how *BRCA1/2*-mutation-positive women move through the major individual- and couple-level domains and processes that comprise the experience

Figure 4: Basic Theoretical Model



Note. Numbers in parentheses indicate dissertation chapter in which element is discussed.

of young adulthood. This model helps to clarify how women make the mental and emotional journey from knowing about their mutation status and understanding family history, to making choices about risk-management. Note that this model is not intended to suggest that risk-management decisions are an endpoint to a staged process; rather, risk-management has been conceptualized in the current study as an important decision around which other model components pivot in important ways. As the data will demonstrate, risk management is certainly not an *endpoint*; rather, making a decision about risk-management is a significant event in the mutation-positive experience for young women and is shaped by other important elements of young adulthood. For many young mutation carriers, several risk-management decisions will be made over time as individual, couple, and family circumstances shift and as new knowledge and medical technologies become available. Further, risk-management is illustrated in an arrow that feeds back into the rest of the model, because women's decisions about and experiences with risk-management may shape their experiences with regard to cancer risk-perception, disclosure to partners, mutation-related partner support, and family formation in important ways.

Six core elements are present in the model: Family Cancer History & Family Illness Narratives, Breast and Ovarian Cancer Risk-Perception (discussed in Chapter Six), Mutation Disclosure to Partner (Chapter Seven), Mutation-Related Partner Support (Chapter Eight), Family Formation (Chapter Nine), and Risk-Management Decisions and Experiences. The next four chapters will discuss in detail the four identified individual and couple-level processes, and explain how they are related to both family cancer history/family illness narratives and risk management decisions and experiences.

Family cancer history & family illness narratives. Family cancer history describes the set of information about which mutation-positive individuals are aware regarding the legacy of *BRCA*-related (and perhaps -unrelated) cancer in their biological families. This is a relatively objective concept; it can be thought of as the “hard facts” surrounding how an individual understands that cancer has impacted her family. Knowledge about family history is influenced by many other characteristics of families and their histories. For example, as was illustrated in Ellie’s story, the availability of information about members of previous generations determines how fully family cancer history can be understood. Dynamics of communication on the side of one’s family from which the mutation was inherited are also influential; families that effectively share information about cancer diagnoses and treatment generally produce young female mutation carriers with more comprehensive understandings of their cancer history than families in which little information is shared (Werner-Lin & Gardner, 2009). In addition, women whose mutations moved down the family tree through successive males may not be as aware of their family cancer history as women whose mutations have been passed down through females, simply because men are far less likely to develop a *BRCA*-related cancer and therefore applicable cancer diagnoses occur in more distant relatives (*e.g.*, second cousins, great aunts). Becoming aware of cancer diagnoses in these distant relatives after learning about the presence of a mutation creates new family history for these women.

Family illness narratives are closely related to family cancer history. First applied in detail to the *BRCA*-positive individual by Werner-Lin and Gardner (2009), family illness narratives “are the stories families tell about their experiences with serious and

persistent illness,” (p. 201). They create a context for members of a family to give meaning to their illness experience. In the stories that they pass along through generations and adapt as new information emerges, families use illness narratives to give continuity and coherence to members’ understanding about “family myths or legacies that include beliefs about who gets sick and why, how families care for ill members, appropriate models of communication within the family and with outsiders, and illness outcomes,” (p. 201-202). Collectively, family cancer history and family illness narratives are represented in the model figure in one element. Family illness narratives are constantly evolving, and young mutation carriers can influence the family illness narratives through the choices they make about risk-management and in the way they approach the tasks of young adulthood that are related to their mutation status (*i.e.*, partnering, family formation).

In *BRCA*-positive families, histories of cancer and the illness narratives they produce are important components of women’s perceived cancer risk, and may produce either an optimistic or pessimistic ideal about what will happen at the individual level. If one’s family illness narrative says that “everybody gets cancer,” then one assumes that she, too, will eventually get cancer. If the family illness narrative is that cancer diagnoses are challenging but not life-ending, women may feel more empowered about their own ability to face a potential cancer diagnosis. Young women’s knowledge about cancer and the way it is understood by the family often bear heavily on family formation; for example, they may believe that they should not produce biological children because doing so would be akin to sentencing children to the same burdensome family legacy and cancer risk with which they are struggling. Family dynamics around cancer may also

powerfully influence one's decision-making about risk-management. For example, women may feel personally empowered by family history and illness narratives that include examples of mutation carriers who have successfully prevented cancer diagnoses by undergoing risk-reducing surgery; or they may feel limited by illness narratives purporting that cancer cannot be controlled, and therefore risk-reducing measures are a waste of energy. Finally, opportunities for sharing and joining with a partner can be created by sharing one's family cancer history and illness narratives with a partner and receiving instrumental and emotional support.

Cancer risk perception. Development of cancer risk perception is a unique individual process influenced not only by one's family history of cancer and family illness narratives, but by many biological and psychosocial variables as well. These include general levels of anxiety and depression, optimism, and recent cancer-related events in the individual experience (Price et al., 2007). It is "a key predictor of risk-reduction practices, health behaviors, and processing of cancer information," (Klein & Stefanek, 2007, p. 147). Data from the current project demonstrate that cancer risk perception can also profoundly shape the choices *BRCA*-positive women make with regard to the other elements of the proposed model. Cancer risk perception may shape decisions regarding disclosure of mutation to partners in couple relationships. A carrier who believes her risk to be very high may fear that a partner who learns about her mutation will choose not to continue a relationship upon learning about the mutation, and so might feel anxious or even scared about disclosure. Developing a sense of control around cancer risk can enable carriers to manage partner disclosure effectively and present information to their partners in a way that promotes connection.

Decisions about family formation may also be affected when very high cancer risk perception produces a belief that one's available time for childbearing, the options that one may utilize to become pregnant (*i.e.*, assisted reproduction), or one's capacity to breastfeed in a way that is experienced as positive and free of risk, are limited. Women who are able to successfully manage their cancer risk anxiety and share their sense of peace with other members of their families can positively influence the family illness narrative. Finally, cancer risk-perception powerfully shapes decision-making about risk management; women with high cancer risk-perception often take more aggressive action to mitigate their risk (*e.g.*, by having RRBM or RRSO at a young age) than women with lower perceived risk, who may opt instead to have surgery later or to rely on screening indefinitely. In either case, carriers' feelings that they are taking or are able to take sufficient/appropriate actions to manage their risk determine whether agency or constraint is experienced.

Mutation disclosure to partner. Disclosure of positive mutation status to one's partner, the first of two couple-level processes in the model, occurs differently depending on the context of a given couple relationship, as will be illustrated in chapter six. Its capacity to impact other elements of the model is notable. Women's choices about risk management can dramatically shape the disclosure process when the results of management decisions are obvious to new partners. Women may feel restricted in disclosing the mutation, as it impacts their partner's willingness to have children together. Many participants, especially those who were single, worried that a partner would find the potential cancer risk to future children intolerable and choose not to participate in family formation; often, this was accompanied by worry about the partners' willingness

to continue the relationship at all. For partnered participants, mutation disclosure in some cases had a constraining effect on family formation because it expanded to the partner carriers' own beliefs about how family formation ideals might seem threatened and options might seem limited.

Mutation disclosure is also an important actor upon mutation-related partner support. Disclosing one's mutation to a partner is the only way to establish a pathway for effective partner support around *BRCA*-related issues; therefore, women who wish their partners to be involved in ongoing challenges and decision-making related to their *BRCA*-positive status must achieve mutation disclosure first. The ability of a partner to provide support, or the carrier's belief that support will be available from the partner, often facilitates or eases disclosure. Couples who are able to successfully share information about and request/provide support around *BRCA*-related issues experience increased closeness.

Finally, an important relationship exists between mutation disclosure and risk management decisions. Many participants described how their decisions regarding when to inform their partners about their mutations were shaped either by an approaching risk management event (*e.g.*, a significant screening visit or a surgery) or by the fact that they had already had a risk-reducing surgery and felt compelled to tell their partners about it before it became obvious. Women who are not actively managing their risk, or managing it in a way that is not noticeable to their partners, have greater flexibility and control in deciding how and when to share mutation information with partners in couple relationships.

Mutation-related partner support. Discussed in detail in chapter eight, mutation-related partner support is a couple-level process and may be defined as the ability of a partner to help a mutation carrier cope (both instrumentally and emotionally) with the challenges that are often part of the *BRCA*-positive experience for young women. In addition to its relationship to mutation disclosure described previously, partner support is common in situations where partners play important roles in decision-making about family formation and risk-management. Opportunities for closeness may occur when a supportive partner helps a carrier cope with her anxieties related to her high perceived risk of cancer, or when anxiety and decision-making with regard to family formation is shared. Finally, a partner's ability to provide appropriate support and input about risk-management decision-making provides another opportunity for closeness and sharing of burden.

Family formation. Family formation is considered both an individual- and couple-level process, hence its placement between these two categories in the model. Dynamics surrounding family formation can impact several other elements of the model, and the ways in which this occurs will be illustrated in chapter nine. Engaging in biological childbearing is considered by many women to be inherently risky because of their fear (often based in anecdotal experience) that the hormonal exposure that occurs during pregnancy and breastfeeding can contribute to carcinogenesis. When family formation is achieved successfully and does not result in a pregnancy- or breastfeeding-related cancer diagnosis, it may (temporarily) mitigate cancer risk perception. When mutation carriers are able to successfully synchronize family formation desires with the perceived need to manage risk through surveillance, chemo-prevention, and/or risk-

reducing surgery so that all needs are met, they may gain a great sense of satisfaction and personal control. The opposite may occur when goals related to family formation and those related to risk management cannot be reconciled, and sacrifices to one's powerful desires must be made. All of this occurs in concert with one's partner; among single participants, the needs and desires of future partners are often considered as part of women's contemplation of future family formation.

Risk management decisions. Decision-making about risk management is largely the product of the other five elements of the model, as described previously; however, its capacity to influence those model components cannot be disregarded. Risk management decisions that bring a sense of relief and lower cancer risk perception often create a sense of competence and mastery. When women perceive that their risk management decisions – or the risk management options available to them at a given point in their lives – are insufficient to protect them from cancer, they may feel limited and powerless. Risk management decisions may also shape the mutation disclosure process in that when a single woman is able to confidently explain her risk management decision-making process to a new partner, she may feel strong and in control of both her cancer risk and the way she is perceived by her partner. Conversely, risk-management decisions can inhibit a carrier's ability to disclose positive mutation status to a partner in the preferred way or at the preferred time.

Chapter Conclusion

Women who test positive for *BRCA* during young adulthood face complex and ongoing challenges in reconciling their high-risk status with the normative tasks of this stage of the life course. Each of the major individual- and couple-level processes outlined

here are intricately tied to the others, producing a confusing and tangled web of choices for young carriers. Women make these decisions both independently and with their partners and families. When this is done successfully, the process creates unique paths that ultimately allow each individual to feel as though she has maintained control and minimized sacrifices to previously held notions of how life would proceed. The proposed theoretical model will be used in the subsequent four chapters to illustrate how each of the four individual- and couple-level processes identified are related to other elements of the model.

CHAPTER 6: RESULTS—RISK PERCEPTION

“I was sure I had two ticking time bombs strapped to my chest.” - Valerie

The quantitative risk of cancer among carriers of *BRCA1* and *BRCA2* is generally well-understood, and can be communicated clearly to potential and known mutation carriers during the genetic risk assessment, education and testing process (van Oostrom & Tibben, 2004). Despite this, the extent to which individual mutation carriers feel vulnerable to cancer is a complex dynamic influenced by information from medical professionals, family and family history, and peers; specific perceived vulnerabilities based on age, mutation type, previous cancer scares, and specific cancer type of concern; and other characteristics unique to each individual. These data indicate that decision-making about risk-reduction is very much influenced by one’s perception of cancer risk, whereby women who feel more vulnerable to breast and ovarian cancer have a different experience of themselves as mutation carriers and are more apt to take early, aggressive steps to mitigate their risk (often by electing to undergo risk-reducing mastectomy or oophorectomy), whereas women who feel less anxious about their risk may be apt to rely on screening for a longer period of time or to believe that surgical risk-reduction is not necessary. For women who have already completed one or both risk-reducing surgeries, there is often a palpable reduction in their perceived risk. However, data indicate that the relationship between perceived risk and risk-management decisions is not simply direct: it involves the other elements of the proposed model as well. Effectively utilizing cancer risk perception to inform and shape decision-making and communication about various tasks of young adulthood is a common experience for these women, and is an important piece of how they take control of their lives as mutation carriers.

Gathering Information about Cancer Risk

Participants demonstrated having gathered information from a variety of sources and synthesizing that information to arrive at individual cancer risk perception. At the time of her interview, Valerie, a genetics researcher, had been aware of her mutation for nearly four years and had recently undergone a risk-reducing mastectomy. She recalled what she felt about her cancer risk shortly after receiving the news that she carried a *BRCA* mutation, and nicely illustrated the synthesis of several different sources of information in making decisions about her own risk management:

I knew all along that I needed to be keeping an eye on my breasts, certainly. As far as I know, there's no ovarian cancer in my family, and we're *BRCA2*, so that's not necessarily surprising. And also, if you [look at] where my mutation is [physically within the *BRCA2* gene], based on the literature, it's in the sort of lower risk spectrum for ovarian [cancer]. But, at the same time, ovarian cancer scares the hell out of me, so I'm not taking any chances. [My doctor and I] basically made the agreement that if anything looked fishy, they were coming out. So my ovaries are basically on notice.

Valerie, 29

Because of her education and profession, Valerie was uniquely positioned to understand her cancer risk. For other *BRCA* mutation carriers, risk information may come primarily from one source, or may be synthesized from several. The way that individuals come to understand that information and the importance they place on any one source appears to be an important determinant of how they approach decision making, and whether they are able to feel a sense of control with regard to managing their risk.

Information from medical professionals. For most *BRCA* mutation carriers, the first substantial interaction with a medical professional in which they receive information about their risk of breast and ovarian cancers occurs in the context of genetic counseling, usually prior to submitting a blood sample for mutation testing. Sometimes, this occurs as part of the same appointment in which they receive their genetic test results. This

communication can be an intense experience for many women, especially if cancer risk is not something they have previously contemplated or thought of as personally relevant. Conversely, women whose backgrounds provide context for the information received from a genetic counselor may find this experience less stressful. Wanda had a doctoral degree in molecular biology, and therefore reached a very comprehensive understanding of the biological phenomenon being explained by her genetic counselor. She recalled the genetic counseling appointment during which she received her mutation test results:

I've always been one of those people who likes to know everything about everything. Sometimes they say too much knowledge isn't necessarily a good thing, but in my case I felt well-informed, I think more so than somebody else who didn't have a science background and had a mutation-positive test. Although of course genetic counselors do their best to try to explain things, patients are hearing, "I'm going to get cancer. I'm going to die." With me, I was like, "Okay. Well, this definitely increases my risk, but there are things I can do, and now I know what I have to do in the future to be better prepared."

Wanda, 31

Wanda's scientific background and ability to understand what was being communicated by the genetic counselor allowed her to feel empowered by the information. In contrast, Elaine remembered being very anxious upon learning that she carried a *BRCA* mutation:

I just felt very anxious I guess. I've never felt that way before, but – I'm a [mental health professional] – I thought, "Oh my gosh, *this* is anxiety," [emphasis added]. I felt kind of tingly and anxious, and [the genetic counselor] was going to give me some information and I just told him, "I have to go. I'll call you back. I can't take any information right now."

Elaine, 34

Similarly, Pauline remembered:

I was in shock and not processing anything for a couple of minutes. [The doctor] came in and said some things and it was sort of like the Charlie Brown teacher voice, like "Wa-wa-waaaw." And occasionally I was able to follow but he said something about high-risk women and that really sort of cut through the fog for me and I was like, "Oh, wait, he's talking about me. I'm high-risk."

Pauline, 30

Elaine and Pauline both felt paralyzed upon first learning that they were *BRCA*-positive. In part, this may be the product of their family histories. Elaine's mother had died of ovarian cancer several years before Elaine's genetic testing, and she lost her grandmother to breast cancer several years before that, so her family illness narrative was one in which having breast or ovarian cancer was a painful and deadly experience. For Pauline, whose *BRCA* mutation was inherited from her father, the effects of *BRCA* were a mystery because no family members with whom she had close relationships had been diagnosed. She felt an acute lack of insight about how the information she was hearing would play out in her own life. These data suggest that women's capacity to hear and assimilate information from providers immediately after receiving positive mutation results may be low if they experience a significant amount of shock or anxiety, or if they are caught up in their family illness narratives. It may be helpful for physicians and genetic counselors who deliver results to women to find out the patient's preference for when to discuss cancer risk, next steps, and risk-reduction options, so that patients can receive the maximum benefit from these conversations.

Once they assimilated the information received from genetic counselors and began regular interactions with physicians and others involved in their ongoing care, participants began to receive frequent messages about their health and cancer risk from the doctors, nurses, and technicians with whom they interact. One frequent observation among participants was that once they were aware that they carried a *BRCA* mutation, it became very easy for them to access needed screening. For many, this was a notably different experience from what had been occurring when they attempted to access

screening based on family history alone (even when they knew that a *BRCA* mutation was present in their families). For example, Rose remembered that

Once I tested positive, my ability to get in to see a doctor, my ability to get scheduled for an MRI just opened up immediately. I don't know if this is true nationwide, but in [major city] if you try to get a mammogram it's like an eight or nine month wait. It's crazy. I couldn't believe that. You've got to be kidding! But once I tested positive for *BRCA* they were like, "OK, you come in in two days."

Rose, 30

The implication of this increased responsiveness from medical professionals is that they acknowledge a significantly elevated cancer risk in these patients, which is often interpreted by mutation-positive women as a confirmation that they are in a precarious health situation. In fact, for many women, the simple fact that they were seeing medical specialists for their care contributed to their perceiving themselves as being at greater risk of cancer; and needing care beyond what a general obstetrician/gynecologist could provide felt significant to many participants.

In their interactions with medical professionals, many women lamented the fact that information they received from different providers was sometimes confusing and/or inconsistent, creating a challenge for them to select the appropriate course of action.

These confusing interactions may begin even before women are aware that they carry a *BRCA* mutation. For example, Leigh recalled a time before she had been tested when she asked her physician for advice about screening as a member of a family with a strong history of breast cancer:

I sort of assumed I had a very high risk of breast cancer but I didn't really know how high. A few years before our family did the genetic testing, I did ask. I told a gynecologist a little bit of my family history and asked if I should be receiving any type of screening now and she thought I was okay to wait until I was 40 to start doing mammograms. So I sort of pushed it out of my mind.

Leigh, 35

This interaction (which took place in about 2003) is noteworthy because, given the current recommendations for breast screening in young women from high-risk families, it is almost unthinkable for Leigh's physician to recommend that she wait until age 40 to begin screening. Given current guidelines, Leigh would be advised to have mammograms, MRIs, and clinical breast examinations every 6 to 12 months, beginning at age 25. The recommendation that Leigh received likely led her to feel that her options for risk-management were constrained, and to feel more anxious about her cancer risk when she did learn of her mutation, than she would have experienced had she been properly screened earlier.

Even for known mutation carriers, there can be barriers to getting desired information from medical professionals. Before she enrolled in the BI study, Monique was receiving all of her mutation-related medical care from local physicians. Having been tested at age 20, she was on the very young end of the range of patients typically seen by physicians at high-risk breast imaging centers. Monique decided to test at an early age because her maternal aunt was diagnosed with breast cancer at age 20 and died at 22; Monique's mother also died from breast cancer at 39 after being diagnosed at 37. Monique's much older sister had already had an RRB, and Monique was interested in learning whether that might also be an appropriate course of action for her:

I think the worst part is that they all were like, "you're so young, you should just get check-ups." I felt like I wanted more of an answer, like, what should I do? They didn't really want me to have the surgery yet, like how my sister had surgery, just for prevention. They thought I was too young until I had a baby, so they were talking to me about what I should do before and after I have kids.

Monique, 26

Although Monique's physicians were being honest with her about the recommended course of action for a mutation carrier her age, it is easy to see why Monique was

frustrated and felt constrained by this experience. Knowing that she was already past the age at which her aunt had been diagnosed, she was understandably anxious about her short-term cancer risk, perhaps more so than other mutation carriers who do not have a family history that includes a cancer diagnosis in someone as young as Monique's aunt. The result of her physicians' recommendations was that she continued to live in a state of high anxiety about her cancer risk, because others did not share her sense of urgency regarding the need to take action to mitigate her risk. Practically, there was no reason why Monique could not have had an RRBM if she chose to do so, with full awareness of the implications of that decision; therefore, her doctors' responses to her request for information about her options for risk-reduction seem somewhat patriarchal. The assumption by the medical community that she is "too young" to make such a decision, that she *should* keep her breasts until after she has children, effectively constrains her choices and eliminates that option for her. Clearly, a family can be created in the absence of one's natural breasts, as has been the experience of several other participants. Increased openness about these "alternative" progressions through the risk-reduction dynamic on the part of the medical professionals who care for women in this population would give *BRCA*-positive women greater freedom and more options to manage and/or reduce their risk in the way that seems most beneficial for them personally, and help them to achieve a sense of agency in making risk-management decisions that are congruent with their chosen path through the life course.

Given the enormous amount of information given to patients in medical settings where young *BRCA* mutation carriers receive their care, it is obvious that many women remember only certain pieces of what they hear from members of their healthcare teams.

When a patient does remember something, it is sometimes because it was phrased in such a way that it makes intuitive sense, strikes a chord, or simplifies an issue in a unique way. For example, Sadie's genetic counselor used a gambling metaphor when discussing the odds that Sadie would develop cancer in her lifetime:

Mostly what I remember is the increased risk for breast cancer, that if you were gambling in Vegas with 80% odds, that if someone said you have an 80% chance of winning the jackpot, you would probably play, thinking, "Hey, that's pretty good." So I always remember that analogy and whenever I'm doubting what I'm doing, I sort of think, "Okay, yes. If I were gambling with those odds, I'm pretty much at, "It's a sure thing."

Sadie, 33

Sadie remembered this comparison often and had even communicated it to other mutation carriers in discussions with them about how to manage cancer risk. She highlighted how this mental checkpoint contributed to her confidence in moving forward with her decision to undergo RRSO at the age of 32, because she was able to return to this metaphor and feel confident that the choice she was making was the right one. Other participants also reported holding tightly to risk-related information given to them by trusted medical professionals. Rachel remembered that her very busy doctor

took the time to really sit there and explain it to us. I still have the piece of paper that she wrote, that I had an 87% chance of having breast cancer and that I had a 40% chance of ovarian cancer and I could do a hysterectomy or I could do an oophorectomy and she literally sat with us in her office and explained everything.

Rachel, 33

Ultimately, many women put a great deal of confidence in the opinion of their doctors and make decisions about risk management based largely on those recommendations.

Asked about messages she received from physicians, Maelie recalled:

There were some conflicting messages, but what I held onto was what's the most certain, what's the best course of action. The majority of people, or articles, or anybody you talk to is if you have the surgery, you have a better guarantee. Not

that it's a guarantee at all, they list all the things they can do, but surveillance isn't always the best option. I think everyone is pretty unanimous on that.

Maelie, 33

Information from family members & family history. One of the primary sources of information from which women draw to arrive at their own understanding of their cancer risk and to make decisions about risk management is the experience of other family members. Like Ellie and Melanie in Chapter 5, participants reported having considered at what age, site, and stage relatives (usually either known or presumed mutation carriers) had been diagnosed with cancer as they considered whether and when they might be at risk themselves, as well as looking to the surgical and reconstruction experiences of their loved ones to help anticipate what their own experience might be like. When women examine their own mutation-positive experience through the lens of family history, feelings of either empowerment or hopelessness may result. Which of these occurs depends partly on what events have occurred in the family and how well individuals are able to imagine having an outcome different from those of family members. For example, Shannon, 31, began to think of herself as a person at high risk of breast cancer at 15, when her mother was diagnosed; after she was tested for a *BRCA* mutation years later, her sense of vulnerability to cancer did not really change, because she had “always felt pretty vulnerable.” Leigh, 35, reflected on both her aunt's and her mother's breast cancer and her mother's difficult experience with surgery and reconstruction in deciding whether and when to have surgery, and which of the two surgeries to pursue. She recalled, “I guess [my mother] had a very hard time with her reconstruction. So it made me want to do it while I'm younger because I figured maybe I would have an easier time.”

Growing up, Dawn was aware that several women in her father's family had died of breast cancer before she was born. When Dawn was three, her paternal aunt was diagnosed with breast cancer at age 27 and died at age 30; shortly thereafter, her paternal grandmother also died from the disease. Asked when she started to think of herself as a person at elevated risk of breast or ovarian cancer, she replied:

Honestly, probably really young. I've always known. I remember being three and five and six and knowing about breast cancer. Even in junior high and high school, I would talk to my gynecologist and ask, "Do I need to start pretty young getting all these tests, the [mammograms] and stuff?" And they all told me, "Honey, it's on your dad's side, you don't need to worry." I never really liked that answer because I always had a feeling that it was something that could happen to me, especially with everyone in my family being so young. I was so determined that they were all not telling me the truth. I always felt like, "Why? My dad passed so many genes down to me, why would he not pass this?"³

Dawn, 27

Though Dawn was clearly attempting to be proactive about her breast and ovarian health, she was inhibited by the lack of knowledge possessed by her healthcare providers. This was largely a product of the era in which these conversations occurred, before *BRCA1* and *BRCA2* and the way they move through families was fully understood.

Dawn and Pauline are similar in that they both inherited their *BRCA* mutations from their fathers, neither of whom have developed cancer. In contrast to Dawn's first-hand knowledge of family cancers, Pauline was only vaguely aware of a few cases of cancer in older relatives before she learned that there was a *BRCA* mutation in a distant branch of her family tree.

It's really scary, though, looking at the [pedigree], because I don't have the experience like a lot of women do who have these mutations, of watching their moms or their aunts or their grandmother go through this. . . .and then basing their recommendations on when to do things based on when their mom got sick, when

³ Dawn was exactly correct: *BRCA1* and *BRCA2* genes are inherited in an autosomal dominant fashion. One average, half the offspring of a mutation carrier will inherit the mutation, and the abnormal gene can be passed down equally well by both males and females.

their grandmother got sick. I don't have that, but I look at my chart and it looks like everyone's getting sick sooner and sooner. I'm seeing people who weren't diagnosed with breast cancer until they were eighty-five and then two generations later people are getting breast cancer at thirty. I know it's not a perfect science but I have two cousins who got sick at the age that I'm at right now. So that's what I think about because I don't have that experience of having watched my mom or grandmother go through it.

Pauline went on to elucidate how, as discussed at the start of this chapter, the absence of first-hand experience of cancer in her family left her at something of a loss when trying to conceptualize and make decisions about her own risk:

Right now, I have absolutely no idea what cancer looks like. I have no idea what cancer feels like. How it affects people, who it takes, who it spares. I've got nothing. I've got just this big fat number that the geneticist flopped in front of me when she gave me my status that says 87%. And so, I know it's irrational to think that I have an 87% chance today, but that's sort of how I feel to a certain extent because I have no sense how else to gauge it.

Pauline, 30

Other participants discussed unique circumstances in which their pervasive family histories of cancer still left them unsure about their own risk. Prior to being tested for a *BRCA* mutation herself, Rachel was aware that her family had a higher-than-usual incidence of cancer, but did not feel that she was at risk herself. She remembered:

We didn't really understand. Everybody on the street [we grew up on] wound up with some form of cancer. Mom and dad used to joke that it was something in the environment where they lived for so many years. That there must have been something in the soil, something in the water, and it literally became a joke. "We almost glow at night," that kind of thing. So, we kept our faith but we also kept our sense of humor. It was still looked at from the standpoint that it must have had something to do with that neighborhood, we were quick to write it off. And in 1998 ... information [about *BRCA*] ... definitely wasn't ... making its way to us. Everyone said to me, "if your mom's already had [cancer] twice, you better be careful. You better start your surveillance early." ... But that's as far as it went at that point.

Rachel, 33

Rachel's experience seems to be similar to Dawn's, in that lack of knowledge about *BRCA* at a given point in time prevented her from fully understanding or effectively managing cancer risk.

After women have assimilated perceived risk based on their family histories, what comes next is often a set of assumptions about what the experience of cancer would be like should they someday be diagnosed. Often, these cancer narratives, which can be either personal or shared, are based on what they witnessed in the breast and/or ovarian cancer treatment given to their mothers, aunts and grandmothers in years past, when screening for cancer was not as effective and therefore cancers were more likely to be detected at an advanced stage. Further, fears may be based on experiences with cancer treatment that occurred when medical technology was less advanced and treatments more physically and emotionally taxing than they are today. In the aggregate, these experiences may exacerbate fears about cancer that are not in line with what current medical practice can provide in terms of manageable treatment. Reina provided a good example of this when she related what it was like to watch her mom go through two difficult breast cancer diagnoses and treatment:

I really had to work through it, just because my mom went through certain experiences doesn't necessarily mean that I'm going to go through those same experiences. Whenever I think about breast cancer, I automatically associate everything that my mom went through and relate that to, well, God forbid if something were to happen to me, that's the type of situation I would be placed in, which is not true. But I really had to work through that because the treatment part of it is so physically wearing. It's hard. There's always that fear of death, but I'd say more so the process of getting into remission, going through the treatments and the surgeries and all that. That weighs on my mind more heavily.

Reina, 29

Reina's level of anxiety about developing cancer seemed quite high compared with many of the other women in the current study. It seems plausible to speculate that more

accurate knowledge about the experience of women whose breast cancers are caught early and managed effectively might have helped reduce her anxiety to a more manageable level and created a greater sense of agency.

Information from peers. Discussing her cancer risk with peers might also provide important support and contribute to a special kind of sharing for Reina and others like her, and drawing support and information from peers seems to be a primary method of maintaining connections with other mutation carriers for young *BRCA*-positive women. Because of the low prevalence of *BRCA* in the general population, it is common for mutation carriers not to know any other carriers personally, outside of their families. Therefore, they often turn to the internet and other media through which young mutation carriers have shared their experiences. Many women mentioned their experience with FORCE as having been very positive and as having helped them find information that was lacking elsewhere. Leigh benefited from “women posting on all their experiences, and it’s all specific to women who have the genetic cancer risk, so that played a big part in terms of gathering information and dealing with everything.” Though this was clearly experienced by Leigh as positive and created communion with these new contacts, it should be noted that a possible drawback of participation in a community such as FORCE is that women may become saturated with negative or troubling stories posted by others looking for support, since those stories may be more likely to be submitted than stories from women who have not experienced significant problems in dealing with their hereditary cancer risk. This was the case for Sadie, who turned to the FORCE message boards for support and information soon after she learned that she was *BRCA*-positive:

When I first went to the FORCE website a couple years ago, it totally freaked me

out. Their message board was all about having surgeries and complications that people were having and whether or not they should do a hormone replacement therapy and what type of mastectomy they were doing. They were speaking all of this lingo that was so foreign to me and there are people on there who were well past having it done but then there are people who are about to have it done and were scared out of their minds, and there were people who had complications right after surgeries and were having trouble. I had like an anxiety attack. I was looking at the message boards and I was just like, “Oh my God! This is so not helping for me to be looking at this stuff!”

Sadie, 33

Although Sadie was open to information from peers when she first viewed the FORCE message boards, the bias toward sharing negative experiences – which makes sense, given that those women are also on the message boards in search of support – created an online environment that overwhelmed Sadie and did not provide what she was seeking at that time. Having real-life access to other individuals in similar situations (*i.e.*, at a similar developmental point in the natural history of their *BRCA*-related experience) is a potential unmet need for young mutation carriers. This would allow them to share their full range of concerns, knowledge, and experiences (both positive and negative) with one another in a supportive and productive manner. Organizations such as FORCE and Bright Pink do a good job of meeting this need through annual conferences and local support groups, but it seems clear that some women have information and support needs beyond what is being provided, or simply are not finding the resources that work best for them.

Cancer experiences of others with whom individuals have personal contact but who are not members of one’s family can also be a source of information about cancer risk. Leigh learned that she carried a *BRCA* mutation shortly after her mother tested positive. Leigh was then the mother of three small children: she experienced high levels of anxiety during that time, and her experience was impacted by the cancer diagnosis and death of the father of one of her son’s close friends:

When I first found out that I had the mutation and that I was at risk now, I was very, very scared of developing cancer in the near term. I would still be busy during the day and to do whatever I have to do, but at night when I was lying down to put my littlest to sleep I would just be terrified of dying and being separated from them. I had anxiety dreams sometimes, like one where I had been diagnosed with cancer and had only a few months to live. It probably didn't help that one of my oldest son's friends, his father died of lung cancer, only six months between diagnosis and death. And he died a couple months before my mom found out that she was a mutation carrier. So I think that was another factor that made it seem very real that a parent could just easily die and leave their children behind. That idea really terrified me for a while.

Leigh, 35

Although the cancer in the father of her son's friend was likely not genetic and was almost certainly unrelated to *BRCA*, the experience of watching a family lose a parent of young children had a powerful effect on Leigh and her own thinking about her health and her family.

Another vehicle through which some women have found support from peers is the book *Pretty is What Changes* (Queller, 2008). Queller's book recounts her experience of her mother's cancer diagnoses, treatments, and eventual passing; as well as Queller's own process of learning about her mutation and deciding how to manage her risk. Rylan mentioned this book in discussing her view of her 87% lifetime risk of breast cancer and relating how she came to the decision to have an RRBm:

I had just read Jessica Queller's book *Pretty is What Changes*, and I think that helped me make the decision. I just decided I was not worrying, but waiting to get cancer. ... I have up to an 87% chance, and if I don't get it, I might win the lottery, too, but chances are I won't. And chances are I won't *not* get cancer. To me it sort of became more a matter of *when* am I going to get it. I mean, I realize I have a chance of not getting it ever, but the risk statistically was so high that it made sense to try to avoid the risk.

Rylan, 34

For Rylan, it was almost as if a switch had been flipped: one day, she did not see her 87% lifetime risk of breast cancer as a threat worthy of her attention, and the next day, she had

made the decision to have surgery. Reading the detailed experience of another *BRCA* mutation carrier close to her age was a powerful catalyst for Rylan to make a confident decision about her own risk management.

Sophie was a participant in the BI study whose family cancer experience consisted of three fatal cases of ovarian cancer on her father's side; nobody in her family had ever been diagnosed with breast cancer. Accordingly, Sophie's perceived risk of ovarian cancer was quite high, while her perceived risk of breast cancer was relatively low. Sophie recalled how meeting other *BRCA*-positive women at the NIH was a powerful experience that became synthesized with other sources of information about risk, namely her family history and the documentary film *In the Family* (Rudnick, 2008). That film, with a young *BRCA*-positive woman named Joanna as its focus, has also become a source of support for many women, who have begun to look to Joanna as a peer and referent. For Sophie, these sources of information combined to prompt reconsideration about the appropriate strategy for risk management:

My dad made my middle sister and I sit down and watch *In the Family* and it was pretty eye-opening and depressing. It's tough to watch. And then when I actually was at NIH this last time, I met a girl who is part of the study, and she was one of three sisters [like Sophie and one of the families in the film], and all three had the mutation. So that was kind of interesting to me. Even now I don't know if a mastectomy would be something I would go forward with, but now it's something that I would possibly consider. I think that is really extreme for me at least. I don't knock anybody who does it, but for me, I think that would be the extreme and that movie just kind of showed that it's something that definitely needs to be considered. A mastectomy really is not completely on the table, but it's in the back of my head. But the oophorectomy is a done deal; I already know. I may be in denial about breast cancer because neither of my aunts had breast cancer.

Sophie, 31

Seeing someone like herself on screen and witnessing how Joanna handled the same issues that Sophie was facing allowed her to see her situation from a new perspective and

begin to open her mind to ideas she had not previously considered seriously. Where she had previously felt trapped by fear and family narratives, Sophie felt empowered by this peer support and information.

Specific Vulnerabilities

Breast versus ovarian cancer risk. While the general term “cancer” certainly elicits feelings of anxiety in most people, women with *BRCA* mutations must struggle to balance their perceived risk of *breast* cancer and their perceived risk of *ovarian* cancer – these risks are not the same, and the health threats presented by the two cancers are also very different. Ruby recalled how learning that she carried a *BRCA* mutation brought with it a good deal of information about breast cancer risk, but left her hungry for more information about the risk of ovarian cancer. Unfortunately, this information has not been easy for Ruby to come by because Ruby’s specific mutation, like many others, is relatively rare, and that has influenced the way she has dealt with her high-risk status:

When I started looking at the information and looking at the risk, and then not knowing what the risk is for our family because ovarian cancer doesn’t have a big enough pool of people to go through ... the breast cancer side and the regular risk is one thing. But knowing the risk was so high and so I feel like my risk was even higher than what was listed on the information sheets. ... they don’t even know what the ovarian risk is ... people first get breast cancer and then they get ovarian cancer later, but that’s not how it’s been in our family. Our mutation was not even on the chart. So that was spooky. They’re giving me the ratios that my risk is 60 to 80% for breast and 40 to 60% for ovarian. But for my family, maybe it’s more.

Ruby, 34

The effect of this perceived lack of information was that Ruby has not felt ready or able to make a decision about moving forward with a risk-reducing oophorectomy, because in order to do so, she would need to have a solid understanding of her ovarian cancer risk.

Pauline also illustrates this important difference when she discusses the timing of her two risk-reducing surgeries. When she was interviewed, she was making preparations to

proceed with her RRBM, which would occur at the age of 31. We discussed how she would know when it was time to move forward with the RRSO:

If I don't have a strong impulse to do it before my fortieth birthday, then I'll know as my fortieth birthday approaches that it's time to do it. As much as I have no frame of reference for breast cancer, I *really* have no frame of reference for ovarian cancer. And unfortunately, the medical community doesn't seem to have much of a frame of reference either because the screening techniques are so crappy. Ovarian cancer is such an unknown in even a bigger way to me than breast cancer.

Pauline, 30

While most *BRCA* mutation carriers interviewed for this study agreed that ovarian cancer is more frightening than breast cancer because of how difficult it is to detect and treat, they also spoke emphatically about their frustration with current breast screening methods. In 2007, the American Cancer Society altered its breast screening recommendations to include annual MRIs for many high-risk groups, including *BRCA* mutation carriers of any age (Saslow et al., 2007). However, for young mutation carriers, even MRI screening can leave a multitude of questions and lingering uncertainty due to the dense structure of young women's breasts and associated difficulty in interpreting the images generated by their MRIs. In relating her experience with screening immediately prior to her decision to undergo RRBM, Rachel recalled her frustration with her physician's interpretation of her MRI:

An MRI on a 33-year-old is very difficult to read because of the type of tissue they are looking at and the fact that when they do an MRI on someone in their fifties, they're not looking at the same type of material as what they are when they look at someone in their thirties.

Rachel, 33

MRI is currently believed to be the best technology available for screening for breast cancer in this population. However, the fact that MRIs are difficult to interpret and many women get called back for repeated tests lends to an increased sense of vulnerability to

breast cancer and constrain their ability to feel confident about relying on screening. Rachel's sense of vulnerability to breast cancer was so high that she decided to proceed with and even completed her RRBM in the time between her initial MRI and the date on which a follow-up MRI would have been scheduled. Finding it intolerably uncomfortable to live with the ambiguity that accompanied her surveillance experience, she swiftly and definitively chose to use surgical risk-reduction instead and was pleased with her outcome and its effectiveness at reducing her perceived risk.

As discussed previously, an important source of information for many women in deciphering their own risk of developing one cancer or the other is what has occurred with other members of their families. Although both *BRCA1* and *BRCA2* are associated with increased risk of both breast and ovarian cancer, some women feel much more vulnerable to one than the other because that cancer site has been more problematic for their family:

I feel like the breast cancer was more of a concern and it's probably not good thinking that, but I'm not as concerned about ovarian cancer because no one in my family has had ovarian cancer. I know that doesn't really make sense, but that's how I think about it. We have only had so many people that we know had the mutation, and that doesn't mean that we can't get ovarian cancer or that I'm not also at increased risk for ovarian cancer. ... I guess I've had so much more experience with breast cancer that that one seems more scary to me ...

Kristy, 29

Charlotte also grew up in a family where breast cancer was well-known, her mother having been diagnosed when Charlotte was in middle school. She recalled,

My mom was diagnosed and beat it, but I've seen so many other people who haven't. It became very real that people do die from this. I think when I was younger I saw it as one of those cancers that people beat and it wasn't as big of a deal as other types of cancers. But the more people I meet in the community, and the more I know about it and the fact that *BRCA* carriers can have a more aggressive type, from what I've read, it's a little bit scarier now.

Charlotte, 26

For both Kristy and Charlotte, this difference in perceived risk actually created a sense of agency in managing breast cancer risk, as they were able to contemplate surgical risk-reduction *via* mastectomy early in their young adulthood rather than accepting an extended period of waiting to see if cancer would strike.

Rose discussed her sense of vulnerability to ovarian cancer, which claimed the lives of her mother when Rose was only 13, and of her maternal grandmother before Rose was born. Based on her family history and long before she was aware of the existence of *BRCA* mutations, Rose was offered entry in an ovarian cancer early detection study at a major university near her home. Rose began a trend of taking charge of her high-risk status by readily accepting this; although she had a sense that her risk of ovarian cancer was increased, she was in grad school and did not have great insurance at the time, and therefore was not being screened:

I was in my early twenties. The genetic counselor told me that there was a test, but probably insurance wouldn't cover it. So I decided not to mess with it at that time. I was young, and had I had a positive result, I wouldn't have acted on it, so it didn't really make any sense to act at that time. I didn't really realize that it was a breast cancer issue, I only thought of it as ovarian cancer, and since I tied ovarian cancer with having my ovaries removed, I knew I would only do that after I had children. I mean, I used to see people with the pink ribbons and think, "that has nothing to do with me. My family history is ovarian cancer." I really dismissed it. Several years later, a second counselor asked me if I wanted to consider the test again and I was like, "why would I want to do that?" And the woman just said it, and I would never have thought about it had she not said it – she said, "Well, you're doing all this stuff for ovarian cancer, but what if you test positive for this gene? You really aren't being aggressive about breast cancer detection." I didn't really get that there was a link. I was so focused on ovarian cancer that it just never occurred to me.

Rose, 30

After she understood the link between breast and ovarian cancer susceptibility and *BRCA* mutations, Rose did not hesitate to be tested. Once she learned that she carried a

mutation, she moved forward with her RRBM quickly, and her surgery was complete within six months of receiving her test results.

Even among women who clearly understand the risks of both breast and ovarian cancer, there are often differences in perception of how negative an experience each type of cancer would be. Trixie had recently completed her RRBM and was midway through the reconstruction process when she was interviewed. Having already minimized her breast cancer risk as much as possible and feeling certain that she did not want children, Trixie had started to think about having her ovaries removed, a procedure that her older sister Maelie (also a participant) had recently completed. Discussing her thoughts on the timing of an RRSO, she stated:

I'm scared to death of menopause. I would get my ovaries out in a second because ovarian cancer is really scary to me, because I can't see it. I've just been afraid of what menopause will do to my body.

Trixie, 27

Similarly, Julia expressed her intense worry about her ovarian cancer risk based on her unique family history. Julia's mother, maternal aunt, and great-grandmother all had breast cancer, and her *BRCA* mutation was inherited from that side of her family; her *paternal* aunt had ovarian cancer unrelated to *BRCA*. All survived except her great-grandmother:

I think that the breast cancer is a scary thought, but ovarian probably scares me more than anything just because it's already on the other [non-carrier] side of my family, I have a higher risk of developing it than the general population, and there's really just not a lot of testing that can be done at this point for ovarian cancer, so I think that's the part that really scares me more than anything. I actually think about having my ovaries out far before I would even think about having my breasts removed.

Julia, 24

Julia carries the same genetic mutation as the three individuals in her mother's family with breast cancer, but identifies strongly with the ovarian cancer risk conveyed by her

paternal aunt's diagnosis given its overlap with the risks inherent in *BRCA*; she was in a unique position in that there were relevant cancer risks on both sides of her family. It is striking that of the two primary cancer sites at risk in the context of a *BRCA* mutation, ovarian cancer is both harder to detect and prevented by what many women view as the more physically difficult risk-reducing surgery. This is an especially challenging dynamic for *young* women to navigate, as deciding about RRSO forces them to contemplate not only ending their ability to bear children, but altering their natural hormone levels decades earlier than would naturally occur, leaving them susceptible to numerous health challenges associated with menopause (*e.g.*, decreases in bone density/risk of fracture, vascular health/risk of coronary disease and stroke, skin elasticity).

Julia's sentiment about the lack of effective screening for ovarian cancer is further reflected in what several women had to say about their confidence in the screening methods available for each potential cancer site (mammogram, MRI, and clinical breast examination for breast cancer; trans-vaginal ultrasound and the CA-125 blood test for ovarian cancer). Almost universally, participants believed that although standard breast screening procedures are sufficiently effective in catching cancer early enough to be treated easily and successfully, current ovarian cancer screening procedures leave them vulnerable to this "silent killer"⁴. Each participant who was currently relying on screening for risk-management was asked how confident she felt that the screening was protecting them from cancer. Here are some of their answers:

⁴ Their assessment of the utility of ovarian cancer screening is entirely accurate. There is simply no evidence at all to suggest that screening with transvaginal ultrasound and CA-125 results in earlier diagnosis or, most importantly, improved cancer-related survival. Despite acknowledging that these screening tools are ineffective, and NIH Consensus Conference on Ovarian Cancer recommended that they be used in women who are at increased genetic risk of ovarian cancer. Consequently, this screening strategy has come to be seen as the medical "standard of care." Health care providers therefore feel compelled to use it.

The breast screening, very confident. The ovarian screening, not confident at all.

Marjory, 30

[The breast screening] does give me reassurance that I can say, 'If I test it, it's going to be caught early and it's a good thing that I'm doing these tests, I'm able to go about this proactively and catch it early. And while ovarian cancer hasn't been in my family, I do worry about that a little more now because you're not able to catch it as early. So I don't have that peace of mind when I'm getting those tests. It's definitely a little scary because I know if the results were to come back, I guess it's more likely that it would be at an advanced stage. I don't have that peace of mind that it would be caught early on.

Charlotte, 26

I'm going to play the game. I'm going to participate in the charades of ovarian cancer screening. I understand that it's a little bit of smoke and mirrors. A lot of smoke and mirrors. But that's what's recommended, so I'm going to do it.

Pauline, 30

Breast cancer, yes. Ovarian cancer, I think it's still concerning to me because the screening methods aren't the best.

Wanda, 32

Taken together, these thoughts suggest that part of why many women feel so much more vulnerable to ovarian cancer than breast cancer is because they realize how inadequate current screening techniques for ovarian cancer are, and rightly feel that their options for successful ovarian cancer risk management are limited. A belief that the only effective method available to protect them from ovarian cancer is to undergo RRSO can feel inhibiting for many due to desires for childbearing. Unfortunately, this is a problem that can only be solved by advances in medical technology that make it easier to screen for ovarian cancer and detect it early, while it is still treatable; in that case, ovarian cancer survivability would improve and it might seem less frightening to members of this population.

BRCA1 versus BRCA2 risk. Because the manifestations of *BRCA1* and *BRCA2* mutations are broadly similar, they are often discussed together in publications to which

women turn for information about their risk. Therefore, some women are exposed to information about the risk differences that distinguish *BRCA1* from *BRCA2* that may mitigate or exacerbate their perceived risk of cancer. In discussing her risk, Leigh appeared very knowledgeable about the differences between *BRCA1* and *BRCA2* and their associated risks, having learned a good deal about her own *BRCA1* mutation by searching for information online:

I think that Myriad has classified certain mutations including my own as deleterious as opposed to unknown or harmless, but beyond that I don't think they have different gradations of deleterious. I think they're all sort of lumped the same. And for my mutation they said the risk hadn't been exactly established but generally for *BRCA1* it's between 64 and 87% for breast cancer. But with *BRCA1*, the cancers tend to be triple negative and those cancers tend to be more aggressive. *BRCA2* carriers, their cancers tend to be more in line with what the general population would get and a little less aggressive and receptive to certain treatments.

Leigh, 35

For Leigh, the knowledge that she was a carrier of *BRCA1* put her in a higher category of risk than she would have been if she carried a *BRCA2* mutation. In other cases, making inappropriate distinctions between the two mutations can lead women down a path that is not quite in line with what is known about *BRCA*-related risk of cancer:

I've heard up to 85% [risk] or so for the mutation. But from the explanations I've heard how my *BRCA2* is mutated but *BRCA1* is there and things like the environment and age are what affects that and eventually could cause cancer if that first one kind of drops out or doesn't start doing its job.

Charlotte, 26

Charlotte interpreted the mutation-specific information she was given to mean that the fact that she did *not* carry a mutation in *BRCA1* was a protective factor in the context of her positive *BRCA2* mutation status, which had the effect of erroneously reducing her cancer risk perception. In fact, a functional *BRCA1* gene is not a protective factor in this situation; almost universally, individuals who have one *BRCA* mutation do not have the

other. This example highlights the need for very clear communication between medical professionals and mutation carriers in educating carriers about their cancer risk, as well as for patients to have multiple opportunities to ask questions or seek clarification about the information they are given.

Specific Periods of Risk

Risk associated with specific ages. Participants had different ways of conceptualizing during which parts of their lives they might be especially vulnerable to breast and/or ovarian cancer. Some women were able to separate the daunting 87% figure associated with lifetime risk of breast cancer from the lower, and therefore much more manageable risk estimates associated with risk of breast cancer for women in their twenties and thirties over shorter periods of time, *e.g.*, the next 5 or the next 10 years.

Rose illustrated this nicely when she stated:

In learning some of the numbers and how you can take some of the numbers to mean certain things and other things, you know, like the 80% risk over the course of your lifetime. But that means, [I learned after talking to the doctor more], I'm only at 30% risk *now*.

Rose, 30

Focusing her attention on her cancer risk during the next 5-10 years, rather than worrying about lifetime risk, allowed Rose to bring her risk perception down to a more manageable level and select a plan for risk management that would allow her to accomplish family formation goals comfortably; she was at home recovering from RRBM when interviewed, and was looking forward to starting a family soon after her recovery. For other women, risk perception changes in fits and starts, sometimes with entire decades being identified as either risky or not so. Marjory worked in a setting in which she

routinely encountered other *BRCA* mutation carriers, discussed how she managed her interactions with them:

[Patients] would tell me that one week they were fine, and the next week they had a diagnosis. And that was very hard for me, knowing that my risk was so high and I was getting into my thirties. The twenties seemed invincible and the thirties seemed like, well, now this could happen.

Marjory, 30

Similarly, Nichelle discussed how she balanced family history and knowledge of other cases of breast cancer in young women in deciding whether she wanted to know about her own mutation status after other members of her family had been tested:

I think the biggest reason [I decided to get tested] was, I'm in a sorority and one of our alumni was diagnosed with breast cancer. I want to say she's about 25. ... So, knowing that...I mean, I know ours typically doesn't show up, based on family history, until mid- to late-forties, but I decided, "you know what? There's always that chance that something could happen. And if I am positive, it could happen sooner than that." So I really wanted to have that knowledge and start doing what's healthiest for my lifestyle.

Nichelle, 21

Marjory and Nichelle provide a useful contrast: whereas Marjory discussed her perception of elevated age-related risk in a way that revealed a sense of doubt, Nichelle's approach was focused on making positive changes to her lifestyle to address cancer risk.

Like Nichelle, an important component of age-related individual risk perception for many young women is the age at which other family members were diagnosed with cancer. Many see the ages at which their relatives were diagnosed as "danger zones" that signify an elevated threat of cancer for themselves (Werner-Lin, 2007). For example, after finding out that she carried a *BRCA* mutation, Melanie thought about her aunt's cancer history, remembering, "because my aunt was only 28 when she was diagnosed, it was scary because I was in my early twenties. But I just kind of said, 'Okay, we know this – now what are we going to do about it?'" For Melanie, the age at which cancer risk

felt more threatening was fairly low, which encouraged her to think about shifting her timeline for family formation in order to allow for risk-reducing surgeries (this concept is discussed in more detail in Chapter 9). In contrast, Serena listed the three cancer cases with which she was familiar in close family members:

My grandmother is a thirty-five year survivor of breast cancer, she had it twice. My mom is the oldest of four and she was diagnosed with ovarian cancer twelve years ago now, and she's completely in remission. Her sister next in line had breast cancer. The magic age in our family is forty-six.

Serena, 30

Serena's experience of young adulthood thus far had been quite different from Melanie's; because the age at which cancer seemed like a major threat was much later for Serena, she was able to navigate through her twenties and into her thirties without her *BRCA* mutation being a major stressor, and without feeling as though she needed to alter her life plans in any way because of it. In fact, Serena stated that "my [biological] clock is definitely ticking, but it's not that loud yet. It's not ringing in my ear right now," suggesting that her risk perception and perceived need to manage risk were not constraining her family formation plans.

Rylan had a somewhat dismissive attitude about her cancer risk for the first several years after she was tested. She attributed her recent change of heart to "being the age mom was. She was 31 when she was diagnosed, and her sister was 39 when she got it, so being in my thirties just sort of woke me up and made me start getting a little nervous." Rachel recalled that the idea that specific periods of risk existed and could be determined based on relatives' ages at diagnosis was reinforced by her doctors:

What happened was all the doctors kept telling me, "you're at risk in the neighborhood of ten years before your mom had it." So, here I was, mom was 40, I was 29 or 30, just shy of my 30th birthday. So yeah, there was definitely a reality of the fact that this could be happening. Nobody tests a 30-year-old woman for

anything like that. So what's to say it's not already growing? Do you really think when you go in to the typical Ob/Gyn that she's very intent and closely watching the breast tissue when she's doing a hand examination on a 30-year-old? I am not sold on that. So I definitely had, I think, almost an undue fear of that and because of knowing that information.

Rachel, 33

Trixie was involved in the breast imaging study beginning at age 23. During the first few years, she remembered feeling fairly confident that screening was effective in ensuring that she remained healthy. However, almost immediately after her time in the study ended, Trixie elected to undergo RRBM at age 27. Asked about how she knew that it was the right time to move forward with surgery, and she stated:

I'm 27 and my sister was 28 when she was diagnosed and I just felt like a race against the clock, like I wanted to get it done before 28. I know, not necessarily would I have gotten cancer at 28, but to me, that number stuck in my head. I want to see 28. I want it to come and go and I want there to be no cancer.

Trixie, 27

For Trixie, the experience of her older sister's two breast cancer diagnoses and eventual death at age 32 served as a powerful marker which she perceived as a looming threat. Although her sister was uniquely vulnerable due to having two *BRCA* mutations (an extraordinarily rare situation that almost certainly resulted in her developing cancer at an earlier age than she would have had only one been present), Trixie's anxiety about meeting a similar fate was enough to lead her to decide to have RRBM fairly early. Together, Trixie, Rachel and Rylan demonstrate that awareness of increased risk based on the ages at which family members were diagnosed with cancer can lead to a greater sense of urgency about risk management, and may move mutation carriers closer to pursuing surgical risk-reduction.

In contrast to the women quoted previously who discussed increases in risk-related anxiety based on proximity to ages at which other family members were

diagnosed, some other participants described situations in which their age-based risk seemed low because they were younger than the age at which other family members had been diagnosed with cancer. Julia, 24, was among the youngest participants in the current study and had not yet started screening; she was preparing for her first screening appointment about three months from the date of our interview. When I asked her about her decision-making with regard to risk-management at this point in her life, she stated that she had chosen screening “because I do have quite a few years before the age that my mom was when she had the cancer. I guess I don’t feel any immediate risk or the need to do anything dramatic.” Similarly, Charlotte remembered:

When I first got [my genetic test results], I would probably rate it pretty low, but because my cousin had it at 37, I guess there are some people that say ten years out is when I should really start worrying and considering prophylactic surgery. And so, as I get closer to 27, I’m definitely getting more and more worried that it could become a reality.

Charlotte 26

Charlotte’s experience also nicely demonstrates the transition from a period of relatively low perceived risk to one of higher perceived risk, setting the stage for significant decision-making about risk-management.

Risk associated with reproductive issues. As discussed previously, family formation is one of the major developmental events that typically occurs during young adulthood (Arnett, 2004; Rodgers & White, 2004). Accordingly, many of the participants in the current study were very much occupied with the idea of having and raising children as they were also learning about their own cancer risk. Dynamics related to family formation and risk-reduction will be detailed in Chapter 9; however, this discussion of specific time-related vulnerabilities would not be complete without touching on the subject here, because knowledge of these issues is a major source of anxiety for mutation

carriers. Libby found out about her *BRCA* mutation during a time when she was undergoing fertility treatments, which was specifically frightening for her because of the potential for increased cancer risk due to very high levels of hormone exposure:

I felt very vulnerable, actually. The more I read, the more freaked out I started to get. I had taken a round of hormones to do an artificial insemination treatment, because I just really wanted to get pregnant. And I was taking a really low dose of hormones, because the doctor knew I had the mutation. After that, I felt very vulnerable. I was like, “oh my God,” because it didn’t work. I thought I might have just put myself in a really bad situation. I felt really vulnerable also with the ovarian cancer, because with the fertility stuff, they track your ovaries so much. I’m constantly getting ultrasounds of my ovaries and I just felt really vulnerable in that sense. It’s good that they’re looking at them, I guess, but they’re not necessarily looking for cancer.

Libby, 32

Clearly, for Libby, the issues of cancer risk and fertility were very much intertwined.

This was also the case for Rachel, who had had one baby with the help of fertility drugs before she was tested for the *BRCA* mutation, but wanted to have more children. In putting these two pieces of health information together, her risk perception was based on the fact that “there is still no real knowledge as to what being on the fertility drugs does to you. Great, they got me pregnant, but especially in a *BRCA* patient, is that increasing my risk?”

The connection between family formation and risk perception was likely intensified for Libby and for Rachel because of their use of fertility treatments, but it is also the case that women not involved with fertility treatment feel a similar sense of increased vulnerability during the period in which they are attempting to get pregnant, while they are pregnant, and in the months (or years) following a pregnancy. Leigh was the mother of several young children when she learned that she carried a *BRCA* mutation, and recalled having conceptualized her risk as

Very high. Also, the early onset of the ovarian cancer aspect of this was especially troubling. By that time I'd read on the internet of several anecdotal cases of young women with very young children who developed cancer while breastfeeding or shortly after giving birth, so I was very terrified of my immediate risk at that time. I thought my risk of both cancers was very high.

Leigh, 35

The elevated cancer risk that many women associate with reproduction is also related to the fact that effective screening is nearly impossible during pregnancy and while breastfeeding, because of the potential dangers to the fetus associated with exposure to radiation and hormonal-related changes in breast tissue which interfere with obtaining clear, interpretable mammographic images. Additionally, the normal palpable changes to breast tissue that occur naturally during pregnancy and while breast feeding may be seen as threatening to women who are already very aware of any such changes.

Previous cancer scares. About half of the women interviewed for the current study had experienced at least one “cancer scare,” usually a test result that came back abnormal and required follow-up or biopsy. In some cases, these occurred before the participants knew that they carried a mutation in *BRCA*, and in other cases they were aware of their mutation when the scares occurred; still others had had multiple cancer scares across different stages of mutation awareness. Chris explained how, having already experienced two cancer scares prior to being tested for the mutation, her positive test results moved her to take action to reduce her risk:

You just kind of feel like you're underneath a black cloud all the time, and I just felt like it was coming, and you just want to beat it before it gets you, pretty much. I mean, you kind of forget about the 13 to 20% chance that you won't get it and focus on “I am going to get it.” And so, you just feel like you're waiting for it. And knowing that I already had two lumps that were giving me trouble, you just kind of think, “Well, this is stupid. It's stupid to sit here and dwell on that and think that it's going to happen. Might as well just have the surgery.

Chris, 33

Trixie further exemplifies this phenomenon. After her oldest sister was diagnosed with breast cancer twice within four years and passed away at the age of 32, it was discovered that both their mother and father carried *BRCA* mutations – one in *BRCA1* and the other in *BRCA2* (Trixie had inherited *BRCA1* but not *BRCA2*). This, combined with Trixie’s own experience at having abnormal test results throughout her years in the BI study, resulted in an intense feeling of vulnerability to cancer during the years between learning about her mutation and having an RRBM:

I felt like I was definitely going to get cancer, especially because I have some weird spots on my MRI. And I was watching someone so young and healthy get cancer in my own family, and you can read varying statistics about your risks when you’re *BRCA1*, and some studies would say 50% and some would say 95% likelihood you’re going to have cancer. And so I tended to cling to those statistics just because I was watching [my sister] have cancer.

Trixie, 27

Soon after her involvement in the BI study ended and she no longer had that access to intensive breast and ovarian screening, Trixie elected to undergo RRBM.

Trixie’s experience is another example of “screening fatigue,” in which women become emotionally exhausted from dealing with peaks in anxiety surrounding their frequent screening visits and requests for follow-up tests based on suspicious or ambiguous results, ultimately confirming their sense of doubt that screening is effectively protecting them from cancer. When asked to describe what it was like receiving abnormal results after her screening visits in the BI study, Grace remembered the experience as

terrifying, because every time you get an abnormal you think, “Is this the time it’s cancer?” So you start mentally preparing for it. If it is [cancer], then what surgery option would I do, and what kind of treatment would I need...you take it to the next level.

Grace, 30

Similarly, Kristy recalled:

It was really sort of traumatic ... having the irregular results, and I had that a couple of times. And I kind of got to the point where I was like, this is great that I'm able to get screened high-risk and the doctors are extra careful with it, but it's kind of to the point where it's just making me uncomfortable because everything is a red flag and everything they want to explore more and everything is just sort of scary, because you never know.

Kristy, 29

For both Grace and Kristy, and for many other participants, the repeated experience of having screening tests come back as inconclusive or in need of additional follow-up and/or tests became too much to bear, and they decided to proceed with RRBM prior to having their first child despite having planned to wait at least until after they had had an opportunity to breastfeed once (again, this will be discussed in greater detail in Chapter 9). For Grace, this surgery occurred at age 29, and for Kristy, at age 28. In cases such as these, it seems that over time, the experience of screening fatigue and multiple cancer scares outweighs the desire to preserve one's natural breast tissue.

Perceived risk of other cancers. Many participants mentioned that they were aware of *BRCA*-associated risks for other cancers beyond breast and ovarian, and there was some variation with regard to how much this was a source of anxiety. Several women identified other cancers for which there was a family or personal history that contributed to an increased risk perception. Recall that Valerie was a genetics researcher and therefore had a great deal of knowledge about genetic issues in general, including many details about *BRCA*. She mentioned both melanoma and pancreatic cancer as specific additional risks:

Melanoma scares me. A lot. I'm incredibly mole-y, and that actually comes from my dad [from whom she had inherited her mutation]. ... He and I are both very, very mole-y, so it scares me knowing that I come with a slightly increased risk for it. ... I feel like that's kind of a bad combo. So, I always knew I was going to have to keep an eye on my breasts and ovaries, but some of the other *BRCA2* spectrum cancers I wasn't even aware of. I didn't know about melanoma. I didn't know

about colon or pancreatic cancer. Unfortunately, there's really not a lot you can do in terms of screening for pancreatic cancer. My doctor said it needs to be in the back of my mind if I have some other medical issue I can't explain. I'm not necessarily enthralled with the idea of always having to go through life thinking, "could it be my pancreas?"

Valerie, 29

Similarly, Libby identified colon cancer as a potential future risk exacerbated by the fact that she had already been diagnosed with a mild case of ulcerative colitis and wondered if this might someday become colon cancer. She planned to monitor carefully.

Cancer in other components of the reproductive system was also a perceived risk for many women. This is an interesting issue because many women describe these other cancers as being "in their family;" however, it is quite possible that when other gynecological cancers are listed as diagnoses in older family members, these may actually be ovarian cases that were mislabeled or misdiagnosed prior to the availability of medical technology accurate enough to label gynecological cancers appropriately.

Marjory believed that even if she chose to remove her ovaries, she might still be at risk for cancer in other parts of her reproductive system: "I wouldn't be worried about getting ovarian cancer after I had my ovaries out, but I'd be worried about the primary peritoneal⁵, because it's in my family." Similarly, Wanda stated that after having an RRSO,

I still think I'll need some form of screening, because I could get cancer. If I only have the oophorectomy done, I could still have cancer in my other reproductive organs, so I still want to be screened regularly. I think I'll feel much better, but I think there's still a chance that I could get, if not a reproductive cancer, I could get some other type of cancer, so I think I just have to be diligent about screening.

Wanda, 32

⁵ Primary peritoneal cancer is a malignancy which strongly resembles ovarian cancer, both under the microscope and in its response to treatment. It is thought to arise in cells within the peritoneal lining of the abdomen, which are known to share a similar embryologic origin with ovarian epithelial cells.

For Wanda and other women like her, it seems reasonable to speculate that completing risk-reducing surgery might completely eliminate cancer risk or the perception of still being at risk with its attendant anxiety, since there is a strong belief that cancer might still occur at another site. The experiences of these women illustrate instances in which completing surgical risk-reduction might leave some carriers with lingering feelings of high risk perception, because of fears that risk is not limited to only breast and ovarian cancers⁶.

Anxiety About Cancer Risk

There are a minority of *BRCA*-positive young women who seem to truly not feel particularly vulnerable to cancer, at least in the short term:

I still to this day don't think it's coming soon. And I hope that's not a naïve attitude. I don't think it's something I could feel vulnerable to until I get told, "Noelle, you have cancer." I think at that point, I'll know. ... right now I'm dealing with it and moving forward with my life. I do research, I'm up on medical journals, and things like that but I don't know how real it will be until it actually happens.

Noelle, 26

It seems that Noelle has found the ideal mix of being aware and educated about her risk of cancer, but not letting it get the best of her or consume her life. For many women, however, learning that they carry mutations in *BRCA* is the beginning of a very stressful period, marked by self-doubt, intense scrutiny over any health-related challenge, and hyper-awareness about risk. Lilly, who had only known about her risk for about a month at the time of her interview, illustrated this nicely in her response to a question about how she thinks about her risk now as opposed to before she was tested:

I definitely could be worse about it, but there's not a day that goes by that I don't

⁶ In fact, the best available evidence suggests that neither endometrial cancer nor cervical cancer occur excessively in women with *BRCA* mutations.

think about the threat. At this point, I'm happy I was able to go for so long not really, really, really worrying about it. It always was a worry, but now I question everything that I put into my mouth, or every bump, every feeling, every pain.

Lilly, 25

Sense of inevitability and fatalism. Many women from HBOC families experience a phase in which they believe that breast or ovarian cancer is an inevitable part of their future. This may often be based on the extent to which family histories seem to be saturated with relatives who have been diagnosed with cancer, or at least in which close relatives have received diagnoses. This sense that cancer cannot be avoided may be problematic when it precludes recognizing that one has the capacity to take actions to reduce her risk. When asked about her previous beliefs about her own vulnerability to cancer, Leigh recalled, "after my mom and her only sister had breast cancer, I sort of used to think I had 100% risk of breast cancer." Similarly, Dawn remembered that before she had risk-reducing surgery, she felt "a horrible feeling that I knew I was going to get it at some point. I would definitely get it." For Melanie, receiving positive mutation test results "confirmed what I already knew. I knew that I was at a higher risk anyway, and you feel like you're broken, and it's a foregone conclusion. Like, when is my time going to be?" Despite knowing that the calculated risk of breast cancer was 87%, Beth believed that "for me it may as well be 100% because with my family history it's just too prevalent." Reina recalled how her mother's two breast cancer diagnoses contributed to her own feeling that cancer was inevitable, even before she was tested for *BRCA*:

I struggled with it for many years because I always had the assumption that I was inevitably going to get it. It just was a part of who I was. I don't think there was a day that went by where I didn't think about it. And then when I got tested and found out that I was positive, then I had to revisit all of those emotions and it just became that much more relevant and real. I struggle with it every day.

Reina, 29

For some women, the threat of cancer is not only inevitable, but urgent. This feeling, coupled with a belief that risk is changeable through action, can move women toward making definitive risk-management decisions. Kate learned that she carried a *BRCA* mutation at age 32 and was from a family in which there had been six known cases of *BRCA*-associated cancers resulting in three deaths; Kate's 39-year-old sister was in the middle of treatment for breast cancer when Kate was tested:

I felt like if I didn't do something immediately, I would get cancer right away. I felt like, "I've got to do this *now*, there's no time to wait. I don't want to take any chances." Maybe because of my sister's age, she is about seven years older than I am. I didn't want to wait for it to happen. I felt like, "It's going to happen." I felt very sure that it would happen sooner rather than later.

Kate, 35

Valerie's family history of breast and ovarian cancer was quite pervasive, with her mother, aunt, several great aunts, and great-grandfather all having been affected by breast cancer. She described her feeling that cancer would be coming for her next:

Cancer always felt like it was hanging over my head ... Really from my mom having it so much and my whole family. It seemed like we just traded off. Mom was sick for so long [and then she died], and then my aunt became sick, and she passed away. And then by that time, my great-grandparents were getting up in age, they were sick. And I just sort of felt like... it kind of felt inevitable. But at the same time, I kind of felt like, if I knew it was inevitable, I could keep an eye out for it. It wasn't going to sneak up on me. But I was sure I had two ticking time bombs strapped to my chest.

Valerie, 29

Valerie's and Kate's recounting of their experiences nicely illustrate the pressure that many young *BRCA* carriers feel to beat cancer to the punch – when women believe that their risk of cancer is very high, it often comes with a belief that they must do something to mitigate their risk *quickly* - to remove the dangerous body parts before they have a chance to develop cancer. That is precisely what both chose to do: Valerie had an RRBM

at the age of 28, and felt extremely confident that she had made the right decision for herself; Kate was planning to have her RRBM within the year following her interview.

Isabelle is the younger sister of Lilly, who was quoted previously in this chapter. Their mother had breast cancer at 25 and again at 36. Isabelle recalled her own conceptualization of her breast and ovarian cancer risk prior to being tested:

I was never really super vigilant about my own breast self-exams. I always just assumed that I would get breast cancer, but I've always had the positive outlook I'm going to get it, it's going to be caught early because we're aware of our family history, and I'll be fine. I think I'm going to get it if I don't have a prophylactic mastectomy. I don't know when, I just have a feeling that I'm going to get it. I don't think I'm going to die from it. I just think I'm going to get it, go through it, and end up healthy.

Isabelle, 22

Isabelle's experience reveals another unique issue related to breast and ovarian cancer risk perception. While living in constant fear of a cancer diagnosis is clearly neither healthy nor productive, the total *absence* of fear – the failure to accept cancer as a genuine threat – may also be counterproductive if it impedes one's ability to take the risk of cancer seriously and be vigilant about screening and/or risk-reduction, thus becoming a barrier to action.

Surprisingly, even some women from breast/ovarian cancer families did not fully appreciate the extent to which the presence of a *BRCA* mutation dramatically increases cancer risk relative to that of non-carriers. This fatalistic thinking about risk is another example of how dynamics of agency and constraint act on the risk-management decision-making process of young mutation carriers. For example, Sophie stated that during the time before she was tested but knew there was a mutation in her family,

I think I thought it would be just like anybody else. You can still get breast cancer with or without the gene, you can still get ovarian cancer, although I think the risks are pretty low. But you still have all these risks. You can never be a

smoker and you can still get lung cancer. And I still to this day kind of think of it that way. You know, it could happen to anybody, my risks are just greater, but at least I know so I can be proactive about it.

Sophie, 31

Sophie's perception of her risk does not align with her scientific risk; however, her binary thinking about risk is not uncommon, and has been observed in previous research (Pilarski, 2009).

Given that the general culture of young women interviewed for this study is quite risk-reduction- and action-oriented, it is interesting to imagine the relationship implications for members of a family or couple in which one individual takes a fatalistic approach and one does not. Sadie provided some insight about this in describing her younger sister, also a mutation carrier:

I think that she and I really differ in our approach to this whole thing. ... I like a lot of scientific hard facts, and she likes religion and fate and relying on the cosmos and stuff like that. ... She just believes the Buddhist philosophy that if it's meant to be, it's meant to be. She is just going with the flow, she lives a happy and full life and she has every intention that being a good person, she won't have cancer. So to her, the statistics don't really mean a whole lot. Before I went in for my surgery, I mentioned again to her if we were going to bet, with an 80% chance that you're going to win, you would probably play all those. And her response was that even if the risk was 99% that you would get cancer, if she's in the 1% then that's 100% to her. So that's really hard for me to understand.

Sadie, 33

The obvious differences in Sadie's and her sister's perception of their breast and ovarian cancer risk and their decisions about how to manage that risk have added a difficult element to their relationship and leave Sadie feeling quite frustrated in her helplessness in convincing her sister to take her cancer risk more seriously. Her sister's willingness to sit back and wait to see what would happen is striking in contrast to the decisiveness with which Sadie has acted to manage her own risk, having undergone RRSO at age 32. It seems quite likely that this has had further implications for family harmony and

interpersonal dynamics between the two sisters and other members of their family, a dynamic that might be repeated in other HBOC families.

Some women may also adopt a fatalistic attitude toward their risk in considering the chance that they might develop cancer even after risk-reducing surgery. In this context, the fatalism seems to be akin to managing residual risk perception even after having done everything possible to mitigate the risk of cancer. Consider Maelie, 33, who stated, “at this point, we’ve done all the risk reduction surgeries and things that we can do. I feel with what I have done, if I get cancer, I’m meant to have it, because there’s really nothing more I can do.” Similarly, when asked how she might feel about her risk after completing both surgeries, Pauline stated,

If I do get cancer after I do my surgeries, I think you’ve got to have a little bit of a sense of humor about it, like “Oh, this was so programmed into my body that it came through even though... well, at least I tried.” And if that’s really, really the way that I’m going to go, then I guess I can’t fix it. I don’t believe much in destiny, that we all have this specific predetermined plan and that we all follow it. But if this is my genetic destiny, if this is truly, truly how my body will expire, even after I have the surgeries, it’s like, “Gosh! Okay.”

Pauline, 30

For Pauline and Maelie and other women like them, there may be a profound comfort and even peace in being able to reach this state of mind. In some ways, this may be the optimal outcome of all available risk-reducing strategies, and is certainly preferable to doing everything that can be done, but continuing to worry that some stone has been left unturned. Instead, they have acted with agency but recognized that doing so has its limits, and some things simply cannot be controlled.

Managing anxiety. Risk-related anxiety seems to shift over time, peaking in the weeks and months surrounding one’s genetic testing and often fading somewhat thereafter. For many participants, these high levels of anxiety reappear with their

regularly-scheduled screening appointments, as discussed earlier in this chapter. Recall that Leigh highlighted her high levels of worry and anxiety dreams when she realized that she was at very high risk of breast cancer, compounded by the recent loss of a peer to lung cancer. Leigh recalled having experienced these high levels of anxiety for a while, “until I realized that just because I have the mutation doesn’t mean I’m definitely going to develop cancer right now. So at some point I relaxed a little bit.”

Some women mentioned lifestyle changes that they believed they could make in order to reduce the negative impact that their positive *BRCA* mutation status would have on their lives and emotional well-being. Reina had been seeing a therapist for most of the time since she found out she was *BRCA* positive. From that, she had taken away the message that

It’s something you really have to make an effort, make a life change, whether it be eating only organic or making sure you go to yoga classes every week, or don’t eat out as much, try and de-stress your life – which is like a joke. All these things that I know can help trigger the mutation, but in the everyday environment it’s really hard to shift in those directions. I definitely fall into the trap of being a workaholic, I have a very busy job that kind of consumes me a bit.

Reina, 29

Reina’s experience suggests another interesting dynamic related to managing cancer risk anxiety, because nobody really *knows* that the lifestyle changes she listed truly impact the likelihood that a given mutation will be “triggered” and cancer will develop. However, thinking this way does seem logical and it provides a rational basis for making such changes. Perhaps some mutation carriers cope with their risk in part by developing a framework in which they choose to view their risk, convince themselves it is correct, and then set about doing all the things that framework would dictate, creating a context in

which they can take action and ultimately feeling that they are taking the very best care of themselves in the process.

Using humor to manage *BRCA*-related cancer risk anxiety may seem impossible, but when it is done with aplomb, this strategy can be quite effective. Pauline was one of the most humorous and good-natured women interviewed for this current study, and her consistent use of humor in navigating the mutation-positive experience was striking. This was her response when asked her to describe what it was like to receive an abnormal result on a recent MRI:

Well, to be honest with you, I was sort of expecting it because I had heard that MRIs are incredibly sensitive technology. So I wasn't entirely surprised when I heard that they had seen something suspicious. I wasn't thrilled, of course, but I just sort of felt in my heart that it wasn't going to be anything big. And I sort of have this joke with myself that my right breast is my bad breast. That's where I found the lump a few years ago and pretty much from the day I got my *BRCA* result, even a few months before that because I knew it was coming, I sort of had pain in that right breast. So when the radiologist called and said, "Oh, we saw something suspicious in your left breast," I thought, "Oh, well then obviously nothing is wrong because that's the good breast." So I actually surprised myself at how well I managed the anxiety.

Pauline, 30

Ultimately, the optimum way that many women manage their cancer anxiety is by taking action that truly reduces their risk, producing a shift from feelings of helplessness or vulnerability to pride and security in having decisively taken control. At 25, Lilly was among the youngest women interviewed for this study, and she had only known about her mutation for a little over one month. Lilly's mother had been diagnosed with breast cancer at age 25 and again at 36, and this was a critical point in Lilly's concept of her own risk. She stated,

I don't deal well with risk and health. So I just said, 'I want to get my breasts removed.' I don't have a boyfriend, I can't spare my breasts assuming I'll find a guy who will have a problem with implants or the fact that I won't be able to

[breastfeed]. So I have pretty much decided that I would like to have the surgery to remove as much of this fear about breast cancer as possible. Then, if and when I get married and have kids, or if I don't, I would like to get my ovaries removed as well. And if there was something to remove the other risks of other cancers as well, I would do that, too. I'd just like to remove as much of the anxiety and fear about these things as I can.

Lilly, 25

At the time she was interviewed, Pauline had only known about her positive mutation status for about two months. She was actively contemplating risk-reducing mastectomy, but had not yet scheduled the surgery or decided about reconstruction options. She recalled how she felt about her breast cancer risk upon finding out that she carried a *BRCA* mutation.

My first thought was that my breasts are expendable. My breasts are not worth the anxiety that knowing that I have a super high risk of developing breast cancer would bring. ... I have obsessive-compulsive disorder. So you can only imagine the idea of having four screenings a year and the attendant anxiety that comes with those things, especially as it relates to my personal brain chemistry. That's going to be really – excuse my language – fucking hard to do. It's going to be really, really hard to be anxious for maybe a week before and maybe a week after, four times a year. That's like two months of my year that I'm just going to be out of commission eating vegan cheesecakes, sleeping, doing all the crazy stress things that I do.

Pauline went on to explain her belief that having the risk-reducing mastectomy will change her quality of life by reducing her day-to-day anxiety about her risk:

Right now I'm walking around like, "I'm *BRCA2* positive, my name is Pauline." It's like my status is more important to me than my name right now. That is the most prominent thing about me. And after the surgery I think that I'll be able to go to a place where I'm just who I am again...if I wasn't going to have emotional relief from this, I wouldn't plan on going into surgery.

Pauline, 30

Valerie is another young mutation carrier who experienced a peak in cancer anxiety, followed by a feeling of relative peace connected with making the decision to have an RRBM. She recalled,

Over time, I realized that just knowing that there's a mutant gene there doesn't mean that anything's actually changed. I've had the mutant gene since I was born. I knew in my mind that I was positive. And when [the doctor] told me I was positive, it didn't alter my mindset much, although it did – I don't want to say that it was the nail in the coffin because that's really kind of an awful way to say it. But that's kind of the best analogy. It was sort of the final piece of the puzzle. I decided, "All right. We've got to get rid of them."

Valerie, 29

Perceived Risk After Surgery

One of the major benefits of risk-reducing surgery for *BRCA1/2* mutation carriers is that it usually brings about considerable relief with regard to perceived risk (van Oostrom et al., 2003). However, the transition from being a "high-risk" person to a person at lower risk is not always smooth or complete, and does not always feel natural for *BRCA* mutation carriers. When she was interviewed, Sadie had already completed her RRSO. Asked whether the surgery had affected her experience of fear and stress about breast and ovarian cancer (both risks are substantially reduced after RRSO), Sadie said,

It's hard for me to identify with a lower risk of ovarian cancer. It's hard for me to say, 'well, I've had the surgery and I guess I don't really have that risk anymore.' Because there's always that slim chance that you're going to get some look-alike abdominal cancer, that it's going to pop up that we didn't get all of the cells, or that type of thing. I haven't really felt that huge relief, I guess. Because in my mind, it's like, 'well, maybe it's gone, but you know, it's probably still going to be there.'

Sadie, 33

It is interesting that the completion of an RRSO does not seem to have brought Sadie any significant relief from anxiety related to her cancer risk. Sadie's family is one in which "all the women get cancer," and many women in her mother's generation had opted to have risk-reducing surgeries even before genetic testing was available. Sadie's mother had both RRBM and RRSO in her twenties and had never been diagnosed with cancer. Having had her own ovaries removed, Sadie believed "I'm always going to have that

little bit of fear. I'm always going to feel like there's some risk there." With regard to her breast cancer risk, Sadie stated, "I know intellectually that by having the ovaries removed, that my risk of breast cancer has diminished just a little bit. But as far as the overall risk, I still think that's probably the same." It seems that although Sadie's evidence-based risk of both breast and ovarian cancer have decreased significantly after her RRSO, her perceived risk is relatively unchanged; going through the surgery has provided her with very little relief from the troubling anxiety she has experienced since learning about her mutation. This would likely be well-addressed by a detailed, thoughtful interaction with a genetics professional who might be able to help her achieve a more realistic assessment of her current status.

At the other end of the spectrum from Sadie is Kate, who had already had a risk-reducing mastectomy but was holding off on her risk-reducing oophorectomy because she wanted to have more children with her second husband. In discussing her confidence that the mastectomy would protect her from breast cancer, she stated:

Oh, 100 percent. I guess from time to time, when I hear about my cousin going back in for a check-up, I have to re-check myself – like, "Oh, am I supposed to be doing anything? Oh wait, no, that's why I had it all done. I don't have to go." I don't have to check for lumps. I make sure that my gynecologist is aware that I have the [mutation], she knows about the mastectomy and she does checks. But during breast cancer awareness month, it doesn't cross my mind that I could get it. I think, "How do I let everybody else know about it?" I don't think of breast cancer as a possibility at all anymore. I know it is, still. I know there is always a risk because they can't get all the tissue out. But I don't worry about it at all.

Kate, 35

Other women also described their feelings after surgery as being not completely without risk, but at a level that was reasonable and livable. For example, Chris stated that after having both surgeries,

I feel really good about [my risk]. I feel like I've done everything I could possibly

do. I exercise, I eat pretty healthy. I keep my weight down, and I take my vitamins. I feel like I do everything. At first I didn't drink any alcohol or eat any sugar. It's like you try to just knock out anything that could increase your risk. But my God, you really have to kind of live your life! So I think my risk now is probably better than that of the average population. Knowing that my aunt has some sort of weird ovarian tissue in her uterus that has caused another cancer, I'll have to give that another thought. But for me, I know that I've done everything that I can do, or that I'm willing to. I do have my uterus. I'm not willing to take that out.

Chris, 33

Chris seems both to have a realistic understanding of her residual risk of cancer and to be coping well with what that risk means for her own life. Like Chris, Leigh seems to be in a sort of middle-ground territory with regard to her feelings about cancer risk after surgery:

Overall I feel like my risk is reduced. When I first woke up from the surgery, I felt a huge sense of relief that I didn't have to worry about breast cancer anymore. I think from what everyone tells me that my risk is greatly reduced now. I still have like a 5% risk so it's not nothing, but it's so much less than it was. So it's not going to be front and center anymore. And I guess that will be an adjustment in itself because I've been thinking about it so much for the last two years. So maybe that hasn't completely sunk in yet.

Leigh, 35

Finally, Audrey, 28, shared her perception, after having completed both RRBM and while contemplating RRSO, that having surgery had been a way to “take hold and do something to where I feel like I have a little piece of the pie that's leaving me in control. You know, ha ha ha! I'm going to get it before [cancer] does.” Each of these women has experienced an outcome that seems like the ideal relief from anxiety that many women are seeking when they act with agency and make the decision to have a risk-reducing surgery, freeing themselves from high cancer risk-perception and re-opening paths toward important life goals.

Chapter Conclusion

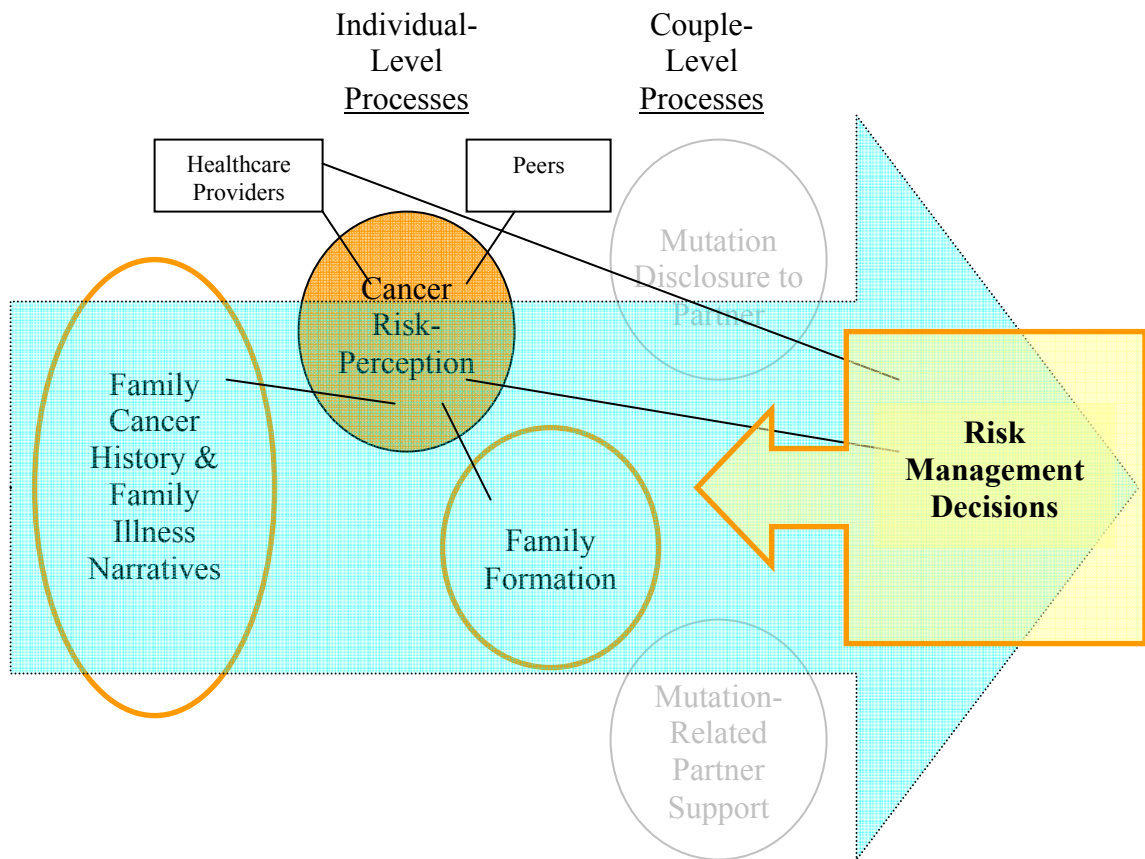
To some extent, the relationship between cancer risk perception and risk management decision-making in *BRCA1/2*-positive young women has been thoughtfully explored in previous research (e.g., Antill et al., 2006; Kash et al., 1992; Neise, Rachfuss, Paepke, Beier, & Lichtenegger, 2001; Schwartz et al., 2004). Believing that “living with risk [is] an ongoing experience which [leads] over time to a subjective development of one’s sense of risk,” (Pilarski, 2009, p. 305), qualitative researchers studying women at increased familial risk of breast cancer (but who were not confirmed *BRCA1/2* mutation carriers) have provided valuable insight regarding how risk perception is formed in this more nebulous situation. However, previous research has failed to richly and comprehensively describe how young *BRCA1/2*-mutation positive women gather information about their risk, think about specific elements of risk related to their unique developmental stage, and reconcile their risk-related emotional experiences with the demands of managing their mutation status, finding ways to act with agency in situations rife with constraint. The data provided here make a significant contribution to filling that gap.

Expanding upon the basic model proposed in Chapter 5, Figure 5 illustrates how breast and ovarian cancer risk perception is related to several other elements of the model. Data presented in this chapter illustrate how cancer risk-perception is powerfully shaped by carriers’ knowledge of their family history and family beliefs about cancer. Participants shared stories about how things they learned in interactions with healthcare providers and peers became prominent features in their understanding of their risk, as well as how beliefs held by providers shaped their understanding of which risk management options were available to them, with clear implications for cancer risk over

time. Finally, several participants touched on issues related to family formation and carriers' worries that moving toward their family formation goals might increase their risk of cancer.

Data indicate that in HBOC families, in which legacies of cancer are often pervasive and quite devastating, women may begin to consider themselves "at risk" very

Figure 5: Role of Cancer Risk-Perception in Theoretical Model



early in their development, and so experiences that contribute to one's ongoing evolution of risk perception may occur across the life course and may be experienced as quite constraining. When this lifelong experience occurs, women appear to reach the point of

genetic testing with firmly held notions of what it means to be at increased hereditary risk of cancer, and learning about one's mutation status is often simply a confirmation of what is already known to be true. However, it may be challenging for women in this situation to set aside their intense identification with the cancer experiences of their mothers, aunts, grandmothers, and sisters and their often strongly held beliefs about being "doomed" to meet the same fate. Participants whose family histories were saturated with stories of cancer-related illness and loss sometimes reported feeling overwhelmed, overburdened, and as though difficult diagnoses were an inevitable part of their future. Such feelings may preclude the ability to feel in control of one's health because the threat of cancer is seen as ever-present, and messages that suggest that periods of very high risk are not immediate may be difficult to absorb and assimilate. This is consistent with early research by Chalmers, Thomson, and Degner (1996), who suggested that early in the development of cancer risk perception, the lived experiences of family members are the primary source from which women draw information. Furthermore, research attending to the family illness narratives present in cancer-affected families suggest that the concept of anticipatory loss plays heavily into the experience of being *BRCA* mutation positive (Werner-Lin & Gardner, 2009), and living with this sense of impending significant loss appears to be managed at least partially through behaviors that demonstrate agency, such as seeking and assimilating information from various sources and creating concrete plans for risk-reduction.

In the absence of information about how family members have been affected, some women are left with an even stronger sense of uncertainty about the meaning of their *BRCA* mutations. Participants whose mutations were inherited through unaffected

fathers, who did not have close relationships with the affected members of their families, or who simply did not have a strong family history of cancer, often reported feeling as though they had little personally applicable information on which to base understanding of their risk. This sense of being suddenly thrust into high-risk status can be alarming and leave women feeling as though they have only the advice of their physicians and peers upon which to rely in understanding their risk; the threat of impending loss becomes even more ambiguous, and risk-management decisions can seem impossibly confusing.

Whichever of these two routes women take to understanding their cancer risk based on family history, working through presuppositions allows one to take a step back and consider personal cancer risk in the context of her own body, previous risk-related experiences (*e.g.*, abnormal screening test results), and the historical context in which her at-risk years occur (*i.e.*, at a time when science and medicine have advanced far beyond what was available to their mothers and grandmothers). It is in this context that participants frequently reported having relied heavily on information gathered from healthcare providers, who have a great deal of influence with regard to the choices mutation-positive women make about risk management. In fact, healthcare providers are often the very first sources from which mutation-positive women begin to gather objective, personal risk-related information, often in the context of genetic counseling. The frequency with which participants related a perceived inability to pay close attention and truly absorb information in this setting immediately upon learning that they carry a *BRCA1/2* mutation suggests that providers should consider temporally separating test disclosure from discussions about the implications of the mutation. In order to provide the maximum benefit to patients, providers should be aware of the different ways in

which their *BRCA1/2* mutation-positive patients take in and retain information; for some, this occurs through repetition; for others, having something concrete to take away from a genetic counseling appointment is key; for still a third group, a useful metaphor provides continuing feelings of certainty about one's understanding of risk information. Providers should be conscious of these differences and attempt to deliver information in the way that works best for each individual patient. Connecting patients with resources such as FORCE, Bright Pink, and existing publications, is another way for providers to direct their patients toward appropriate support; however, ongoing in-person support from a knowledgeable, supportive healthcare provider should be available to all mutation carriers as they move through the ever-changing challenges presented by their mutations.

Perceptions of variations in risk related to specific mutation (*BRCA1* or *BRCA2*) different cancer sites (*i.e.*, breast, ovarian) and developmental stages (*e.g.*, during pregnancy, breastfeeding) were a major source of cancer worry for many participants; further biomedical research providing insight with regard to these variations in risk would decrease ambiguity for women in this group if, for example, better ovarian cancer screening techniques were available, or breast screening during lactation were reliable. As more individuals are identified as mutation carriers and their natural history is studied, the medical community will come to understand more and more about how to counsel women about managing their risk. In the meantime, learning to listen to the unique and intricate stories of today's young women, and how *they* make sense of their risk given their own knowledge, life experience, and interpretation of both, is key to supporting and assisting them through the ongoing decision-making processes that come along with being high-risk, congruent with recommendations by d'Agincourt-Canning (2005).

Among participants, it seemed that those with the most desirable outcomes related to assimilating and making decisions about risk and risk-management were those who effectively came to see their breasts (and, to a lesser extent, their ovaries) as parts of their bodies that were not integral to their sense of self; that is, they came to understand that they are not defined by these body parts, bringing an easier acceptance of risk-reducing surgeries. Beyond this cognitive shift, some women are able to take a humor-based perspective on their risk, accepting that once they have done what is available to them to reduce their risk, the rest is out of their hands. In other words, they have found ways to overcome their situations and resolve some of their risk-related anxieties, but do not get caught up in needing to continually seek ways to *act*. Rather, they can take comfort in having done the best thing available to manage their risk. This seems to be the healthiest path to adaptation to the *BRCA1/2* experience, and would be an effective focus of ongoing psychosocial support if it were routinely made available to mutation-positive women in the settings in which they receive their mutation-related care.

Finally, it is important to note that risk-management should not (and in many cases, does not) end with surgery. Appropriate follow-up with physicians and genetic counselors is often necessary to help identify lingering feelings of anxiety or misunderstandings about risk, so that carriers have an opportunity to recognize and shift conceptual glitches that leave them feeling vulnerable to cancer. Ongoing screening for other gynecological cancers and a lifelong awareness of other *BRCA* related health threats are also necessary, but are far less intrusive and constraining than living with the very real and often constant threat of breast and ovarian cancer.

CHAPTER 7: RESULTS—MUTATION DISCLOSURE IN COUPLE RELATIONSHIPS

“I expected it to be a very difficult conversation and I expected someone would be turned off by it.” – Kate

Although *BRCA* mutations occur within individuals and are passed between biological relatives, their potential to impact and be impacted by couple relationships is both real and significant (Hoskins et al., 2008; Manne et al., 2004; Matloff, Barnett, & Bober, 2009; Werner-Lin, 2008b). Establishing, nurturing, and maintaining couple relationships are primary tasks for women and men in young adulthood (Arnett, 2004). When a female partner carries a *BRCA* mutation and is making ongoing decisions about how to manage her cancer risk, several additional relationship-related tasks become necessary. The presence of a *BRCA* mutation must be disclosed by the mutation carrier to her partner; this is often precipitated by a perceived need to discuss issues related to risk management. The timing and tenor of such discussions vary significantly with the longevity and depth of the relationship, and may be followed by a period of education (either by the carrier or through research of other sources) for the non-carrier partner. Couples already in serious relationships when the female partner learns that she carries a mutation in *BRCA* must adjust to a new phase of life in which the demands of managing the mutation must be consistently and supportively taken into consideration. Couples must establish bi-directional exchanges of information and support (both instrumental and emotional), and cooperate in decision-making about risk management. Ongoing discussions about the implications of a *BRCA* mutation for family planning and medical management may also become part of the fabric of these couple relationships indefinitely.

Disclosure in Dating Relationships

Informing one's partner in a dating relationship about the presence of a *BRCA* mutation was described as an intimidating task by many participants, and the manner in which this exchange of information occurs varies depending upon, among other things, a woman's choices about risk-management. Those relying upon surveillance may be disclosing, in addition to the presence of the mutation and risk to future children, the potential for future bodily changes as a result of risk-reducing surgery; however, because they have not yet done any risk-reducing surgeries, these women are able to choose when to disclose their mutation to a partner, thus having greater flexibility and control over the process. For women who have already completed one or both risk-reducing surgeries, disclosure often occurs earlier in the development of a relationship because of a belief that disclosure should precede any physical intimacy that would make the effects of the surgery obvious to the partner. This sense of obligation to disclose earlier in the development of a new relationship can be restraining to carriers if they feel unable to control how this highly personal and sensitive information is shared with a partner.

Timing of disclosure within relationship development. For many mutation-positive women in dating relationships, deciding *when* to inform a partner about their mutations is a delicate process. Factors that influence this decision include the perceived solidity of the relationship, an individual's desire for the relationship to become permanent, and what actions have already been taken to mitigate risk (*i.e.*, risk-reducing surgeries). In this study, the belief that disclosure of one's mutation should occur only after a partner has demonstrated commitment was widespread:

I guess I'd have to be really close to them. It would have to be a very serious relationship. It would not be something I'd tell somebody on the third or fourth

date. It'd be somebody who's already met my family, somebody who's already demonstrated commitment.

Noelle, 26

While many participants reported desiring some commitment prior to mutation disclosure, they also discussed the delicate balance between disclosing early enough to avoid being seen as secretive, while waiting long enough for the partner to feel sufficiently invested that simply ending the relationship would be less likely. As illustrated by Ellie in Chapter 5, Charlotte also believed that the timing of disclosure with a future partner could significantly impact the relationship trajectory. This derived from a previous experience in which a relationship had ended shortly after Charlotte and her partner discussed her mutation. Single when interviewed, Charlotte thought about disclosure to a future partner:

I have dated a little bit and not felt like I needed to bring it up right off the bat. But I think probably before it got too serious, if I could tell that it was going to probably be an exclusive relationship or that things were going in that direction ... if he wasn't OK with it – I experienced that before, I wouldn't want to be so involved that it was heartbreaking. So I think that I definitely would tell him before it got too serious, just in case it wasn't something that he was able to handle at that point in his life. And it wouldn't be so hard if it just wasn't the right time for the two of us, so maybe before it got too serious. I don't think it's something that on the first date I'd need to share, but as we're getting to know each other, I'd just kind of let him know my situation in case it progressed into a serious relationship. But I would almost feel like I was keeping a secret if I waited too long. I would feel like I'm not being fair, I guess.

Charlotte's plan to disclose her mutation to a future partner before the relationship had become serious may be a way to protect herself from the hurt she felt when her previous relationship ended unexpectedly after disclosure. Her acute attention to disclosing early enough to prevent a future partner from accusing her of being secretive—and before becoming too emotionally involved herself—is a testament to the gravity of this information. She also noted how dynamics in her family of origin, specifically with

regard to their worry about her, were related to the disclosure process with a future partner:

I think the hardest part definitely is, I feel like I would wait until I felt close enough with someone. And at that point I feel like they would care about me obviously and I think it'll go back to seeing my family worry about me, I feel like it'll be adding another person onto that burden. Almost that this new person's going to have to end up worrying about me when I don't want anyone else to. So that'll be hard.

Charlotte, 26

Charlotte's concern was connected to pre-existent guilt feelings that she experiences when her family members worry about her, a state of mind that may be exacerbated because, having inherited a *BRCA* mutation from her father and with a mutation-negative sister as her only sibling, Charlotte was the sole member of her immediate family dealing with a very high cancer risk. Although her father was a mutation carrier, his risks (as a male) were much lower. Her words suggested that disclosing her *BRCA*-related status was akin to inviting her partner into this group of worriers, a perception that may account for her deciding to delay sharing this information until further into the relationship.

In addition to balancing the status and demands of the relationship itself, carriers must also select a time for mutation disclosure based on what is happening in their lives with regard to the mutation itself. For many, involvement with surveillance or progress toward risk-reducing surgery may move them to disclose earlier than they would otherwise, as Ellie experienced (see Chapter 5). Trixie recounted how informing her partner about her yearly NIH visits, and the reason behind them, was a useful segue between telling him about the history of cancer in her family and about her own mutation:

I told him pretty quickly about our family. You know, I had pictures and I would talk about different members of my family. And then ... shortly after [I met him],

I had to go to NIH for my yearly visit. So that's when it came up to him that that's where I was going and why. He thinks it's great that I'm involved in something, that I'm managing my risk. That I'm not just sitting around thinking about my risk, I'm doing something. So he was very supportive.

Trixie, 27

Disclosure prior to risk-reducing surgery. Ellie and Trixie had some flexibility to time their mutation disclosure because they had not yet had surgery; consequently, there was no outward evidence of their status. For them and for other participants who experienced mutation disclosure to a partner before undergoing risk-reducing surgery, disclosure was often focused on conveying lifetime cancer risk and the challenges of ongoing screening and prevention, as well as the potential risk to future children. Many, but not all, participants anticipated at least one risk-reducing surgery as a likely event in their future. Accordingly, they felt compelled to disclose this eventuality to partners in new relationships before they become permanent, out of a sense of obligation to inform their partners about an important life challenge in which the partner would be expected to be involved (Hoskins et al., 2008; Werner-Lin, 2008). This feeling fit with women's conceptualization of the seriousness of the two surgical procedures, and suggested that they were aware of the demands that these procedures would put on their partners and on developing relationships.

Many participants who were not in relationships when interviewed gave thoughtful consideration to how to approach informing a future partner about their mutations and plans for subsequent risk-management. Isabelle lucidly outlined her strategy, which would depend upon the risk-management decisions she had made up to that point, as well as how the discussion would likely be influenced by her cultural background and that of her partner:

I am Jewish, Ashkenazi – there are a lot of genetic issues going on with our group of people. For all I know, he might have a history he wouldn't want to bring up, or would want to bring up at a later point. If he's overly caring and sensitive, it might just come up in conversation. If I'd already had a prophylactic mastectomy I would bring it up a lot sooner just to warn him what my breasts look like, but if I hadn't and I'm going for surveillance, I feel like he would want to know why I'm going to the doctor. If it was too much for him then it would be too much for him, or I would take it a little bit at a time, give him little tidbits and then if he stayed long enough and was still interested I'd give him the whole megillah.

Isabelle, 22

“The whole megillah⁷,” to Isabelle, was comprised of her family history of cancer (*i.e.*, her mother's two very early-onset breast cancers) and their implications for her own health; her risk-management plans, including both risk-reducing surgeries; the implications of her mutation on the timing and pace of family formation; and potential genetic risks to future children. For Isabelle, these latter issues were confounded by the threat of other Ashkenazi-specific genetic health risks (*e.g.*, Tay Sachs, Gaucher, Niemann-Pick diseases), which she thought might be present in her future partner's family history, if he were also Jewish. Given the higher frequency of *BRCA* in the Ashkenazi Jewish population, other carriers may be similarly concerned with the possibility of co-occurring genetic issues within a couple considering future family formation. Isabelle suggests that the appropriate way to handle disclosure to a future partner would be to share information in pieces, ensuring that he could handle each piece before giving the next.

Women's sense of confidence about how their partners will receive mutation disclosure directly impacts the experience of the disclosure itself. Recall Ellie's thinking (in Chapter 5) on informing her partner about her decision to undergo RRBM as indicative of strength, and her expectation of a positive reaction. In contrast, Annie

¹ Megillah: a Yiddish term for a long, boring, tediously detailed account.

recalled disclosing her mutation and long-term risk management plans to her long-distance boyfriend after they had been dating only a few weeks. In the same short period of time, she had met this partner and decided to move forward with RRBM, so disclosure in Annie's case meant informing a very new partner of a significant health-related decision by which he would certainly be impacted if their relationship continued. Annie's decision to disclose came from her sense that they were a "good fit" and had similarly positive ideas about moving forward together:

It was a total mess, to be honest. We'd been dating a little under a month, and he had come to visit and we had spoken about how we felt about each other, and I just felt like before we made a commitment to each other he needed to know what was going on ... So I just said, "Hey, this is something I want you to know," and of course I started crying immediately. He goes, "What's wrong?" and he wants to try to fix it and all of that. But it was very emotional for me, because there was a part of me that was very worried that he was gonna be like, "OK, this is a little more than I signed up for." But in the end, his reaction was one of concern about me; he wanted to make sure I was okay *versus* what that meant for us. It was a huge relief. It was almost euphoric ... like, "Did that really just happen?" ... He took it all in stride. But I told him, "This is what I'm planning on doing. This is how I'm gonna manage what this means for me," and he didn't offer an opinion. He just said, "Whatever you decide. It's your body and I'm here for you. ... you know what you need." So that part of it, not trying to insert his opinion, was great. He's in healthcare ... so I guess I was concerned he was gonna second guess the decisions I had already made, but instead he just had nothing but support for it. He's really special and I feel fortunate in telling him and his reaction about it, that that's the way it is. ... even though I bumbled through telling him and trying to explain it, but his having a different reaction would be very devastating.

Annie, 32

Annie's experience illustrates what seems to be a fundamental truth about disclosure for many young *BRCA*-positive women: they often genuinely fear that even a committed and seemingly wonderful partner will find the challenges presented by *BRCA* to be too much to handle, and choose to end the relationship, rather than facing the challenges together. Her recollection of the disclosure as "a total mess" reflected the level of emotional

intensity she experienced during the conversation, having found herself in tears at the start of the conversation because of her overwhelming fear that she would be rejected.

Disclosure after risk-reducing surgery. As previously stated, women who have undergone risk-reducing surgery while single face the challenge of informing new partners about their risk-management decisions early in the development of new relationships. Kristy believed that information about her mutation would be difficult to hide from a new partner:

I'm hoping that I'll just deal with it as it comes along for whatever comes up. It's not something that I think I would keep a secret for a long time, because it's had a major impact on my life and obviously I have a lot of family things that are going on that would be hard not to talk about. I think it would come up as a result of that. And because I've had reconstruction, when I start becoming physically intimate with somebody, it's going to be pretty obvious. Especially because I haven't had the reconstruction finished. So it's going to come up.

Kristy, 29

Kristy was in a relationship that has since ended, when she underwent RRBM. She has not yet had the experience of informing a new partner about her mutation and surgery.

Lynn was the youngest study participant who had already completed RRBM, having done so at 23. She was in a relationship when interviewed, and had informed several previous partners about her situation since learning that she was a mutation carrier at age 19:

I haven't really had any bad experiences with it. Like I said, I'm really open about most of that information, it's a take it or leave it sort of deal. If you don't like it, I can go meet somebody else. I'm not who I am because of some guy. I am who I am because of me, and if they can't understand why I've made a decision like this, then so be it, I'll find somebody else who does.

A strong sense of self, coupled with a straightforward attitude about her mutation and the changes it had brought in her life, were the keys to Lynn's open communication with partners. She believed that diving straight into it was the best strategy; in fact, she had

disclosed her mutation and information about her RRBM to her current boyfriend on the night they met! She believed that had she not disclosed this information to this new partner quickly, he would have soon figured things out on his own. When they met, she was fitted with tissue expanders (small pouches behind each pectoral muscle) that had small external ports implanted in each breast through which liquid was gradually added, until the desired breast size is achieved, and she had no nipples. When they progressed to a moderate level of physical intimacy, he would have immediately recognized that something was amiss. Unsure whether her current relationship would become permanent, Lynn went on to comment about disclosure in potential future relationships:

I think before I get married ... this is something that I would tell him right off the bat. Not necessarily the kids part, but the *BRCA* mutation, the statistics that are added to that, what I've done already, what I'm going to need to have done. And if the relationship moves forward, I'd mention, "This is something I have to do. I have to remove my ovaries. I don't need them for anything but having children. If I'm done having children, I expect to remove them, and all the lower parts." And it might put a little bit of pressure on the time table as far as having children, but hopefully that person will agree with me and think that it's a smart idea. Especially if I'm the person they want to have children with, they would be concerned about having me around to see the children grow up, and so forth. The more open I am about all of it, the more comfortable I am with discussing those things with people, and when you're trying to find your mate for life, your husband, you want to be open about those things. You don't want that to be a surprise, like, "Hey, we're getting married, but by the way..." I just don't want it to be one of those things.

Lynn, 24

Lynn's belief was that as a mutation carrier who had already undergone RRBM, both facts should be disclosed very early in a new, developing relationship. Other information, such as the implications for children and intention to undergo RRSO in the future, could be appropriately held back until the relationship had progressed and was more solid.

Not all participants found it as easy to be forthcoming about their mutations as Lynn. Kate was hesitant to share information with her new partner, after having gone through a divorce while she was undergoing RRBM and breast reconstruction:

I think probably the first couple times that we went out, the thought of telling him about that hadn't even crossed my mind because it had been so long since I dated. I was more worried about what to do on a date. But it wasn't very long after that I thought, "Well, I know I like this person, and we're going to probably continue to grow our relationship, I need to let him know about this." So I brought it up like, "Remember how I told you my sister had cancer? It was breast cancer..." and so I started to tell him all these things, and I guess I expected the worst kind of reaction, "Oh my gosh, you have *that* wrong with you?!" that kind of thing. But he said, "Oh really? My good friend had the same thing happen to her." So it ended up being remarkably easy. He knew all about the surgery, so that wasn't a shock to him at all. It was totally no big deal. I was stunned that it could be that easy. I expected it to be a very difficult conversation and I expected someone would be turned off by it.

Kate, 35

Kate's surprise at the ease with which her partner accepted news of her mutation and surgery indicate that her fears about his reaction were real. This positive outcome was partly facilitated by his familiarity with *BRCA* from his friend's experience; clearly, most women who disclose *BRCA*-related information will not be so fortunate.

Sense of urgency and pace of relationship development. The sense of urgency that some carriers feel to move forward quickly, to "fit in" all the desired tasks of young adulthood before their time "runs out" due to a cancer diagnosis or risk-reducing surgery, is another important dynamic that influences the tenor and tempo of couple relationships in this context. Charlotte was in a relationship when she first learned about her *BRCA* mutation, but that relationship ended shortly thereafter. Having been single for several months, Charlotte discussed her concerns about starting a new relationship for the first time with knowledge about her cancer risk:

When I first broke up, I was just kind of enjoying being single. But definitely now, I do feel a lot of pressure and just because I know my family wants me to have surgeries and things like that I do feel a lot of pressure and kind of like a ticking time bomb that I need to get married, have babies, so I can do this and not have to worry about it, definitely. I know that I feel emotionally ready to date again or be with someone again. It does almost feel like it's something on the to-do list, like it's something I need to do and get done. And hopefully I'll be able to take the time and really get to know someone that I care about and not just be doing it because I feel like, "okay, I need to get a boyfriend and get married and have babies and do this." So, I think it's definitely something that I just need to make sure that I'm not just doing because I feel like I need to get married, but because it's someone I care about.

Charlotte, 26

This stress is a real threat for young female mutation carriers – differentiating the feelings derived from their mutations from those that might be present if the mutation were not an issue. Charlotte's awareness of the risk of making rushed decisions based on needs imposed by her mutation is a positive sign that she can avoid this threat.

The sense of urgency to establish and make permanent a couple relationship may come not only from one's own feeling that time is short, but also from outside sources.

As noted by Charlotte, family members can be one source of this external pressure;

Serena also provided a good example of this:

I'm getting constant pressure from my mom, my dad, my grandparents, aunts, uncles, cousins. Everyone's like, "Who are you dating?", "Nobody," "When are you going to date somebody?", "When I find somebody." It's horrible. And it puts a lot of pressure. It's hard enough to date people when you're not under the pressure of "when are you going to find somebody, when are you going to get married, when are you going to have kids?" And it's become internalized as well. It doesn't make me happy and I know that about myself. And that is very frustrating because I don't want my life to rotate or depend of me having to find a guy. It is hard. They want me to get married and have kids. My brother is eight years younger than me and he's going to turn out a nephew any day now. My sister who is four years older than me has five kids.

Serena, 30

Unrelated medical conditions or challenges bring additional pressure on young carriers to progress quickly through couple relationship stages. Concurrent with discovering her

BRCA mutation, Monique learned that she was at high risk of cervical cancer⁸. This combination further reduced the perceived time available to have her family:

It's frustrating. I just want to get married, but I don't want to just marry anyone. I feel like I'm forced to do this. And then I step back and I'm like, uh... it's kind of frustrating, what to exactly do. You don't want to make the wrong decision. So I think definitely the cervical cancer issue made it even worse.

Monique, 26

Charlotte, Serena, and Monique all illuminate the experiences of young female mutation carriers who are not in relationships, although similar pressures exist for women who are in relationships as well: many experience a strong desire to make their dating relationships permanent. Noelle had been dating her boyfriend for several years and was ready to take their relationship to the next level, a plan that was not congruent with his timeline:

I think he's afraid of marriage, whether it's because he's too young, he owns a business so I know he thinks the whole business side of his life needs to be stable and perfect before he gets married. To me, there's never a perfect time. ... And so I really think he's focusing on proving himself in the business world before he can dedicate himself to marriage. And he has said that he thinks being married means you have to have a child right away, which I thought was the silliest comment ever. ... I was like, "Well, that works out perfectly for me and my situation here." I really think I'm the backbone of this relationship in that I can deal with a lot more and I just think he wants problems to be other people's problems, he wants kids to be other people's kids. That's not to say he's not the most excited friend or sibling when other people are going through these major life moments. That's what confuses me because there's such a disconnect between how excited he gets for other people and wants to blow off that himself.

Noelle, 26

While Noelle was frustrated by this impasse, Kristy recalled how knowledge of her *BRCA* mutation catalyzed her relationship with her boyfriend Spencer to move more quickly toward permanence than it might have otherwise:

⁸ Cervical cancer is caused by persistent infection with the human papilloma virus (HPV). It is not part of the spectrum of *BRCA1/2*-related malignancies.

In our case, I think in some ways it kind of accelerated our relationship because we were pretty newly dating when I found out that I had the mutation. And I went to this high risk center and everybody was asking me, “When are you having kids? You need to have kids early and have your boobs taken out so that you reduce your risk.” And at that point Spencer was very supportive, and it’s something that we worked through together. And it kind of got to the point of, well, I need to get married quickly and we got along so well, and we loved each other. In some ways I think that news accelerated our relationship. He was very supportive and it brought us closer together, but in the big scheme of things it probably had an effect that maybe we didn’t want.

Kristy, 29

Ultimately, Kristy and Spencer concluded that they were not the right long-term partners for each other. However, Kristy was emphatic that being with a warm, supportive and loving partner during her mutation testing and her RRBM was very positive: she truly needed him then. She has no regrets about their relationship; rather, she appreciated that Spencer was a part of her life during such a difficult time.

Beth’s experience further illustrates how women’s sense of urgency complicates their lives. Before she knew she was *BRCA*-positive, Beth and her boyfriend unexpectedly conceived a baby, despite consistent contraceptive use. One year post-partum, they were struggling to balance their desire to get married with issues related to Beth’s risk-reducing mastectomy and her sister’s ongoing breast cancer treatment. Plus, Beth felt compelled to complete her surgery before her son turned one, because her sister’s breast cancer diagnosis (which occurred one year after delivering a baby) dramatically exacerbated Beth’s sense of vulnerability:

Originally we were going to get married this summer, this was before my sister’s [breast cancer] diagnosis. My sister’s diagnosis kind of threw a wrench in that because now if we do a destination wedding my sister won’t be able to go. Then I thought we could still do a destination wedding and just have it be the two of us, because I’m thinking if my sister can’t go I don’t know if I want anybody there. So that kind of put a wrench in it. And then me having the mutation, knowing that I want to have surgery, I told him I need to have the surgery before my son is one year old because my sister was diagnosed when her daughter was 11 months old,

so that feels like a ticking time bomb to me. But a wedding is just not as important as the surgery and getting well. So then after my sister's diagnosis, and after learning about my mutation, we thought OK, she's going to have chemo, then she's going to have surgery in February, if I want to have my surgery before September we could still get married late spring or summer. But it just keeps getting pushed back because my sister's surgery was pushed back, it's hard enough to plan a wedding if you have an infant, but then throw cancer and the mutation into it...

Beth, 30

The urgency to accelerate making a couple relationship permanent was consistently articulated among participants who were in positive, stable dating relationships when they learned about their mutations. However, learning that one is a mutation carrier may have the opposite effect on women who are not confident that their relationships have long-term potential. Annie illustrated this beautifully; she was dating someone when she learned of her mutation, and chose to end that relationship soon after:

I got the results and shortly after that we broke up. It wasn't because of it, by any stretch. Well maybe, you know what, maybe it was, because I realized I was in a very casual relationship and, ultimately, I had a lot of much bigger things going on and wanted to focus a little bit more on myself, and wanted to find someone that I wanted to talk with about it and wanted to be involved in the process of it. Ultimately, I think that very well could have been a catalyst for me to realize, "I'm not really into this person and this isn't going where I want it to." That probably moved things to an end quicker than maybe I would have otherwise.

Annie, 32

Annie felt empowered by this decision, and commented that her experience might differ from other mutation carriers; rather than worrying that her mutation would make her undesirable to a partner, she instead viewed her mutation as a catalyst to making positive changes, focusing on herself, and positioning herself to meet a partner who was a better long-term fit.

Charlotte's experience with her then-boyfriend was similar to Annie's, although she did not describe it in the same way. Charlotte and her boyfriend had been dating

several years when Charlotte learned that her family had a *BRCA* mutation. That information was particularly difficult for her partner to cope with because a member of his family had a serious disability, and serious medical issues in general were a tough subject for him:

He very much backed away and just wasn't able to be a part of it and was pretty open that he didn't know if he could deal with another sick person in his life. Even though I wasn't getting diagnosed with cancer, the potential of having to deal with that was pretty hard for him, which was hard for me to hear when I needed the support. And so the testing process was pretty hard for him. He was one of the people among my friends who kept saying, "Your cousin will be negative. Your uncle will be negative. Your dad will be negative. You'll be negative." When it came down to my dad was positive and it was time for me to get tested, I asked him if he would come with me, and we actually ended up not talking for three days. He just said he needed some space and we were living together and he went and stayed somewhere else. And then he came back and he did apologize, but that's when he was open about how he was feeling with all of it. He did come to the test with me, but it was not the kind of support that I needed or was looking for. After that he felt guilty because we had dated for so long, and he wasn't able to be there for me. We had been growing apart after college and things like that. So we were just dealing with, "Is this because of this? Or is it because we're just growing apart?" So it was very on-again, off-again for probably about nine months or so, and then that ended up just ending.

Charlotte, 26

Collectively, participants' feelings of doubt about couple relationships and their experiences in disclosing information about their mutations and susceptibility to cancer are powerful illustrations of the intense emotional work that comes with the physical state of being *BRCA1/2* mutation-positive. They also illustrate how decisions about risk management can have a profound impact on when and how mutation information is disclosed in a dating relationship.

Fear of rejection or confirmation of commitment. It was a powerful emotion for several single subjects to feel that their desirability to the opposite sex was or would be reduced as a result of being a *BRCA* mutation carrier. For some, these beliefs stemmed

from doubts about a partner's willingness to commit to someone whose body will likely change as a result of risk-reducing surgeries or cancer, or to cope with the implications of a *BRCA* mutation on family formation (*e.g.*, risk to children, condensed timeline for family formation). For others, external factors, such as messages from family or societal beliefs about feminine attractiveness, were the source of doubt.

Many participants who were uncertain regarding how a partner would handle mutation-related information were not in relationships when interviewed, but had considered how partner disclosure might occur someday. For example, Lilly discussed how she felt as she started dating several months after positive mutation testing:

It is – pardon my potty mouth – fucking scary. It's not a diagnosis, but who's gonna want to deal with this? I mean, implants are fine and nice for strippers, but I don't know too many guys who want that. I don't know. There are just a lot of "ifs." As much as I try to think it's not, this is a big deal, and I don't know too many people who are gonna want to walk into this situation.

Lilly was already contemplating how a future partner might feel about her body after she eventually has risk-reducing surgery. In part, this feeling may stem from Lilly's own uncertainty about how her body would look and feel after RRBM; she also questioned the ability of a future partner to deal with all of the uncertainty that comes along with being a mutation carrier. She then discussed her belief that the demands of her *BRCA* mutation would not align with a partner's ideas about what young adulthood should be like:

I can't help but think that he'd walk. At 25, 26, 27, or even 31, 32, 33 it's like you're focused on wanting to get married or wanting to have a family. Hopefully someone would be able to look past [the mutation]. That's the route that I'm hoping to take. But in all honesty, if the tables were turned and someone said to me, "There is a strong chance of schizophrenia," I don't know how I would deal with that.

Lilly, 25

Equating the challenges presented by her *BRCA* mutation with those presented by a serious mental illness demonstrates the profound negativity with which Lilly fears a partner might view her mutation and its implications. This is a genuine fear for many young women. Nichelle, who was in a very casual relationship with a partner with whom she had not yet shared any information about her mutation or the presence of cancer in her family, illustrated this concern:

I think that could be one of the worst things is to tell someone that you've tested positive, and then they just wouldn't react well. Or them not being able to handle it and kind of removing themselves from your life. I think there's always a fear that it would limit a relationship. It would possibly end a relationship because of finding this out. A partner not being able to handle it would be really a devastating thing because if it's someone you really care about, and you tell them this, and they can't handle it and would walk out of your life...that's probably my biggest fear in regards to having this, just having a person walk out on me.

Nichelle, 20

Understanding Nichelle's deep fear that a partner could simply walk away, it is easy to imagine how difficult it was for Nichelle to consider sharing this information with the man she was dating when interviewed.

Some participants had theories about partner traits that might increase the likelihood of his successfully coping with *BRCA*-related information. Most theories centered on his having previously experienced cancer or other medical hardships in their families, giving them increased capacity for empathy. Lilly suggested that a partner who was also from an HBOC family might be a good fit:

After going to a lot of these [*BRCA* support] meetings, a lot of the women have sons and they say that their sons, after having a mother with cancer, are in fact different. It just seems like if someone has dealt with something heavy, then maybe they can deal with this. [Interviewer: So maybe someone who is also from a *BRCA* family would be a good match for you?] Perhaps ... then again, I don't want to double whammy a child if I opt to have one.

Lilly, 25

Lilly's quote also illustrates her belief that having breast or ovarian cancer in the family, especially when one's mother is affected, is a truly life-altering experience that can and often does lead to an increased capacity to deal with the difficulties that might arise for a couple in which the woman is a mutation carrier. She also acknowledges the heavy burden that could befall the child of parents who are both mutation carriers – the “double whammy” to which Lilly refers would be a child who inherited *two* mutations, a potentially devastating scenario⁹.

Participants' fears that their *BRCA* mutations might be more than a future partner could handle come not just from their own imaginations, but from their loved ones as well. Isabelle, Lilly's sister, shared her memories of their mother's concern about how negatively both women might be perceived by potential partners; this was a topic of discussion in the weeks leading up to their genetic tests. Isabelle remembered:

Going through the whole testing process, my mother's huge concern after us having the mutation was, “if they do have it how are they ever going to get married? Who's ever going to want to marry them if they have this huge possibility hanging over their heads?”

Isabelle, 22

Hearing this message from their mother during the time that they were deciding about testing was a negative experience, and doubt about their desirability as partners lingered long after learning that they were mutation-positive.

Some participants identified specific reasons upon which future partners might base their rejection. For Charlotte, the bodily changes created by risk-reducing surgery

⁹ Because of the rarity of *BRCA1* and *BRCA2* mutations in the general population (estimates range from 1 in 280 to 1 in 1000), it is even more uncommon for two randomly mating individuals to both carry such mutations (except for persons of Ashkenazi Jewish heritage, in whom the mutation prevalence is 2-3 per 100). Inheriting a *BRCA1* mutation from each parent may not be compatible with fetal survival. Inheriting a *BRCA2* mutation from each parent results in a totally different disorder - Fanconi anemia - a devastating disorder with susceptibility to cancer and diverse congenital anomalies. With any combination of two *BRCA* mutations, lifetime risk of *BRCA*-related cancers may be extraordinarily high.

were the biggest threat because of their incongruence with what she believed men would find attractive:

I think the biggest part will be the physical change and them having to deal with that. Having your breasts removed is a pretty big deal, I feel like, to a guy. That would be a pretty big issue in a relationship if they care about you, just having that worry that you could get cancer. I think that would be pretty hard for a guy.

Charlotte, 26

As a woman relying on surveillance for management of her cancer risk, it seems natural that Charlotte would view her own likely next hurdle – risk-reducing surgery – as the most likely threat to a future relationship. For Kristy, who had already undergone RRBM, the risk to future children seemed like the bigger threat:

It's hard to think about this because to me it's kind of at that point where it's just really not a big deal, but I know that for a potential partner it could be, especially considering that I have a 50% chance of passing it to my children. So I think that could be something that could be scary or difficult to deal with. I guess I just assume that if it's a problem with him, then oh well, move on to the next person. And if it's not, then great.

Kristy, 29

Acknowledging that a future partner might find it difficult to cope with the physical and psychological impact of a mutation on their children, Kristy had a realistic view of potential future relationship challenges. Her expectation that the “right” partner be willing and able to accept those challenges also seems predictive of good long-term coping and relationship health. This speaks to the other end of the spectrum: if the fear that a partner will leave the relationship because of *BRCA* is not confirmed, there is an opportunity for the relationship to be solidified by the partner's ability to prove his dedication to the carrier. This was true for Marjory, who was “a little bit nervous that [her boyfriend] would reject” her and who felt enormously relieved when he responded in an enthusiastically supportive manner that did not indicate any desire to end the relationship;

in fact, she felt that their relationship had become considerably stronger since she told him about her mutation. Similarly, Reina recalled her decision to tell her then-boyfriend (now her husband) about her mutation fairly early in their relationship partly as a way to ascertain whether there was potential for the relationship to become serious and/or permanent:

I guess I thought, I'm at an age now where I want to be in a serious relationship, something that could potentially be long-term, and if I'm going to be with somebody who is right for me, then I need to know that this is something he can handle. I need to know that this is something where this person is going to understand and be supportive because it's going to be a very big part of my life. So I opened up to him pretty quickly, and I don't think it's something that he understood fully but something that he was very supportive about, he said he could only imagine how difficult something like that must be. And shortly after that was my first [BI] experience, and he was there for me through and through, 110%. And I said, "Wow, this guy is a keeper." And even my mom was like, "He's somebody special." So I kind of knew, you know, "I think this guy might be in it for the long run." To deal with something of this magnitude after only having dated a short time, he was really there for me.

Reina, 29

Like Reina, Serena viewed the sharing of information about her mutation with a partner as a way to test the fit of the relationship:

He asked a couple of questions, just general ones, and I answered them and he said, "No worries. Sounds like you're doing everything you're supposed to do." He didn't make a big deal out of it. And if he had, that would probably have ended things right there because I'm not going to date somebody who's not going to understand; just like I wouldn't date somebody that didn't like my dog.

Serena, 30

Surprisingly, participant's fears about their partners being unable to cope with the implications of their mutations were generally not confirmed, and most relationships not only survived but were strengthened by the disclosure process. However, for a few women, relationships devolved in the period immediately following disclosure. Although the breakdown of these relationships was attributed to a variety of causes, the mutation

was always among them. It is possible that the extra stress of a *BRCA* mutation on an already faltering relationship may push partners past their limits and bring a swifter end to relationships that likely would not have survived anyway.

Disclosure to Spouse

The process of disclosing one's mutation to a partner is very different for women who are already married when they learn about their mutations. Because married partners have already agreed that their relationship is permanent, the threat that a partner will leave the relationship is at least partially mitigated. Close, intimate relationships between marital partners mean that mutations are almost always disclosed immediately, and usually long before any risk-management decisions are pondered or reached. Almost universally, participants who were married when they learned about their *BRCA* mutations involved their husbands prior to even having the genetic test. However, there was a diversity of experience among married female mutation carriers regarding how openly they shared other *BRCA*-related information with their spouses, and regarding the manner in which their spouses received and handled this information.

When interviewed, Rose had known about her mutation for six months, and was at home recuperating from her RRBM. She and her husband had made decisions about testing and risk-reduction together:

Everything I know, he knows. We're really open, really close. He's absolutely my best friend in the entire world. It's almost seamless. It's hard for me to even say what did I know and what did I tell him and at what point did I tell him things. It's just whatever happens in my life happens in his life, so it's what we did together.
Rose, 30

Chris's experience was similar to Rose's. She recalled that after she learned her results:

He of course asked [what the results were], and I told him. He cried. He's pretty emotional when it comes to me. I unfortunately make him that way. But he cried,

and he told me he was sorry. And I told him I just want it all off, I just want everything out, I want it off. I just was ready to be done with it all.

Chris, 33

For Rose and Chris, there were no barriers to exchanging mutation-related information with their husbands. Both couples functioned as teams in making risk-management decisions, and this was experienced positively by both wives. In contrast, Marie's husband's was less involved in her genetic testing decision and process. Working to navigate the challenges of her husband's own family history of cancer, Marie chose to take her brother with her to genetic counseling, leaving her husband at home. She later regretted this decision:

Not taking my husband to genetic counseling was a combination of me being stubborn and assuming that he was feeling a certain way, and him not really knowing exactly what the situation is. His mom has cancer and when he gets in emotional, illness-related situations, he really kind of shuts down. I was assuming he wouldn't want to be part of it because either he was going to freak out and shut himself off or get annoyed with the situation. I wanted a positive reaction from him and I was thinking, "If he doesn't give me the reaction I want, I'm going to be really hurt. So I'm just not even going to go there." The day I got the results, after the appointment when I came home and I sat down and went through with my husband what everything meant, he was totally shocked. He was like, "I had no idea it was this serious." I was very regretful that I didn't bring him to the appointment. I wished that he would have been there with me.

Marie, 28

Since her genetic testing, Marie and her husband have come to a better understanding about his desire to be involved in *BRCA*-related issues and her need for positive support.

For some of the married study participants, unique family and/or health situations created their own dynamics with regard to *BRCA*-related issues. Adopted as an infant, Jane grew up never knowing that her biological family had a high incidence of *BRCA*-related cancers. Only when she chose to contact the adoption agency as an adult (in an attempt to learn more about her biological family generally and not because of any

concerns about cancer risk) and was able to access records made available by her biological mother did she start to learn about the possibility of risk for herself – this information was a complete surprise to her. Suspecting that genetic testing might be appropriate, Jane consulted with her husband about next steps:

When I learned of information from the agency I immediately shared that with him. I think he felt sympathetic, but I don't think he felt super concerned about it. It wasn't alarming to him, it was kind of, "OK, this is informational, it's good for you to have," and that was about the extent of it. When I talked with my doctors to see about getting the test done, he put it more upon me that it was my decision to make.

Jane, 30

Jane disclosed in her interview that her husband's response was not as warm and supportive as she wished; although it was clear to her that he cared about her, she lamented that his leaving the decision completely in her hands felt isolating.

Like Jane, Elaine also expressed disappointment with her husband's initial reaction to news that she was mutation-positive:

I was so pissed at him. I usually work late, but I ended up coming home at like 10:00 and just walked in the door and said, "I ended up testing positive for one of those mutations," and he looked at me and said, "Oh, I'm so sorry." And I'm like, "You're so sorry?" I'm thinking, 'I'm going to die, aren't you sad, too?' I was quite upset. But he didn't realize what the risks were. He wasn't making it a "we" thing. He was very much removed and I felt like, 'oh my gosh, whoa, I'm your wife! Come on!' He had had a little scare with cancer a couple years ago and I felt scared then. And I felt like he wasn't empathizing at all! I felt like he was really removed. But then I told him right away. I said, "Maybe I'm PMS-ing or maybe I've just had a long day but your response really kind of hurt my feelings because it's not very personal. Here I am thinking I'm going to die and you're saying you're sorry, like 'Doesn't that suck for you?'" I was able to vocalize it right away and then he responded with, "OK, well, you've got to quit your job. I'll sell the farm." And I'm like, OK, let's slow down, you know... I have meaningful work and all.

Elaine, 34

Elaine's perception that her husband's initial response did not demonstrate enough concern given the gravity of the situation was similar to Jane's; however, unlike Jane,

Elaine chose to address this disappointment right away and to clearly define her expectations, allowing her husband to alter his approach to the situation. Offering to “sell the farm” (a second property they owned and planned to retire to) was his attempt to create a financial situation in which Elaine could stop working, slow down, and enjoy life and their children, rather than continuing to work at her demanding mental health job. Although this was not exactly what Elaine wanted, it was an effective way for her husband to demonstrate his understanding and concern.

Audrey’s journey toward awareness of her *BRCA*-related cancer risk was completely unique among study participants. Rather than learning of her risk because of a high incidence of cancer in her family, Audrey discovered her *BRCA* mutation after her young son was diagnosed with a rare genetic disease called Fanconi Anemia (FA). In a very small number of FA cases, the condition is the result of a child inheriting a faulty *BRCA2* mutation from *each parent*, as happened with Audrey’s son. Therefore, Audrey and her husband learned together in a genetic counseling session related to their son’s health that *both* of them were also *BRCA*-positive; each of them had one copy of an abnormal *BRCA2* gene. She believed that their reactions to their son’s genetic diagnosis were unique: there was no opportunity for blame or anger at a parent who was “responsible” for their son’s genetically inherited disease:

I think we just kind of looked at each other and went, “Huh. Go figure.” I know in most families’ situations it’s one or the other, and that wasn’t there for us to place blame and say, “Oh, well, it’s your fault. You’re the one who had it.” Not that it’s anybody’s fault anyway. But we weren’t left with any would-haves, could-haves, should-haves, maybes, because we didn’t know. And it was both of us, and it’s nothing anyone did intentionally. As far as our relationship, we don’t have anything to be mad about. We both have it.

Audrey, 28

Audrey felt that the fact that both she and her partner carried *BRCA* mutations eliminated any possibility for either partner to react negatively.

As a final example of spousal disclosure, Pauline's experience came at a very intense time in her life and relationship. One month before their wedding, Pauline and her husband received an e-mail from Pauline's father informing them that a *BRCA* mutation had been identified in one of his cousins, and that the next step was for Pauline's father to get tested. He told them that if he tested positive, then Pauline should also get tested.

Pauline and her then-fiancé had some time to ponder what this meant:

Once we realized we were going to need to wait until my father got tested, my attitude was to continue to worry and start acting like [my dad] had it and I had it. And [Graham] was really much more positive and optimistic, saying "Oh don't worry about it, neither of you have it, there's nothing we can do right now anyway until we find out, so it's not worth worrying." I was so convinced that this is my destiny in some way that I was sort of like, "No, we have to pretend like I have it because it will make it easier for us to be able to deal with it when I do." So there was a bit of tension there but not like any sort of thing that would escalate into a fight or a true disagreement.

Ultimately, Pauline's father and then Pauline herself received positive mutation test results. Looking back on that period as she approached her first anniversary and during the process of making a decision about RRBMs, Pauline stated:

It's made me very, very grateful that we found out about the mutation after we were married. Not because I think that he would have chosen not to marry me, and I've actually wanted to be really specific with him about this, he's been incredibly supportive of me. But I want him to be able to say if he's angry that he married a woman who has a genetic mutation, not that he's going to leave me. I know he's not. I can separate those two things. But if he ever has anger about these things I think it's appropriate to express, because I certainly have anger. If he says "It's not fair," I know that he doesn't like me less. It's simply the truth; it's not fair.

Pauline, 30

Pauline insightfully acknowledges that in committed couple relationships, learning about the presence of a *BRCA* mutation is not just significant for the carrier, but for her partner

as well. Psychosocial intervention targeted at helping both members of a couple process this, both individually and as a couple unit, would likely help all couple members to do just as Pauline suggests – express and deal with all of the positive and negative feelings, including anger and fear, that come along with confronting this complicated journey together.

Non-Disclosure

The decision not to disclose their mutation status, or to not share fully the consequences of their mutations, was deemed to be the best strategy for a few study participants, for reasons which varied from subject to subject. Sometimes, non-disclosure was simply a matter of circumstance, or a reflection of a participant’s progress toward assimilating the risk information herself. For example, Valerie recalled a previous significant couple relationship in which she did not disclose the presence of a *BRCA* mutation in her family or the fact that she was actively considering genetic testing:

I was sort of the lone biologist of anybody I knew, and he was an engineer. I just don’t think it ever occurred to him that because my mom had cancer, and my grandma, and my aunt and everybody, that I might too. And I really don’t think we ever discussed it. I don’t remember ever actually telling him that my family had been tested and that I could be.

Valerie, 29

Valerie went on to state that the end of that relationship was just part of a natural evolution in her life, and did not have anything to do with her potential status as a mutation carrier.

As a 20-year-old undergraduate student, Nichelle was involved in a casual relationship with a partner for whom she cared deeply, but she described their relationship as mutually “open,” and therefore not perceived as stable or potentially long-

term. She first attributed her decision not to disclose her status to the history of cancer in her partner's family, but this larger reason for not disclosing soon emerged:

I think the biggest reason I haven't told him is because, based on his family's history that has recently developed, it's like I don't want to burden him with that, finding out someone else that he's really close with could possibly have it. I think, at a younger age, it could be harder for people to process, especially if you're being told that someone else at a younger age does or possibly could have [cancer]. I don't know how he'll react, so I don't want to tell him. I'm a little afraid of doing that because I think that would be really hard if I did tell someone I truly do care about and he... [LH: So what would you need to happen in your dynamic with him to feel comfortable enough to tell him? What's missing that you need there?] I truly think that if we were in an actual relationship, I would be willing to open up to him and tell him that because I've been very open and upfront with other people about it. I just think that the fact that we're in an open relationship is what makes me hesitant. If we were in an actual one, I think I'd feel more comfortable and have faith in telling him that. It's a fear that he'll decide it's too much to handle and say, "I can't deal with this," and will just kind of disappear out of my life.

Nichelle, 20

Clearly, at the core, Nichelle's fear that her partner would be scared away by news of her mutation and its consequences is what kept her from sharing this information with him.

This fear had its origin in the nature of their relationship, which was not seen as "real."

Annie was described earlier in this chapter as having felt compelled to end a relationship she did not perceive as potentially long-term after learning about her mutation. Here, she described what it was like to be in that relationship and not share information about her mutation with her then partner:

It didn't even cross my mind to talk with him about it, because [the relationship] was still something so new, and it just wasn't serious in my mind, which probably should have been a sign. Rearview is 20/20, but he was very supportive when it came to my cousin and what was going on with her. I just never really took the time to explain it to him, or he didn't want to know, or I felt like maybe he didn't want to know. I think it kind of went a little bit over his head, and maybe that was because I didn't really want him to think about it too much.

Annie, 32

For Annie, the instinct not to share this information with her then-partner was an effective internal barometer about the quality and potential longevity of their relationship; recognizing her hesitation in sharing this information, she understood that her heart was not really in the relationship and that it would probably be best to move on.

Although she was not in a relationship when interviewed, Monique had definitive ideas about whether, when, and how she would tell a future partner about her mutation. Because she was also at high risk of cervical cancer, she faced the challenge of informing a future partner about two weighty pieces of information. Describing her reasons for keeping this information to herself in past relationship and her plan to take her time in disclosing it to a future partner, Monique stated:

It might change his outlook if I have kids with him. I don't want someone to think differently of me. I just don't even like talking about it. They just might think their child's going to be almost 50% likely to have the mutation, and some people just don't want that. Or even, just because I have it, am I different? No, but you know what I mean? If you tell someone that, you don't want to scare them away either. Some people just don't understand. I don't usually share that kind of personal information yet. Until I know they're with me for the long term.

Monique, 26

Monique's perspective on mutation disclosure was reminiscent of themes discussed earlier in this chapter; specifically, she doubted a future partner's willingness to continue their relationship given the mutation, and connected this worry with the threat to future children (something she worried about herself).

Even when disclosure occurs in couple relationships, some women are not particularly open with their partners about what the mutation means in their lives, or choose not to involve their partners in the myriad tasks and decisions that come after learning that one carries a *BRCA* mutation. Marjory recalled a previous relationship in

which she informed her partner about her mutation test results, but did not involve him in what came next:

He knew about the testing and he knew about the result. I think he knew when I had my appointments, but that was about it. We didn't really talk about it at all, probably because the relationship was deteriorating. We had a lot of other issues, so I think that just wasn't something that was a priority to talk about. I was doing fine with it on my own. I didn't really need a lot of support for that, and I think I avoided it because we were having issues, and if you talk about genetics, you're talking about family, and I think that's kind of why, as we got into the relationship, it didn't come up as much. We were talking about marriage less, and so I think I protected that information a little bit more. That's the kind of information I was only going to really talk about with someone who I could see myself having a future with, and I think there was a point when I was in grad school where I was questioning that.

Marjory, 30

Marjory's experience suggests that, just as awareness of one's interest in disclosing or not disclosing mutation status can be a barometer of relationship quality, the quality of a relationship is an important factor in women's desire to involve their partners fully in their mutation-positive experience. Marjory's current partner is not only aware of her *BRCA* mutation, but participates actively in supporting her through and helping her make decisions about the full range of *BRCA* related issues.

Finally, Sophie described the dynamic that had developed between her husband and herself, wherein she did not involve him fully in her *BRCA*-related management, but nonetheless reported receiving sufficient support from him:

I don't know exactly when I told him; it might have been a few months [into the relationship]. I don't go around telling anybody and everybody, but if the conversation is brought up I'm not one to really shy away from it. I think it just came up, and again, I don't think he really knows all that much about it. I've told him what I want to do as far as surgeries and everything like that, but I don't think he really knows that much. He's always interested in anything that has to do with me, and he's very, very supportive.

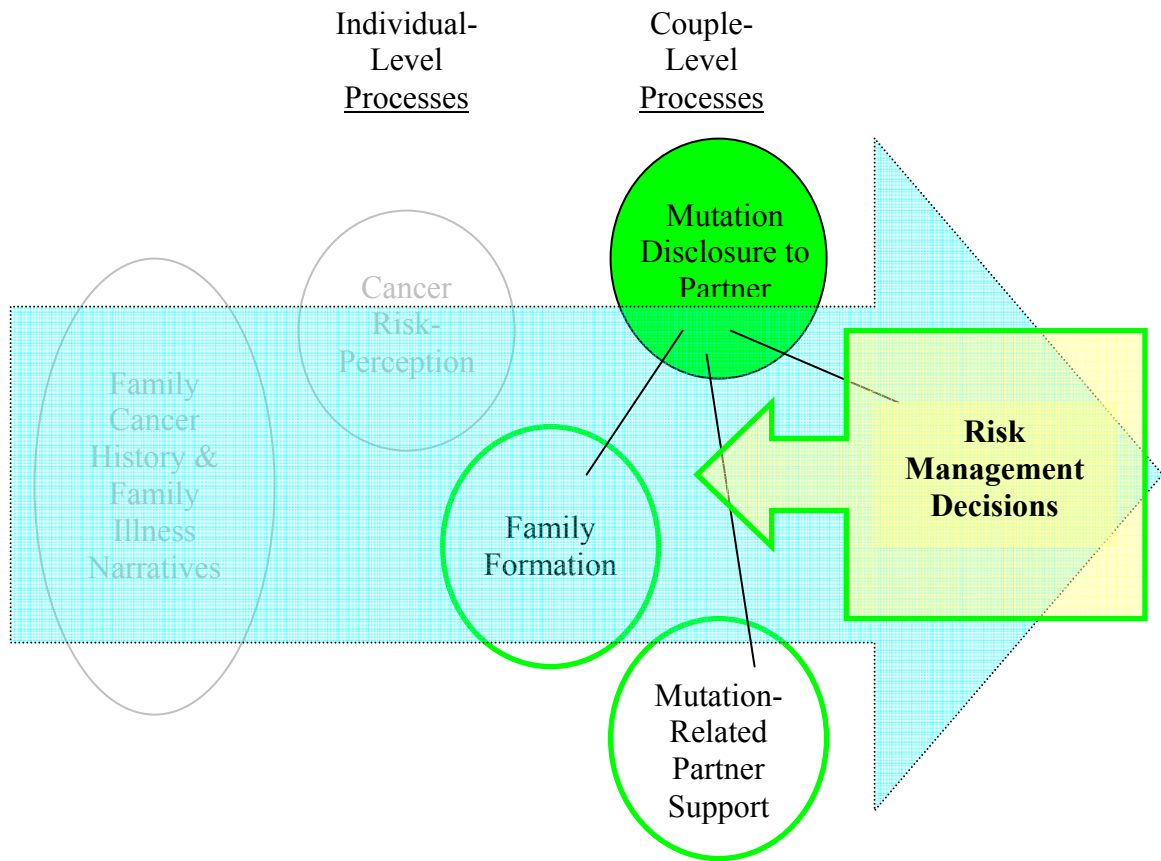
Sophie, 31

Sophie's more independent approach to her husband proved quite workable in the context of their relationship. Thus, dynamics related to exchanges of information and support may vary greatly between couples; while Sophie and her husband have developed a pattern in which he "does not really know that much," other participants perceived such relationships as unsupportive. Accordingly, many study participants worked hard to educate their partners about the meaning and implications of a *BRCA* mutation, to draw necessary support from their relationship partners, and to engage them in *BRCA*-related decision-making. These dynamics are the focus of Chapter 8.

Chapter Conclusion

Participants' reported experiences suggest that the process of mutation disclosure within couple relationships is closely related to mutation-related partner support, relationship quality, family formation, and risk-management. Often, mutation carriers in non-marital relationships disclose because of their belief that a partner's understanding about the issues related to *BRCA* will impact his ability to provide support and to make decisions together about family formation and management of cancer risk. Decisions women have already made about risk management are also highly influential in the disclosure process because having already completed surgery often forces young carriers to disclose information about the mutations to their partners very early in the development of new relationships, perhaps even before they feel comfortable doing so. For this reason, the disclosure process can look very different depending on whether or not a carrier has had a risk-reducing surgery (usually RRBM, since its sequelae are so much more readily apparent). Data further suggest that for women in dating relationships, a

Figure 6: Role of Mutation Disclosure to Partner in Theoretical Model



sense of urgency to achieve important life course goals of young adulthood, such as solidifying relationships and completing family formation, complicate the disclosure process. Carriers may fear that partners will reject them because of their mutation, but in fact the opposite was often true among participants in this study – couple relationships may be solidified by partners’ responding to disclosure in a way that makes carriers feel unconditionally loved, cared for, understood, and as though the partner is willing to participate in the ongoing challenges of the mutation positive experience.

For participants already married when they learn of their mutations, the process of partner disclosure typically occurred very swiftly, and spouses were often involved in the genetic testing process itself. In fact, many were unable to explicitly remember the

process of disclosure because it had happened so naturally; they could not imagine going through such an experience and *not* sharing it with their partners. For women in this group, thoughts about disclosure were often focused on whether or not spouses would meet expectations about providing appropriate support in response to the situation. When this did not occur, carriers felt disappointed and they varied in their ability to discuss their unhappiness candidly and openly with their spouses in order to elicit a different response.

Participants of various relationship statuses sometimes chose not to disclose, or not to share fully their mutation status and what it meant, for reasons ranging from perceptions that partners could not handle the information or that the relationship was not strong enough to bear such a burden, to carriers' own desires to be independent and handle mutation-related stressors without significant input from their partners.

Across participants, a personal focus on the next upcoming mutation-related challenge often created a sense of anxiety with regard to how a partner might respond. Once they had undergone a particular event or complication related to *BRCA*, and been forced to cope with it themselves, they often found it easier to envision that someone else (*i.e.*, a partner) would find it acceptable too. Not surprisingly, it may be that women find scenarios that they have not yet experienced as more threatening, both to themselves and their partners. Overall, however, these women display remarkable courage and strength as they negotiate one complex situation after another and, in the process frequently solidify rather than damage their relationships.

CHAPTER 8: RESULTS—MUTATION-RELATED SUPPORT PROCESSES IN COUPLE RELATIONSHIPS

“I just simply cannot imagine taking my shirt off for another man after I have this surgery, and I’m really, really glad that I have to only take my shirt off for a man who loved me before and who I know will love me after.” - Pauline

As young female *BRCA1/2* mutation carriers traverse the complicated path that begins when they learn that a mutation is present in their family, relationship partners are powerful sources of support. The type and quantity of support desired by carriers varies across individuals and over time. Participants who were in relationships identified various ways that their partners had succeeded or failed in providing support, and those who were single were quick to identify what they believed they might look for in a future partner with regard to his capacity to meet their mutation-related support needs. Interestingly, both single and partnered participants reported having considered the needs and desires of their partners in making decisions about risk management; for single women, this was done by imagining what a hypothetical future partner might need. Among participants, support was primarily viewed as directly related to the challenges imposed by a *BRCA* mutation, and as being provided by the male partner to the female mutation carrier. Some participants also discussed the ways in which they provided support to their male partners. Throughout their experiences as mutation carriers, many participants dealt with issues related to body image and sexuality, with clear implications for their couple relationships; having a loving and supportive partner was a key component of women’s ability to successfully manage these concerns.

Changing Support Needs over Time

As noted previously, most young female *BRCA* mutation carriers utilize a variety of risk-management strategies over time. For many, this means starting out with breast

and ovarian cancer surveillance, which may last from only a few months to several decades, depending on each individual's tolerance for the discomfort and uncertainty that accompanies regular screening examinations, and her willingness to take a more definitive step (*i.e.*, surgical removal of the target organ[s]) to reduce her cancer risk. After some period of time, many *BRCA*-positive women ultimately elect to undergo risk-reducing surgery. This decision is usually based on knowing that surgical risk-reduction is currently the most effective strategy to mitigate cancer risk. However, some women choose to continue surveillance indefinitely, and describe themselves as unwilling to remove healthy tissue from their bodies when they may never develop cancer at all. Regardless of women's ultimate decisions about risk-reducing surgery, chemo-prevention (*i.e.*, medications taken specifically to reduce cancer risk, such as tamoxifen for breast cancer, or oral contraceptives for ovarian cancer) may or may not be utilized as an additional risk-mitigating strategy. Across participants, our interviewees highlighted the role of partners, either current or future, in providing support with regard to, and making decisions about, cancer risk-management. Regardless of the different ways in which men provided support to their *BRCA*-positive partners, the recipients universally described their doing so as critically important. Opportunities to request and provide support begin when a woman discovers that a *BRCA* mutation is present in her family, and continue through and beyond the ongoing surveillance and/or risk-reducing surgery process. For this analysis, only support related to risk-management will be discussed.

Support related to surveillance. After learning that they carried *BRCA* mutations and deciding to utilize surveillance to manage risk, many participants reported finding themselves in need of support in the context of regular breast/ovarian cancer

screening. Factors contributing to surveillance-related stress include fear that a cancer will be detected, pain related to the screening procedures, delays in receiving test reports, and the frequent occurrence of false-positive test results, which require additional testing to resolve. This process is often accompanied by prolonged periods of ambiguity and uncertainty. Shortly after she began to date the man who is now her husband, Reina visited the NIH for an annual BI screening appointment. An abnormal finding on one of her imaging tests necessitated an extra overnight stay and CAT scan the next morning, which frightened Reina greatly:

I was on the phone with him and I was explaining to him what was going on, and he said, "I'm here for you, whatever you need, everything's going to be okay, you'll see," just very supportive words. When I came back home, he said he was so relieved and so happy to hear that the news was good. I mean, I still needed surgery but it wasn't cancer. And he came and visited me at my apartment with roses and hugged me and just said that he would always be there for me, that he couldn't imagine something like this happening to me, and he said, "I knew everything was going to turn out fine; I just had a feeling," and just very supportive in general. I think in my gut I was already feeling like he was the one, but this sort of sealed the deal.

Reina, 29

Her boyfriend's supportive response to this *BRCA*-related crisis not only helped Reina cope; it also gave her insight about his suitability as a long-term partner. Marie, who was already married when she learned that she was *BRCA* positive, benefited from her husband's willingness to come with her to her first round of screening appointments:

When I went for my MRI, I knew that it could possibly be a scary and he came with me for that. I told him I really wanted him to go, and he was like, "Are you sure? It's supposed to be real easy." And I'm like, "I really want you to come, so let's get a babysitter and come with me." And thank God he came with me to that, because I needed him for that, for sure.

Marie, 28

MaryAnn, 26, reported that her fiancé supported her by insuring that she adhered to her recommended screening appointments schedule, stating, "...he'll get on me because I

would always procrastinate before about going to doctors' appointments and going for my checkups. And I don't think he would ever let that happen anymore or let me get away with doing that."

In the previous three examples, partners of young *BRCA*-positive women were able to provide essential support during breast/ovarian cancer surveillance. However, support does not always flow naturally in these situations, and several women identified ways in which their current or former partners had been unable to meet their *BRCA*-related support needs. Jane described that her husband was unsure about what his role in her *BRCA*-related care should be:

I don't know that he knew what to do. He was supportive. He went to a couple of appointments with me that I had scheduled with the gynecological oncologist, and then also with the breast specialist. I think he felt really uncomfortable at those meetings. He didn't know what his role was or what he was supposed to do or say or to be. And I don't know if they know what to do with the spouse. He's there, he's present, but I don't know that they really made any effort to bring him into the process – they just didn't have any way of including him. They weren't rude or anything, but I could definitely understand that he felt that it was awkward.

Jane, 30

Jane's experience suggests that some physicians and other medical professionals who work with couples dealing with *BRCA* may not have or communicate clear ideas about how mutation-negative male partners can be involved in meetings, screening appointments, and other settings in which their female partners may require support.

Noelle noted a similar lack of fit between her needs and the support her partner provided, but for an altogether different reason: her boyfriend's apparent lack of concern for the ongoing management of her cancer risk through surveillance:

I would want him to say, "We can get through this." Right now I feel like it's my thing. It's my family thing. If he were to say, "Let's do this together, what can I do?" or even ask, "Hey, I know you had your blood drawn last week, did you get those results?" He doesn't follow up. Or he could say, "I know you're going in for

your MRI, I remember you saying last time that it was wrong or annoying or whatever it may be, is this going to be like that this time?,” just asking questions. And he doesn’t want to ask questions.

Noelle missed the sense that her partner was truly interested in her problem, and that they would face its challenges together. Instead, he maintained emotional distance by not demonstrating interest in her surveillance. She went on to say:

I haven’t asked him [to go to any appointments], but I thought that he would say, “Do you want me to go with you?” I have girlfriends here who are like, “Could I go with you? What can I do?” He’s never done that. It’s interesting because sometimes I go to our cancer center here in the hospital, and sometimes I see people and it’s just so upsetting, just to look around in the waiting room, and then other times I go and it’s like any other doctor’s office. So it really varies on how my appointment goes, whether I feel he should have come with me himself. But I have not asked him. I’m waiting for him to volunteer.

Noelle had begun seeing a therapist to cope with the stressors related to her mutation status, and had recently found her sessions focusing on “how [the mutation] plays into my relationships.” She had repeatedly invited her boyfriend to come to therapy with her to talk about how her mutation had impacted them, but he refused. Her focus in therapy had thus shifted to

learning how to deal with him and how people handle information differently. ...I’m learning ... to communicate in styles that he’d be responsive to. But I wouldn’t say it’s made our relationship stronger or brought us closer together. It’s taught me lessons on interpersonal communication, but that’s really it.

Noelle, 26

The impact of breast/ovarian cancer risk-management on sexual relationships comprised another important theme among women relying on surveillance. For Reina, this manifested as a serious emotional hurdle that arose when she and her husband became physically intimate. She first noticed this after a particularly difficult screening appointment, during which she had to have a painful and humiliating ovarian ultrasound.

Since then, they had been struggling to come to a common understanding about this issue and to communicate effectively about her experience:

I've had issues with my body, sort of like a disconnect. It's something I've been working through slowly and he's been incredibly patient with. But there have been some fights this year about it and I can understand from a man's position that it's very hard to truly identify. That's been one of the hardest struggles for me. I don't think he'll truly ever know why that happens or what I feel during those moments, but through many, many conversations I think he understands a lot more than he initially did. I think he automatically thought there was something wrong with him, or something wrong that he was doing, or a lack of attraction to him, but through going to therapy and learning more about why I have these issues and how I can work through them, I've been able to communicate to him that there are so many different levels of reasons and explanations that have to do with this. It's very deep. At one point, I had told him that it's easier for me when I'm in control of the situation, when I can instigate, when I can have the control of when things happen because the experiences that I've had with the gene and the NIH, I've had absolutely no control. And I've explained to him in detail the physical aspect of the screenings, and what has happened in my experiences, and how I was touched, and how I felt and how I was naked, and all of these different things. I just try to be as open with him as possible in the moment if emotionally I'm having a difficult time and he can tell, it's obvious, and he'll ask me to talk about what I'm feeling and it's very difficult, because sometimes I just can't express it.

Reina regretted that she was missing out on having a “normal, healthy” sex life, as she imagined other women her age experience, and communicating with her husband about how to sustain a healthy, mutually enjoyable sexual relationship had become an ongoing struggle. Because Reina did not view risk-reducing surgery as a viable option, these intimacy-related challenges will likely persist while screening continues, unless she is able to successfully disconnect one from the other. Reina said of her current sexual relationship with her husband:

If it's been a little longer period of time since the last time we were intimate, the automatic pressure is there whether it's spoken about or not, I kind of know that at some point sooner or later something has to happen. And I know now that he's not going to instigate, so I'm always mentally struggling with being able to get myself in that place where I'm OK with doing it. And it's hard for me to stay there for a long period of time because it's almost like forcing myself to only

think about the experience of being intimate in a positive light, only focusing on the feel-good part of it, allowing myself to connect with my body and allowing it to feel good. And it's getting easier and it's getting more frequent for me but it's still a major struggle. The issue is not him; it's not touching him, it's not anything having to do sexually with him. It's just me and my body. He wants that closeness and that's what's hard for me.

Reina, 29

As Reina continued to struggle with this issue, her husband's consistent and gentle support and understanding allowed their relationship to continue to function healthily despite the hurdle presented by issues of sexuality.

For women in established couple relationships, the experience of effective partner support during surveillance-related challenges was positive, but not without challenges. Marjory noticed how health-related communication with her partner had shifted since they learned that she was *BRCA*- positive:

I'm a little more hyper-aware of things, like if I'm feeling bloated or just not feeling well, nauseous, or any of those things, he jumps on those more readily than I do. So I think maybe I hold back a little bit, and I don't always tell him everything until I know for sure, because I don't want to worry him. So that's been kind of hard, if maybe I feel something in the shower, knowing I'm going to be getting it checked out in a month, I won't maybe mention it to him. Whereas if it was something like a headache, of course I'm going to tell him. But because this might actually mean something, I don't want to jump to any conclusions until I have to. I just don't want to put undue worry on him.

Marjory, 30

Marie also noted new dynamics in her marriage as a result of ongoing communication about mutation-related issues:

I feel like it's been more challenging than anything because it's just something that's on my mind all the time and I feel like it takes over sometimes. And talking about it too much can definitely be pretty irritating for him, just because he is thinking about all these bad things that can happen to me.

Marie, 28

Other participants noted how being supported by a partner through surveillance had significant positive effects on the couple relationship, as was the case for Melanie (see

Chapter 5). For example, MaryAnn found that working through *BRCA*-related issues together with her fiancé reinforced her already strong belief that he was the right partner for her:

I think it's had a positive influence. Every time you go through something really intense with somebody else, I mean, I just think it's been positive. I realize he will really be here for me no matter what. All in all, I think it's made me realize how fortunate I am to have someone like that.

MaryAnn, 26

As long as *BRCA*-positive women choose to employ surveillance for management of their breast and ovarian cancer risk, they will likely continue to encounter the anxiety and uncertainty that accompany regular screening. For some participants, this had become a consistent feature in couple relationships as well:

I think it's always on the back burner. We have our good weeks and we have our bad weeks. When it gets closer to screenings I get stressed. When I've had a very intense therapy session with my therapist, things get a bit more stressful with us just because a lot of these sessions that I have with my therapist doing a lot of this PTSD treatment, it's emotionally wearing, almost like I'm not myself for the next day or so because my mind is just exhausted.

Reina, 29

Repeated, long-term involvement in preparing for, doing, and following up on each intervention contributed to some participants feeling like a burden to their partners. Jane, who previously discussed her husband not having a clear role when he accompanied her to screening appointments, talked about how his reaction to her reporting about screening visits led her to feel she was imposing on him:

When I go in for screenings – and maybe I'm partly responsible for communicating it to him so much – but I just went in for a mammogram this month and it does make me anxious not knowing what the results are going to be, especially now that I'm thirty, it's something that weighs heavily on my mind. And I think at times I feel guilty to ask him for support or to really tell him how I feel because I feel like I'm making a big deal out of nothing. I feel like I'm burdening him.

Jane, 30

Like Jane, Reina reported that bringing the negative experience of *BRCA* to her husband made her feel badly, which might be related to her own anger regarding her situation and a way to shift her own feelings onto him:

I think instead of it just being a burden on my shoulders, it's a burden on both of our shoulders. In some ways it makes it easier, in some ways it makes it harder; easier in that if he can help me carry that burden a bit more then it makes it a little easier for me, harder in that it's something that is going to personally affect him because of the decision he made to be with me for the rest of his life, something that he could have chosen to avoid and leave but decided to stick through with it. I think that in many ways he tries to stay very strong for me because he knows that I struggle with it deeply and if he shows any type of negativity or weakness about it then he kind of feels like he's not being as strong and supportive as he should be. But it must be very draining for him to be like that all the time.

Reina, 29

Both Jane and Reina were relying on surveillance for risk management, so their support needs were focused on managing the anxiety and uncertainty of their regular visits for mammograms, MRIs, transvaginal ultrasounds, *etc.*, each one of which contained the potential for a new cancer diagnosis. Their feeling like a burden during this phase of their management raises serious questions about their partners' ability to cope with the more demanding and difficult scenarios related to risk-reducing surgery or a cancer diagnosis.

Dealing with the ongoing uncertainty of surveillance often necessitates a real shift in organization and expectation for both partners. In cases where this does not happen effectively, real damage to the relationship can be a result, as Ruby reported:

I think knowing that cancer could be my future, I probably focused more on the children, and my relationship with them, making that a priority in my life. My husband was working a lot, and I was really giving more to my relationships with my kids because they're so cherished. It's kind of hard. My relationship with my husband is kind of on life support. ... Our relationship kept taking a backseat to his work and the kids and the stress of life, and then this. And in about February, he wanted to call off the marriage. And two weeks ago, we went and retained divorce lawyers and then he decided to go to a counselor to see if this is really what he wanted or not. It's just a shock because your husband is the one who's said that he would never leave you, and him saying that he wants to call it quits is

new to me. He's coming up with a list of resentments this weekend, but I can only imagine that as I was focusing more attention on the kids than I was focusing on him, he probably felt not valued and he shows that to me, and then I feel not valued and I show it to him, and it's a cycle. And gradually things start to happen that wouldn't have been okay early in our relationship, like not talking for a day or two, then that goes to three or four days, then five days and then a week, and then two weeks and then, a month.

Ruby, 34

Ruby was moving toward RRSO when interviewed, and wondered whether her husband would continue to provide critical emotional and instrumental support during that process. She recognized that she might have to "supplement that support elsewhere" if their marriage came to an end and he was unwilling to continue in a supportive role for her.

Support related to risk-reducing surgery. For some participants, discussions about partner support naturally focused on the time surrounding their risk-reducing surgeries. Partners were able to provide support to carriers in making the decision to use surgical risk reduction; in preparing for surgery and on the day of surgery itself; during recovery in the days immediately following surgery, and during potentially lengthy reconstruction processes; and in moving beyond surgery, both physically and emotionally. Pauline provided a useful example of how her partner became her primary support at the very beginning of her journey as a *BRCA* mutation carrier. They learned that there was a *BRCA* mutation in Pauline's paternal bloodline just before their wedding, and learned that her father was positive shortly thereafter. Pauline quickly began to consider the possibility of early RRBM, because she knew herself to be intolerant of risk. She described her new husband as the person with whom she spoke most frequently and whose opinion she regarded most highly, stating:

I basically started thinking of myself as positive the day my dad got his results and I learned more about my cousin's approach to things, which means I started having conversations with my husband about risk reduction even before we knew the test results. It was sort of like an insurance policy – if I'm not positive, then I can just forget all this information, but if I am, I'm not a lost individual. My husband was a little reluctant to go there. He said it wasn't worth thinking about or talking about before we knew the results. And I would say to him, "But just in case, we should probably start getting comfortable with this topic." And it turned out to be a good thing we did because we were able to hit the ground running when I actually got my results.

Pauline, 30

Pauline's husband had continued to provide this crucial support over the next several months, as she moved toward and through her experience with RRBM.

Making decisions about surgery. For participants who were single when interviewed, thinking about a partner's involvement in or opinion about risk management was a theoretical exercise and sometimes burdensome. Sisters Lilly and Isabelle, both single, spoke about how they envisioned discussing their desire to have an RRBM with a future partner. Their mother had had bilateral breast cancer, first diagnosed at age 25; this made cancer risk an immediate/specific, rather than somewhat distant/vague, threat for both women. Although their family history was identical, Lilly (the older sister) was more eager to pursue RRBM than Isabelle. Having turned 25 shortly before her interview, Lilly's desire to pursue surgery in the near future far outweighed any concerns about what a future partner might think:

I just [decided], "I want to get my breasts removed." You know, I'm not married, I don't have a boyfriend, and I can't spare my breasts assuming that I'll find a guy who will have a problem with implants, or that I won't be able to breastfeed. I can't make these decisions without having anything concrete.

Lilly, 25

In contrast to her sister, Isabelle planned to delay any risk-reducing surgery until after she had completed childbearing. Her primary concern regarding future partner's involvement

in her surgery decisions were focused on how she would “sell” the procedure to her future partner:

I have pretty big boobs to begin with, so I have a feeling I’m going to have a guy who’s going to like boobs. And [after breastfeeding] I would jokingly say, “You know what, they’re droopy now. If you want me around for our kids’ future, and if you want a new set of boobs on me, this is that we’re going to do.” I can’t see myself becoming the type of person who would not do it because he doesn’t want me to. It’s my body, my life. I’m going to do what I want. But I would definitely make sure that he knows everything and bring him along to doctor’s visits. I don’t know if I’m hoping for this Prince Charming, idealistic guy, but I’d like to think that somebody like that’s out there who would care enough about me to know that this is what I have to do, and I think at that point, because I’m that much older, and if I haven’t had anything until then, I have an even greater chance of developing breast cancer. So I would think that he would be able to understand that. Like, “I have to get this done even more so now. I was going to do it in my twenties, but I decided not to so that I could have my breasts for our children, and now that I had them and they got put to use I would like to get new ones so that I can live.” I know that there are so many different [reconstructive] options, so if he wants it one way as opposed to another, I would look more into that.

Isabelle, 22

As a final example of a single participant, Charlotte shared that among the characteristics she would seek in a future partner, the ability to provide strong, consistent, and definitive support was paramount; this would make him a unique support provider among Charlotte’s many very worried family members, whom Charlotte did not always find helpful in making tough decisions about how to manage her cancer risk:

I would definitely want him to be supportive, probably just because my family is so emotional about it and so worried. I’d kind of look to him more to be a rock or a solid person in my life that I could go to. I think definitely because there are so many options out there and I tend to be indecisive by nature that I’d want someone that I could really talk to about different options and go to appointments and help me make decisions just because making decisions so permanent can be hard. So I think somebody that could be kind of a stronger person for me.

Charlotte went on to describe how her opinions regarding the timing of risk-reducing surgery timing should be communicated to a partner so that he could share his input about whether and when that should occur:

I think also having to tell them about the decisions I'd have to make like having my breasts removed and eventually, after I have children, have the oophorectomy and different surgeries like that, just basically that I'll be physically a different person, which is something that is a choice I feel like they'd get to make a little bit at that point.

Charlotte, 26

Partnered participants reported varied ways of involving their partners in decision-making about risk-management. For some, a belief that decisions about one's body should be made autonomously led them to convey information about what they had already decided, rather than asking for a partner's input. For example, Reina felt strongly that risk-reducing surgery was not an option she was interested in pursuing, and told her husband so:

We had a conversation a while back about having my breasts removed, and I told him right off the bat, "That might be the right decision for some women; it's certainly not the right decision for me." That was one of the reality-type conversations that we've had about how to potentially tackle it head-on. I told him, "It's not for me; I can't have surgery to remove a very large part of my body just because one day, what if." He supports any decision I make. He really does.

Reina, 29

Similarly, Shannon discussed her sense that she should inform her then-boyfriend (now her husband) that she intended to undergo RRBM at some point, so that he would know that surgery would be a part of their future, if they got married:

I did share with him when we were dating that I had already decided to have the prophylactic mastectomy someday. I wasn't quite sure when. But when I knew our relationship was getting serious, I wanted to make sure he knew that information and that that wasn't going to be an issue for him. And he was totally supportive and it wasn't an issue at all. And that's stayed true. He's actually, in the past two years, he's wanted me to move forward with this probably quicker than I have myself.

Shannon, 31

Interestingly, Shannon demonstrated that although her informing her partner about her planned RRBM was initially structured to give him a passive role, he subsequently

became more actively engaged and is now even more enthusiastic than she is about moving forward with surgery.

Trixie had also already decided to have surgery, but was pleased to find that her partner responded in an ideal fashion to both the news and the reality of her RRBM:

He stood with me through the whole decision-making process about my next step. He had no input into the surgery because I already had my mind made up, but he has been so great in reassuring me that it's me and not my breasts that he wants. And I think because he's older, he has a different set of priorities, where a guy my age who I would meet at a club might still be more interested in my breasts than me. I think I was seeking somebody like that to be with.

Trixie, 27

Trixie illustrates that some women seek support and input from their partners regarding the specific type of reconstruction they will select, described as her “next step.”

Dawn involved her partner in the decision between traditional implants (a relatively simple reconstruction procedure wherein a saline or silicone prosthesis is placed behind the pectoral [chest wall] muscle to simulate natural breast tissue) or the more complex autologous TRAM (transverse rectus abdominus myocutaneous) or DIEP (deep inferior epigastric perforator) flap procedures (wherein skin, fat, and muscle from the abdomen is used to reconstruct one's breasts). The latter procedures are more difficult, complicated and expensive, and require a longer, more painful recovery. Despite this, some women choose these procedures over traditional implants because the reconstructed breasts formed from one's own body tissue look and feel more like natural breasts, and age in concert with the rest of one's body (breasts reconstructed with synthetic implants tend not to age at all) (Djohan, Gage & Bernard, 2008). Dawn recalled her husband's position about the RRBM after she received an abnormal screening result and felt compelled to move forward with surgery:

When they came back with the ultrasound results and there was something there, he said, “You probably need to have this done. I don’t want to worry about it. They want to repeat in six months, that’s way too long.” So he was definitely with me on that one. And he’s been great, especially with the plastic surgery. He was like, “I don’t care if you don’t even have the reconstruction, I just want you to be here.” So I think that helped me make my decision just to get the implants just plain and simple. No 18-hour surgery to have all this plastic surgery done from my stomach.

Dawn, 27

For Dawn, the belief that her husband would accept and be happy with whatever choice she made allowed her to set aside her fears about having “fake” breasts and move forward with the simpler, less painful reconstruction process. She was scheduled for mastectomy exactly one month after her interview.

Maelie reported that her husband left the decision-making about whether and when to have an RRBM up to her, but was invited to give input about her reconstruction:

He really wasn’t involved in the decision-making about the surgeries. We talked about it, and he kind of listened and it was, “Well, whatever you want to do to take care of it.” He did have some say in the reconstruction part of my breasts, as far as if I was going to have reconstruction, what they were going to be, how they would look. Just because that would really be for him anyway. And he gets the brunt of the side effects.

Maelie, 33

Rachel’s husband took a different approach to post-RRBM reconstruction:

He said, “It’s your decision. If you want to do reconstruction, do reconstruction. If you don’t, don’t. These are your decisions that you need to make.” And that’s an advantage that I had being that I was with him for 13 years and that we already had some obviously strongly laid foundations.

Rachel, 33

Rachel reported feeling that her husband’s approach was a positive experience for her.

Their “strongly laid foundations” underlie her confidence that her husband will love and accept her no matter what choice she makes.

Wanda also noted her husband's powerful influence in her risk-management decisions. Having reached full agreement about whether and when she would pursue RRSO, Wanda and her husband were still not quite on the same page about RRBM:

I think we're both pretty certain that I'm going to have my ovaries removed once we decide we're done having kids. As far as the mastectomy, we're still talking about that. I feel that the screening methods for breast cancer are much more advanced so the chances of catching it before it became something big might outweigh having the mastectomy. He's like, "Just take it all out. Get new ones made. It's good. It's fine." But that's a much more major surgery, as opposed to the oophorectomy which is almost a same day procedure in some places.

Wanda, 32

Kristy recalled how conversations with her then-boyfriend Spencer helped her feel confident about and move toward RRBM:

Spencer's really who I had the most conversations with about that because I felt like it impacted him a lot, too, in terms of breastfeeding our kids, and what I would look like, and things like that. And he told me flat out, "I want you to have it done. I don't care about you breastfeeding our kids, I just want you to be around to be a mother to them." So I obviously knew that he wanted me to have it done.

Kristy, 29

Grace provided one of the most powerful examples of a partner's influence over risk-management decision-making. Having been in the BI study for four years during her mid-twenties, she and her husband Jason had decided that as soon as the study was done, they would try to have a baby. After she breastfed, they would proceed with the RRBM before trying to have additional children. This compromise would allow Grace to experience breastfeeding one time, but would not delay her RRBM more than two or three years. Shortly before implementing this plan, Jason had a change of heart:

He came to the decision before I did. He and my mom had gone to Race for the Cure while I was out of town, and I guess being there without me it sort of hit them, the reality of thinking about starting a family and what if something were to happen to me in the next nine months? So I got back from my trip and he sits me down and says, "We need to have a talk. I think the plan needs to change." And he was really sweet about it but I'm like, "Why are you being so sweet and yet

giving me this crappy news?” And so he gave me all of these reasons that it made sense to do the surgery before having a baby, and he could help feed the baby at night and we wouldn’t have to worry. There were just a million reasons he listed. The next day I met with a social worker and the breast center here at the hospital, I was hysterical, I was bawling my eyes out all day long. She really helped me validate that even though I agreed with all of his reasoning, and it wasn’t like he was pushing me into the decision, it was just that he was a couple steps ahead of me in feeling more willing to accept it. And even though I knew I wanted and needed to accept it, I just wasn’t quite there yet. She helped me figure out how to just grieve about it and take the time to accept it and know that it’s the right thing.

Grace, 30

Ultimately, the two proceeded with Jason’s plan, and she became pregnant a few months post-RRBM. In retrospect, she feels confident about her choice and thankful that Jason foresaw that this plan would work better for them and was willing to work to bring her around to that perspective.

Rose was interviewed during her recovery from RRBM, which she had decided to have prior to her first pregnancy. She highlighted her husband’s role in reminding her of their shared priorities of family and health, especially when she found herself doubting her choice and wondering, in the immediate pre-surgery weeks, if she should delay the RRBM until after she was done having children. Her husband’s consistency allowed her to remain focused on getting through the RRBM so that they could put the stress of her breast cancer risk behind them before they tried to have their first baby:

He was really supportive, and the part where I would stumble would be, “Well, wait, maybe we should have kids and I’ll do something later.” I think some of those stops along the way I was trying to convince myself otherwise, when deep down he knows how I think and that at the end of the day I want to be healthy and he wants me to be healthy. He supported the mastectomy from day one. There was no discussion really for him. It was like, “This is the right thing to do.” He did a ton of reading and anything I wanted to read, he wanted to read. And he’s always been so proud of me being part of that early detection program, so anything that was being proactive is something he’s the biggest cheerleader for. His attitude is, “Do it. Do what you have to do.”

Rose, 30

Although Marjory saw herself as the lead decision-maker with regard to her surgeries, her partner's opinion was highly valued. After independently concluding that proceeding with RRBM was the best choice, they worked out the specifics as a team:

For a long time, I didn't know if I wanted to do [the mastectomy]. And then probably within the last year and a half, I'm almost 100%, I'm about 95% sure that's something I want to do. So at that point, I had to involve him and let him know that's what I was thinking. For that particular discussion, he was already there. It would have been harder for him to understand if I wanted to continue to screen, rather than have the surgery. In his mind, it was a no-brainer, like "of course you want to have the surgery, why would you not want to do that?" So, we've been talking about that. And we've been planning on when I am going to do it. We have appointments to meet with plastic surgery and general surgery and I don't even remember who all else. I have a book that I'm reading, and then he's going to read it. And we're planning to do it maybe this fall.

Marjory, 30

Sadie's decision to have an RRSO at age 33 came after a long, difficult, and ultimately unsuccessful attempt to have a baby (the details of her struggles with infertility and family formation decision-making will be discussed in Chapter 9). Once it became clear that a biological child was not in their future, Sadie and her husband began to discuss whether RRSO was an appropriate immediate next step. This decision was difficult for Sadie because she was worried about the long-term effects that this hormone-altering surgery would have on both her physical health and her body image; she equated menopause with being fat, old, and ugly, and worried that she would be seen this way after having her ovaries removed:

My husband was really supportive through the whole thing, and he was there and listened to the argument. He said, "You know what? I want you to be around. I want you to be here and we're not going to be having kids. We might as well just go forward. I want you to be around for a really long time, and if that means having these surgeries, then that's what we do." And his argument was that a lot of people have their ovaries removed, his mom had it done and my mom had it done. He pointed out that they're not fat, or ugly, or old, and so he was just really supportive through this whole thing. I ended up having the surgery about six weeks ago, and through the whole thing, I think for him to actually have one of

the surgeries done has been a big relief for him. Because for him, it's like, "Well, that part of it is done, I don't have to think about ovarian cancer anymore." I mentioned before that we were talking about cancer and the diagnosis of cancer; I think it was in that conversation that it came up, and he said, "You know what, I'm really glad that you had the surgery because it's not something that we have to think about anymore." It's like a huge relief off of his shoulders. I think for him, surgery has been really positive and he's been really supportive about them.

Sadie, 33

Having gotten past one major risk-reduction challenge and feeling more confident that her ovarian cancer risk had been reduced, Sadie and her husband began to discuss whether and when she might have an RRBM.

In sharp contrast to Sadie's story, Leigh and her husband have struggled with regard to his supporting her. Leigh's attempt to engage her husband in her care is instructive, in that she reported his having made clear that his capacity to listen was limited:

Overall he would get tired of me talking about the topic after a while. Initially it was all-consuming after my mom found out her results, so I wanted to talk about it a lot and he got tired of talking about it within a week or so, and after that he didn't really want to hear too much more about it and I didn't share as much with him. Only if something really important came up, I would share it, but other than that, he wasn't someone who I talked about it with all the time.

For Leigh, her husband's inaccessibility led her to seek the needed support from friends and family members. It is unclear what kept Leigh from more emphatically stating her need for support to her husband. A useful psychosocial intervention for couples dealing with *BRCA* might be to teach them to express their needs and communicate more effectively – a common challenge for all couples that may be exacerbated by the stress of *BRCA*. In addition to this challenge, Leigh found it difficult to help her husband understand why she feels that an RRSO is necessary. Having recently completed her RRBM, Leigh was scheduled for RRSO shortly after her interview, and was hopeful that

after she came through it successfully, her husband would accept the decision she had made. For this couple, effects on Leigh's sexuality and implications for intimacy seemed to be primary concerns:

The last time we talked about it, which we haven't talked about that one as much recently because lately the breasts have been front and center, but he didn't really understand why I wanted to do that. I have explained my reasons to him, and I think he'll be OK with it, but I think we're both afraid that I'll be castrated like a cat or a dog. On the FORCE boards, most women seem to be OK with hormone replacement. So I'm hoping I'll be OK and if I think I'm OK, he'll be fine with it eventually.

Leigh, 35

Jane was also able to clearly identify support deficits in her relationship with her husband. Noting an overall lack of detailed communication about the implications of her *BRCA* mutation, Jane specifically noted that her husband's tendency to leave major mutation-related tasks and decisions to her felt unsupportive:

I think he wants to be supportive but I think guys kind of want to fix it, and this is something you can't fix. So he just kind of puts in back on me, like "Whatever your decision is I'll support you in that." I just wish he would take more of an interest in understanding what this mutation means specifically to me. You know, if he were to say, "I found this article and it said this, and what do you think about that?" it would make me feel like he really cares and wants to learn more about it. If he had been able to help me in some of the research. And one of the things that I said to him in the very beginning, it was so overwhelming for me to read thing after thing about women getting diagnosed so young and dying and very aggressive cancers. And I think that raised my anxiety a lot, and so if he had been more helpful in that piece of it, it might have helped me to feel more supported.

Jane, 30

In the process of working through the decisions to have a risk-reducing surgery, several women recalled having important conversations with their partners about how they would adjust together to their altered bodies after surgery. Valerie stated:

I've always had my boobs and I'd come to my peace with them. They were what they were and they filled out my tops or whatever. But he and I both had to start looking at my breasts and thinking, "how much longer are we going to keep these around for, and what are we going to replace them with, and how?" And we really

had some frank discussions about my breasts and how I feel about them and how he felt about them, and that's a conversation I'm sure many people do not ever have. I told him that one thing I would not put up with was being lied to, and it's helped me to know that he will give me the honest answer on this stuff. When I told him I thought it was time to do this and what did he think, he said, "Well, I'm thinking that I don't know what's bigger than a double D, but I'd like to find out." And we both laughed about it, and I think we do kind of approach things that way. Just as honest and straightforward as we can. We've just had to be that way about things that most people don't have to be that way about.

Valerie, 29

Acacia also sensed that her husband would experience a loss when she underwent RRBM, but highlighted how his continued involvement would help him cope:

We talked about it and I think his initial reaction, while he is very supportive, it's very scary for him and it's obviously the part of my body that you kind of have to say goodbye to. Obviously he understands the whole reason behind it but it is definitely sad, especially being a male and looking at a feminine body like that. But we've talked about it and I think he'll definitely go with me to the plastic surgeon and talk about that.

Acacia, 30

Beyond recognizing how issues related to body image and their own and partners' feelings about their altered bodies might pose challenges in couple relationships, several participants described how they had reconciled these and other issues related to sexuality with their planning and decision-making. Libby was also concerned about how the ways in which she might be different after a future RRSO could be difficult for her partner:

I'm going to have my ovaries out once I have kids, and that might pose some issues, just not having ovaries and stuff like that. I'm fine with it, but just afraid about the whole early menopause thing. He doesn't really have much view on it at all.

Libby, 32

Recognizing the likelihood that the changes to her hormone levels precipitated by the oophorectomy would pose some challenges in her relationship with her husband, Libby had talked to him about how to handle these. She did not describe his lack of a strong opinion as problematic, but the fact that they have not fully decided together about how

they will respond to changes to her mood, physical appearance, and sexuality could set the stage for significant difficulties in the future.

Pauline had given considerable thought to how her physical relationship with her husband might both be impacted by and be a comfort during her RRBM and recovery:

My status has materialized for me in this physical form as the removal of my breasts because that's what I'm choosing to do. That's obviously a very intimate and sexual part of your body. And I just simply cannot imagine taking my shirt off for another man after I have this surgery, and I'm really, really glad that I have to only take my shirt off for a man who loved me before and who I know will love me after. That simple act of disrobing, it's almost like going back to adolescence, being so timid about taking your clothes off or getting naked with a boy. We're going to have to rediscover ourselves, probably. He's going to have to re-learn about my body. I'm just really, really glad that I already know who the guy is that's going to be seeing me the first time I take the bandages off, and that's going to be my husband.

Pauline, 30

Pauline is not alone in being concerned about how her partner would respond to her body after her surgery, or in weighing this as part of her decision about the surgery itself. For example, Isabelle had already started to consider how having the surgery might impact a future sexual relationship, although she had only recently learned about her *BRCA* mutation and was likely years away from having either surgery:

The first support group I went to the topic of discussion was basically sex after cancer, and all the hormonal changes and issues relating to sex that go along with going through cancer or having your ovaries removed. And after that discussion, I'm hoping something better comes along, because a lot of these women were just not happy, and I know the general stress of marriage and sex is natural. It dwindles after a while, but for everybody's sake I hope they come up with something that doesn't put you through what these women are going through.

Isabelle, 22

Beth, scheduled for her RRBM, was concerned that she would not be comfortable enough with her own body to allow for the desired level of intimacy with her partner:

I'm going to have a deformed body for a while, but you know, I think when I was pregnant I didn't like him to see my body because it was changing and weird. So

having this surgery and the idea of being kind of deformed for a while is kind of [difficult].

Beth, 30

Marie also related her concerns about how her body would feel after surgery to her experience with pregnancy and childbirth:

I just think having the mastectomy and the reconstruction, I feel like I might have some... I can imagine that it'll be odd to be intimate after the surgeries have happened because, for example, I had a C-section with my daughter and I just remember that feeling of being cut open and so exposed and feeling like I didn't really have any control over my body. And then down the road, I guess I always assume that after I have the hysterectomy, oophorectomy, that it's going to affect my sex drive or something. So I think about that. It's kind of in the back of my mind.

Marie, 28

As demonstrated previously in this chapter, many young female *BRCA* mutation carriers in this group of participants relied on their male partners to help make decisions about the details of their risk-reducing surgeries. For others, their partners' reluctance about surgery resulted in the carrier deciding not to have surgery at all. Jane recalled her husband's position with regard to surgery shortly after she learned that she was *BRCA* positive; he asked her to take her time with her risk-management decision, rather than rushing into the risk-reducing surgery:

In the beginning, I had talked with my husband about a prophylactic mastectomy, and I think he was very put off with that idea. I think he could understand the reason why some women would do it, but was really concerned for me to make a decision so quickly after getting my results. He really wanted me to step back from it and not to make a decision that was totally based on fear, but that it was something 100% I wanted to do. And he kept saying, "Whatever you decide, I will support you, but you know, maybe you just need to sit on this for a minute and think about it." Which I think was good advice. Because if I had made a decision when I first found out and had acted on that by choosing a surgery I think I would have an incredible amount of regret now. And then with surveillance now he's supportive whenever I go in for my screenings. And when I tell him the positive news that nothing came up that was concerning, he's really happy for me.

Jane, 30

Jane's experience suggests that her husband's advice to slow her decision-making process and not make it purely based on fear has worked well for her in the long run.

Chris, who found her husband very supportive in her process of deciding *whether* to have risk-reducing surgery, became frustrated when he had different ideas than she did about the specific *timing* of her surgery:

He was extremely supportive. I mean, he's always been very supportive of whatever I've wanted to do. And he just wanted me to have it done. I wanted to have it done as soon as possible. It was like, "Get it done!" But we have a high deductible insurance, of course that factors in, and he really didn't want me to do it at the end of the year, so we would have to pay that deductible price. He wanted me to wait. And then he kind of started saying, 'well, the holidays are coming, just kind of enjoy the holidays, and then you can do it all.' And I was angry at him at first for that because I thought he wasn't understanding the importance and the whole thing. But in hindsight, that was the best thing, because I think going through all that would have been really miserable. So, he definitely encouraged me the right way. And he's always been very encouraging with that.

Chris, 33

Chris's perception that her husband's focus on having the surgery at a time when it would be most logical and convenient with regard to their insurance was unsupportive highlights the potential conflict that arises when a carrier finally arrives at a surgical decision and feels compelled to move forward as quickly as possible. Like Jane, Chris was able to see in hindsight that her husband's ability to see the bigger picture was useful. These participants provide useful examples of how members of couples in which the female partner carries a *BRCA* mutation can work together effectively (even if they do not feel as though they are doing so all the time) and provide balance and perspective in coping with the many challenges and decisions regarding breast and ovarian cancer previvorship.

Preparing for and experiencing surgery. Once they have made the decision to undergo risk-reducing surgery, mutation carriers are faced with a variety of tasks in preparing for and getting through the procedure(s). Undergoing risk-reducing surgery

brings substantial additional support needs. Surgery in this context is an intense and emotionally draining experience, which also brings significant physical limitations and reduced independence, especially during post-RRBM recovery. Bathing oneself, lifting almost anything, and even sitting up for any length of time are off-limits in the days immediately following the procedure. Partners again provide critical emotional and instrumental support through this part of the process.

Beth was working on scheduling her RRBM when she was interviewed, and she discussed how planning for and anticipating that event had become a regular feature in conversations with her fiancé:

If I talk about it he listens, he's just a listening ear. I keep asking him, "Are you ready for this?" Because it's going to be a big burden on him when I have the surgery and he's like, "That's just how it is." He had several knee surgeries over the past year and a half where I've had to kind of wait on him hand and foot, so he's like, "You did it for me, I'll do it for you." I hope he knows what he's getting himself into. I try to tell him that my surgery is a little bit different than his – it's a lot different. I'm not going to be able to lift our son, or help bathe or son or do a lot of stuff with our son, and he's going to have to do everything. But I think for the most part he just listens. He doesn't try to fix anything he knows he can't fix.

Beth, 30

Beth appreciated her fiancé's intuitive sense of how best to provide the appropriate level of pre-surgical support, *i.e.*, by listening, rather than trying to find and impose solutions to the unfixable problems. Rose's husband, however, played a more active role during her surgical preparations, discussing decisions with the various doctors involved in her care:

He was the one always asking the hard questions throughout the surgery, or even discussing the pre-op, he was the one taking the notes and asking very specifically, "Is this the day that we talk about which procedure we're doing? Will you do the nipple sparing? Is it going to be this kind of surgery?" And just asking some of those hard questions that don't seem real, but then the surgery's in a week, so you better ask them now.

Rose, 30

Pauline's husband had been an active participant in her RRBM preparations, accompanying her to appointments and demonstrating his determination that her pain be well-managed during her recovery. She expected a similar level of active involvement on the day of surgery and during her recovery:

I think that he will be one of the last people I see before I go under and one of the first people I see when I wake up. And I want it that way. I can't imagine going into this without him. I imagine that he will probably take some time off work to be with me and take care of me. I think that he will be really, really worried about me. I know he already is. ... his fears are seeing me in pain. So I think that he will be very involved. I think he'll see the dirty gross part of it. I think that he'll probably bathe me and do those sorts of things too. I think that he's going to be pretty hands-on, but I know I would do the same for him, and when we're old people I just hope he'll be bathing me or I'll be bathing him, too.

Pauline, 30

Pauline's husband provided another kind of instrumental support. Knowing that her RRBM was scheduled for early winter, "We planned a vacation for this summer so that I could take my boobs away one last time."

The period immediately surrounding surgery is one in which some carriers noticed negative impacts on their partner relationships. Some felt as though they were putting an unfair burden on their partners in expecting such intensive support. Preparing for her upcoming RRBM when interviewed, Libby stated:

It definitely caused a lot of pressure on the relationship, because now I'm having surgery. I have to rely on him for help. So that causes stress I think. I feel like maybe he feels like I'm a bad egg or something now. I mean, I'm sure he doesn't feel like that, but that went through my mind. I feel bad that he has to deal with it.

Libby, 32

Chris, having already had both surgeries, remembered what it was like in the weeks and months leading up to her surgery date:

Before the surgery, at times I was kind of angry. And so, I didn't want him to acknowledge my breasts or anything. It's almost like, "Can we just forget that they're there?" And especially when you're getting ready for the surgery. I have

always been really stimulated with nipples, and you're just not really sure what to expect after the whole thing. You kind of try to prepare and act like that's the way it is, like there's nothing there. That's kind of hard.

Chris, 33

Elaine was swiftly moving toward both risk-reducing surgeries when interviewed.

A busy working mom of three young kids, most of her free time was absorbed by thoughts about the surgeries and their impact on her and her family. She sensed that her husband did not understand the frequency and intensity of her mutation-related thoughts:

I think about it all the time. For me, it's just pervasive. We're walking by a tattoo shop and he'll say, "Hey, you want to go get a tattoo?" Well, I'll be getting my nipples tattooed. I'm thinking about it all the time but I don't think he gets that. I don't want to paint him out to be self-absorbed or anything, but I think that he really has a hard time empathizing, not just because he's a man but he's not tremendously close with his mom and so, even with my mom dying, I don't think he gets it. I don't think he gets how it can still affect me.

Elaine, 34

Elaine went on to speak eloquently about how her concerns about sexual dysfunction and changes to her intimate relationship with her husband played into her decision to pursue risk-reducing surgeries and her fears as they approached:

I'm especially concerned about sexual dysfunction. That's the biggest concern. It's already affected me sexually because my libido is so low since I learned. I think because I'm thinking about it a lot, and also life is just kind of hectic. You know, if he's touching my breast, it's like I'm thinking, "They're not going to be there." And my biggest concern about the oophorectomy is, "Oh, gosh, am I going to have sexual dysfunction?" I feel like, you know, we've got three kids. I nursed them all and was really affected with lactation and how it suppressed my own estrogen. I didn't get a period for a year. So that's probably my biggest concern is, "Is this what menopause is going to be like?" But I'm getting less worried about that. I mean part of it is that I really do feel like I don't have a choice. I know I do have a choice. I mean I'm making the decision, I don't mean it like that. But in my mind the choice is so simple, regardless of whether I'm going to have sexual dysfunction I have to go ahead and do this. And so, I'm worrying about it less because there's not really a point. It's not going to affect my decision. My mom's cousin who had the oophorectomy, she ended up having a little bit of hormone replacement therapy that was okay for the vaginal dryness and so she didn't feel like sexual dysfunction was such a big deal. So I'm just going to hope for the best I guess.

Elaine understood that her husband's attitude was one of apparent confidence that any changes to her desire for sex would be workable within the context of their relationship, as they were a small tradeoff for keeping her around:

He's just sort of like, "Well, let's wait and see and it'll be fine." He is not concerned about it from a selfish perspective at all. I think if we never had intercourse for the rest of our lives he's be fine. I mean I'm sure he'd be a little sad but he's really said, "This is no big deal, I'd rather have you alive."

Elaine, 34

Support during recovery and reconstruction. The length of the recovery and reconstruction process after risk-reducing surgery varies depending on which surgery a carrier has had and the reconstructive options she selects. Partners have an opportunity to provide support in a very close and intimate way during this period. Having recently completed her RRBM when interviewed, Valerie reflected on how integral her husband had been to her recovery, and pondered how difficult that phase of her life would have been had he not been there to rely on:

I really do feel awful for women who have to make these decisions, and go to all these appointments, and have the surgery, and everything on their own. Even just in that week after my surgery, I don't know what I would have done without him here to help me up and down the stairs and, you know, wash my armpits for me and everything. I mean, I'm sure if you don't have somebody, you get through it because you have to, but I don't know how I would have done it.

Valerie, 28

Because she chose to have her RRBM in another state, Rylan was touched that her then-boyfriend (now her fiancé) was willing to make the trip with her:

When I had my surgery this past year, he came up there with me to New York where I had it, and stayed with me the whole nine days I was up there, and also was able to not have to take any vacation time, he just worked while we were there. That was the reason he was able to go, otherwise he'd have to take all of that vacation time, and we would feel like it would have been wasted because we like traveling and vacationing a lot. So that was nice, and he was physically and

emotionally supportive of me when I was recovering from that. He never challenged any decisions I made, or he never questioned them.

Rylan, 34

For Rylan, the fact that her fiancé not only chose to accompany her during surgery, but also arranged his work schedule so that they could still enjoy a vacation together after she recovered, was a powerful expression of his love for her.

Trixie, having recently completed RRBM, reported feeling guilty at times because she believed “I’ve been such a burden to him for the past four months, but he assures me that that’s not how he’s feeling.” She discussed her appreciation of her partner’s willingness to ensure her comfort and facilitate a quick recovery after her RRBM:

He does everything in his power to make me feel comfortable, which is so nice. He’ll prop the pillows up for me if I’m uncomfortable sleeping and make sure that if we go to a bar, the stools have backs on them, he’s just completely focused on making me comfortable and getting me through this process as quick as possible.

Trixie, 27

Happily, it seems that for most participants, male partners did not experience these support needs as burdensome; rather, they were willing and happy to provide support to the women they loved.

Though Rylan, Valerie, and Trixie’s partners were vital to their surgery and recovery processes, other participants reported some conflict regarding how their partners would fit into the team of people providing support. Kristy had been dating Spencer for several years when she had her RRBM; although he was present, and eager to do whatever he could during her recovery, Kristy’s mother was also there and they sometimes disagreed about who should be responsible for which tasks:

My mom stayed for three weeks with me and it’s sort of like she and Spencer had to figure out their division of labor. I think that Spencer thought he was going to have more of a role because he was my boyfriend and he wanted to take care of me. But, for a lot of it I needed my mom to help me. And it caused a little bit of

tension in the relationship. I was very emotional because I was exhausted and I was on drugs. And we ended up sort of bickering a lot. I don't even remember it. I would get mad at him. He would say, "You did whatever, it wasn't very nice." I was like, "Can you just get over it? I'm obviously not myself. I'm on these drugs and I'm going through a tough thing. Can't you just sort of let it go?" So, in that way it created a tension and I think that overall it brought us together and we got through it, and he's been very supportive and he's still very supportive, but it definitely added some extra tension to our relationship.

Kristy went on to describe how the after-effects of the tension created by her surgery were the beginnings of the unraveling of her relationship with Spencer; they broke up several months after her surgery was complete. However, Kristy felt strongly that

it [was] something very positive, because if I wasn't in a pretty serious relationship, I'm not sure that I would have had the surgery. So in that regard I think things happen for a reason, and I think that was one of the reasons.

Kristy, 29

Kristy's experience suggests that the presence of a supportive partner, even when that support was not ideal, can be a powerful predictor of effective preparation and readiness for the challenge of undergoing a risk-reducing surgery at a young age. Because of Spencer's acceptance and encouragement of her decision, Kristy was able to feel that she was making a good choice; she moved forward with her surgery sooner than she would have without him.

Rose and her husband experienced some strong feelings about what it meant to them individually and as a couple to work through the process of reconstructing Rose's breasts after her RRBM:

Going through the process of mastectomy and reconstruction, it's this really weird thing about seeing yourself, "does this look good, does this look good," and that puts pressure on the relationship. But then there's this chance for rebuilding my body. I think it puts a lot of stress on someone to have to be like, "how does this look? Does this look good? Is this fabulous? Now I have big beautiful boobs. Isn't that awesome?" I can't imagine how hard it is for my husband sometimes. What's he supposed to say? When we're in the doctor's office he feels really awkward because he's talking to the doctor and he [worries that the doctor's thinking], "Is

he trying to get custom made boobs?” You’re going to have to readjust to the body image of your partner, you know? That’s a weird dynamic and something that doesn’t usually happen in a lifetime. I still sometimes wonder, I really want to know what he really thinks. I looked pretty good before, I was a very small-chested woman, just average, like a B cup. And I just wonder. I just sometimes wish I could crawl in his head and really know if he thinks this is even better? Does this look great? It’s one of those things you kind of wonder. I’ll never know.

Rose, 30

Rose’s experience illustrates a likely common challenge for women who are in relationships when they have RRBM (and reconstruction, for most): it is impossible to ever *truly* know what one’s partner thinks about the look and feel of one’s reconstructed breasts. There was a sense among participants that their partners were giving up the breasts that were present when these partner relationships were formed, just as the participants themselves were giving up their natural breasts; for both, this often required a genuine process of grieving and accepting the loss. While this uncertainty about one’s partner’s opinion may turn into an ongoing source of low-level anxiety for some women, it is likely that in the context of a loving relationship, a post-mastectomy female mutation carrier would be able to feel that her partner fully accepted and even embraced her new, less cancer-prone body.

Moving past surgery. Once surgery, recovery, and reconstruction are complete, women may face ongoing challenges coming to peace with their altered bodies after surgery; this is another opportunity for male partners to provide necessary support in helping carriers feel confident that they are still loved and that relationships are still secure. Participants provided powerful examples of this:

Sometimes we do have conversations when I’ll say I feel much more normal, or I feel back to myself pretty much. So it makes me a little bit stronger in some ways because I know that he’ll be with me through whatever. It’s almost a relief to think that he’s 20 years older than me, and I don’t look the same. I mean, my

surgery result was not perfect. And to think that I have a great husband who doesn't care, it makes me feel much better about that.

Chris, 33

Chris suggested that her husband being older made him less concerned with physical attractiveness and the sexual component of their relationship, and facilitated his accepting the changes created by surgery. Valerie also found comfort in her husband's acceptance of her post-surgery body, and highlighted how the presence of her *BRCA* mutation has added a new dimension to their relationship:

I think that sometimes I don't really want to lean on him. I try to be independent. And [*BRCA*] is one of those things that I get to lean on him for. So I think, to a degree, I'm not going to say he likes it, but I think he at least appreciates that I lean a little bit. I vent, I cry when I get the call that I have to come in for another test or a biopsy. And I let him look up stuff on PubMed for me sometimes. I don't necessarily let him do that for other things. I'm fairly independent. So I think he likes that I let him do that for me. And he's been very careful to tell me how much he likes my new boobs and that I'm still just as pretty as I was before.

Valerie, 28

Grace spoke powerfully about her husband's acceptance of her post-surgery physique; unique among participants, Grace elected not to have breast reconstruction after RRBM, at age 29. Shortly after her surgery, she became pregnant, and was several days past her due date when interviewed. She spoke lovingly about how her husband shows his acceptance of her newly breastless body:

I would say with the pregnancy – well, even before the pregnancy because I chose the risk-reducing option and had my prophylactic mastectomy almost a year ago, he's been totally just wonderful and supportive. Each night, being nine months pregnant and really, all through the pregnancy, he would lotion on my scars and most nights he'll kiss each side of my scar and kiss my belly each night before bed. And so there's a real sweetness about his acceptance of the scars. It's definitely made it way easier for me to accept it, knowing that he fully embraces it. That makes a huge difference.

Grace, 30

Grace's husband's unconditional acceptance of her appearance made it much easier for her to make the bold decision to decline reconstruction, which can be a long, complicated and frustrating process, without a guarantee of a cosmetically pleasing outcome.

Negotiating or renegotiating one's sexual relationship with a partner after risk-reducing surgery was a challenge many participants spoke about openly. For some, this task was experienced as relatively straightforward; for others it presented a major hurdle. Having started a relationship with a new partner several months after her RRBM and before her reconstruction was finished, Lynn remembered:

I was definitely nervous about it at first when we became physical and I did remind him. I was like, "Hey, by the way, I don't have any nipples, don't be freaked out by that." But he didn't care and it didn't change our physical relationship at all. The only thing I miss is the nerve endings in my breasts, the feeling. I do miss that.

Lynn, 24

Lynn exemplifies how well a young mutation carrier can cope with the bodily changes resulting from cancer risk management. Similarly, Kate and her partner, whom she met after her RRBM and reconstruction were completed (sans nipple reconstruction), communicated effectively about their feelings about Kate's post-surgery body:

He thinks [nipple reconstruction] is a waste of money, since it won't be a real nipple, why bother doing it? After we began our physical relationship, for a long time I would never take my shirt off in front of him. And he's never said anything. And I asked him one time, "Is it uncomfortable? Does it bother you?" And he said very honestly that he doesn't like to look at all, he doesn't like to look at the scars, but it doesn't make it impossible for him to see me naked. He just doesn't look there. So he does not have a preference whether or not I keep a bra or a top on. He just doesn't look in that area, and he said that works for him. For me, it's a little bit sad. I mean, this is the first time since I had my surgery where it makes me a little sad that I did have to have a mastectomy. It doesn't make me regret my choice, but it makes me sad that I had to choose it, because I miss – I feel okay about the fact that he doesn't look there, but I wish that wasn't an issue. In that physical sense, it's sad to me that I don't have my whole body.

Kate, 35

Their ability to have an honest conversation about her body and what her mastectomy meant to their sexual relationship was an important step in moving beyond any awkwardness that might have been present. His avoiding looking at her bare chest, and the loss of her breasts as a part of their sexual relationship are clearly sad for Kate, but do not impede their ability to have a healthy relationship.

In contrast to Lynn's and Kate's successful adaptation to their post-mastectomy bodies in the context of their sexual relationships, other participants have encountered more significant obstacles. Recounting her sexual experiences with her husband after completing both risk-reducing surgeries, Chris stated:

It's like a whole different ball game, intimacy afterwards. You don't feel your chest if you have expanders. I mean, how does he even get on top of you because of all the squishing? Plus, for me, I'm really emotionally high maintenance, and so, I really wanted to know that he was still attracted to me after the surgery. That's a big thing on the message boards that you'll see people talk about, like, "When did you have sex after the surgery?" So, we did -- I think it was ten days afterwards, I still had drains and everything. But I think it was comforting to know that he still found me attractive. After you have your boobs and ovaries out, then it like really takes it to a whole new level. You know, you don't even have the hormones there. And I can go for -- I mean, I was reaching my sexual peak, I thought I had been there for like two years, you know. So, you go from that, where you're like constantly begging him for sex, to "Oh, it's been a week? Well, okay." It bothers me because I like our physical relationship and I like to be close with him, but I could live without sex, totally. I could definitely live without it.

When interviewed, Chris and her husband were still struggling to connect intimately in a way that would work for both of them:

I don't realize when we haven't had sex in a while, and he doesn't say anything. Add onto that that he's 20 years older than me, so sex whenever you want to is not always going to happen. All the stars have to be aligned. So that's been a big conversation. You know, Oprah has this sex map thing on her website, and you're supposed to put numbers where you like to be touched and in what order. So I started having problems because I'm missing a hormone now. So I went on the Oprah site and we did the whole sex map thing, and we talked about it. I would have thought he would remember, but he doesn't. He just does his same thing. It's like he just acts as though I'm still the 32-year-old with raging hormones, instead

of the person who's 60 years old on the inside. I want to tell him, "You have to do some foreplay, you know?" It's been terrible. I even started on testosterone to help a little bit with it.

Chris, 33

Menopausal hormone therapy (MHT) was a frequent topic in post-RRSO discussions of sexual relationships. Participants commented often of tension between using MHT to alleviate surgical menopause symptoms such as hot flashes, vaginal dryness or lack of libido, and the fear that doing so would increase their breast cancer risk, thus blunting the protective effect of RRSO.

Participants who had completed one or both risk-reducing surgeries were able to reflect on their surgery experiences and observe its impact on the partner relationship.

Valerie believed that she had provided insufficient support for her husband in the preoperative weeks:

I think, in the weeks leading up to my surgery, I know that I checked out of our relationship a little bit. I really just was kind of worried about me. I was worried about getting all my work done, and getting all my ducks in a row, and getting everything I needed, and getting all my tests, and, you know, everything basically. And I know that I was kind of distant, but not really meaning to be. I wasn't necessarily pulling away from him, I just... I was always up over my head at work and everything. And after my surgery, he said that he was upset, but that he understood it needed to happen. But, you know, he kind of wished that we'd been able to have a few more quiet dinners at home, maybe a couple of romantic evenings because he knew I was going to be out of commission for a while. I do regret that in hindsight, not making more time for him and I right in that lead-up to surgery.

Valerie, 29

Valerie's awareness and insight about how the pre-surgery days might have been different may facilitate her responding more effectively in the future, *e.g.*, if she chooses to have an RRSO.

Several participants noted the process of coming to feel closer to their partners as a direct result of the support they experienced in the immediate surgical aftermath. Dawn

believed that her relationship with her husband had only been strengthened by her *BRCA*-positive experience:

Truly, this has brought us closer. I mean, we've had a great relationship and – not to be all gushy, but – he's my soul mate, and I think that if anything, it has brought us closer together and we talk more. He's really shown me he's there for me and so that has definitely brought us closer.

Dawn, 27

Trixie felt that her partner's post-op physical and emotional care helped bring them closer together:

I think it's brought us closer together. I don't see how you could go through this with somebody and not be ... I mean, it's not pretty right afterwards, the way I looked, and I was emotional. I wasn't crying but I was mean after my surgery. And he stayed with me through it and understood that it wasn't me talking, it was the trauma my body had just gone through, and just seeing somebody who's able to take care of me when it's not that fun to do so has brought us closer and made me respect him more, and made my family like him more. It's all been great.

Trixie, 27

Similarly, Rylan felt that she and her fiancé had bonded through the experience of her surgery and recovery, and that this helped to solidify their relationship not only to them, but in their families' eyes as well:

We trust each other that we're going to take care of each other, which is really important to his mom and dad. They are always telling us to take care of each other, no matter what. They've been married for thirty years now, and they want the same thing for him. I think this has kind of helped us do that better. It definitely makes you more health conscious when you're going through something like this, and I think not only yourself, but your friends and family around you, which I think is really important.

Rylan, 34

Looking back on her risk-reducing surgeries, Chris noted:

You kind of realize that obviously life isn't forever. And I think the biggest impact or the change that we had to our marriage was after I had the surgery and how much care he gave me, and you could just tell, really, how much he cared about me and how much he loved me because he was so doting, and he was so good with me.

Chris, 33

Kristy was unique in her discussion of how experiencing RRBM with her now former boyfriend had brought them closer in a way that transcended the end of their relationship:

Even though we're not technically together, it's something that has made us closer, especially with my family situation. Every week we have something. My mom calls and tells me about my aunt and it kind of gets to the point where with my friends, it's kind of hard to tell them about this every week. I kind of get the feeling like I'm Debbie Downer. So really, Spencer is kind of in that perfect position where he knows my aunt and has spent time with her so I can talk to him about it. But he's not so close that he's like a family member and will get upset if I talk to him about my feelings. So, in that regard, it made us very close because I can share with him and he knows, he's very supportive of that. And I would assume that with the next person it would be similar and if it's not, then that's probably not the right person for me.

Kristy, 29

Together, these examples illustrate the significance of partner support in the *BRCA* mutation-positive experience. When given freely and in a way that effectively meets a carriers' needs, partner support can help smooth the process of surgery; when withheld or not given in a way that is experienced as helpful, partner support can become an added stressor during this difficult phase for young mutation carriers.

Unique Ways of Supporting

In addition to providing support during surveillance and through the surgery, recovery, and reconstruction process, partners of young female *BRCA* mutation carriers provide other types of support that is specific to the unique situations surrounding each couple relationship. Several participants discussed the creative ways they had found to make something positive of their situations as carriers, and this often involved sharing their experiences with other (potential) carriers in some way. After the completion of her one-step RRBM, Rylan decided to create a blog to provide other women with information they might find helpful in deciding about this relatively new procedure. The product was

a collective effort between Rylan and her partner, Byron, and allowed Rylan to be generative in a way that felt powerful to her:

We both designed it together, but he wrote all of the code for it and he has been teaching me HTML a little. He was pretty much willing and happy to do it, he was really excited about it, because he loves his job but he also thinks about doing projects on the side, and this became his little project. That definitely brought us closer together because he really has a concern about my blog and wants it to be the way I want it. He wants it to be functional. He tells me he's proud of me, like almost every day he tells me how proud he is of me, and that helps me a lot.

Rylan, 34

Rylan and Byron had worked together to find a creative and educational outlet that would allow them both to utilize a specific expertise, and that also gave them an additional opportunity to share their *BRCA* experience with each other. Similarly, Valerie and her husband, both laboratory scientists, created a way to focus together on *BRCA*-related research, which Valerie has taken on as her primary focus at work:

I've decided to try to get into it. It's my first project as a post-doc, I'm going to start looking at *BRCA1* and *BRCA2*. So I've been bouncing a lot of ideas off of [Jake], and he has more "out there" ideas than I do. I can think of the "A leads to B, which leads to C," but then he comes up with something totally different that I never would have thought of. So we've been talking a lot about *BRCA* lately, in a research context. It's nice to not have to feel like I'm being run by it anymore like I did in the weeks leading up to my surgery. I sort of felt like I was just *BRCA* all over. But we talk about it a lot lately just in terms of research.

Valerie, 28

Finally, partners of some participants were uniquely positioned to provide emotional support when others with whom carriers had close relationships did not do so. MaryAnn noted that the unwillingness of some of her family members to support her *BRCA*-related decisions made her more appreciative of the support she received from her fiancé:

I also think it's important how other family members perceive your results, and I've had some interesting reactions from my sisters. One of them is vehemently against being tested and thought that I was being overly paranoid to go through

with the testing. She was negative when I told her that I got tested, and I felt like she was being kind of judgmental. But I think it does make me rely on my fiancé more just because he is supportive.

MaryAnn, 26

Single Carrier's Support Needs

The previous sections provide many examples of both successful and unsuccessful provision of support by partners across various stages of the mutation-positive experience for young female carriers. However, single participants acknowledged a support deficit resulting from their not being involved in a couple relationship.

Several unpartnered participants discussed characteristics they hoped would be present in a future partner. For Lilly, knowing that she was a *BRCA* mutation carrier made her more aware of how important it was for her future partner to possess compassion, in addition to the other traits she had been looking for all along:

Before, I was looking for one type of guy, and maybe now, it might be the same guy, but there's got to be a strong element of compassion there too. I always went for funny, sarcastic, goofy guys. To some degree I'm like a giant kid, not wanting to grow up and all of that, so I always went for those types of guys. Maybe I still would, the element of humor, but there would have to be some strong sense of a mature aspect of their life.

Lilly, 25

For Nichelle, fearing that others would believe that her abilities were limited was a major *BRCA*-related worry. Accordingly, it was important to her to have a partner who believed that she could have a perfectly normal life despite her mutation:

I think just having a positive response, like, "I'm really sorry to hear that, but you know what? You know ahead of time and you're going to be alright. You have nothing to worry about. Don't stop yourself, don't limit yourself because of this.

Nichelle, 21

Ultimately, for many single participants, discussions about desirable traits in a future partner boiled down to one crucial element: staying power. This is consistent with themes discussed in Chapter 7 about participant's fears that partners might choose to end the relationship because they saw the mutation as too much to handle. Serena stated:

I would want him to just be supportive. You know, try to get involved. So I would hope that if he was interested in something long-term, possibly marriage and kids and stuff that he would really support me in that way. I think that's really all I would ask. I would ask for the support and the love and if something happens, stand by me.

Serena, 30

Chapter Conclusion

In summary, effective partner support seems to be a critical component to successfully navigating the myriad decisions and challenges presented to young female *BRCA* carriers. Participants who did not receive the desired quantity or quality of support from their partners clearly noticed this deficit and were easily able to identify ways in which their partners could have been more supportive, and how that would have altered their experience. Those who did receive sufficient support highlighted the various ways in which that occurred and was helpful, as Marie summarized:

He is supportive because he listens to my different things that I'm thinking about. And he is very logical and planful in his thinking. When I'm freaking out about things, he'll always make me stop and think. He kind of makes it like it's not as bad. I'll make it like it's really bad, but he'll be like, "No, it's not really that bad. Let's think about it logically, and everything's going to be OK." So, he's like my strength, my rock, my normal when I'm freaking out and being crazy.

Marie, 28

Effective support and cooperative decision-making about cancer risk management within couples can be an intense bonding experience, as was the case with many participants:

I think it's definitely brought us closer. I think he and I both think about our health a lot more, and we do that together. And it actually has facilitated us making decisions together a lot more. With this, there comes a lot of decision-

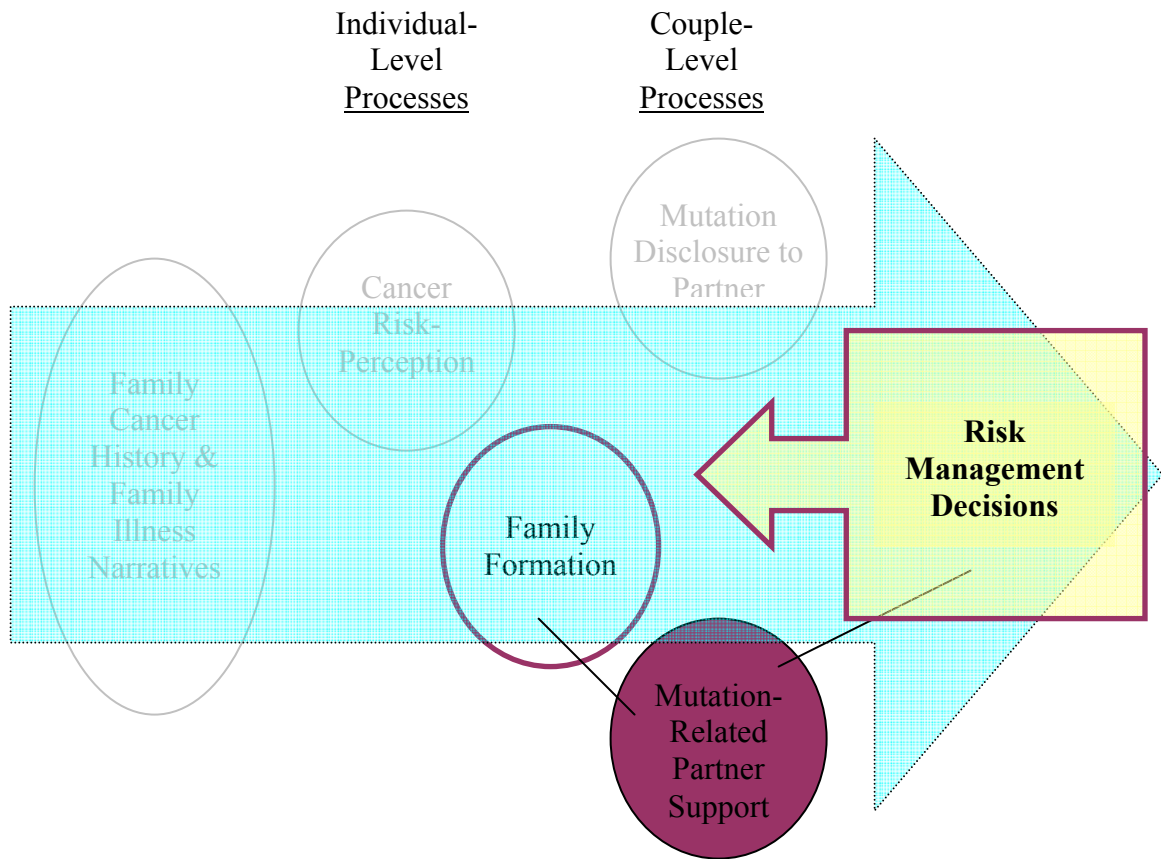
making and he very much wants to be involved in every one, which is great. But it's really helped us as a couple to make those decisions together. I understand more about what he's thinking, what I'm thinking, whether I'm OK with it. Like, do I want to continue screening or do I want to look into surgery? When should we start having children, given time constraints and things like that. So it's helped us in terms of being closer and able to talk about these issues more comfortably. I think in a lot of couples, if you're not presented with serious issues, you never learn how to talk about them. Because we've had these to deal with, we're just comfortable talking. There's really not a whole lot I can't talk about with him. It forced us to talk about family and if we want children, how many do we want? When do we want them? He's definitely made me accountable. Those would be the main benefits I would say.

Marjory, 30

Understanding the importance of effective partner support to partnered carriers raises an important question: how do young female mutation carriers who are *not* in couple relationships identify and receive the needed type and quantity of emotional and instrumental support? This important question should be addressed in future research.

As illustrated in Figure 7, support from relationship partners clearly plays an important critical role in how women make decisions about risk management. The role of partner support in family formation also started to become clear in this chapter, as women discussed how their partners participate in making decisions that would accommodate both family formation and risk-management goals. This will become more evident when discussed in Chapter 9. With regard to risk management, participants highlighted different ways of inviting their partners to be a part of this process. However, a sense that partners (either current or future) should be involved in some way was nearly universal. The lack of a clear role for partners during the period in which mutation carriers rely on surveillance suggests that strategies aimed at helping providers recognize and respond appropriately to spouses' needs can be developed and would be beneficial to these couples.

Figure 7: Role of Mutation-Related Partner Support in Theoretical Model



Concerns about one’s body image and sexuality were common throughout the experience of moving toward, through, and beyond risk-reducing surgery, and this is consistent with previous research on *BRCA* pre-vivors (Matloff, Barnett, & Bober, 2009). Participants’ who reported feeling disconnected from or angry about their breasts may be reflecting the ambiguity that arises just before surgery, which may be part of their method of coping with the impending loss. Awareness of partners’ sense of loss after risk-reducing surgery was common among participants, and anticipating this may certainly be a complicating factor in the process of making a decision about surgery. Couples’ abilities to communicate honestly about their fears and anxieties seem to facilitate

successful coping with women's altered bodies, or altered feelings about their bodies, throughout the *BRCA* experience.

Overall, the data presented in this chapter suggest that partner support is important to young *BRCA* pre-vivors, whether or not it is present. Because couple relationships are such an important facet of this phase of the life course, young adult women are acutely focused on their current or future partners and the role that those individuals will play in their lives. The lack of empirical research about these relationships, and the almost complete absence of partners' voices in the literature, suggests that the medical and psychosocial communities have not effectively investigated or understood how best to serve this population. Physicians, genetic counselors, and medical professionals who are able to involve partners in a way that feels inclusive, welcoming, and helpful to both members of a couple may find new and effective ways to alleviate anxiety and help mutation carriers feel confident about their choices with regard to risk management.

CHAPTER 9: RESULTS—FAMILY FORMATION

“It’s just this weird thing where getting testing and doing more mammograms are recommended all around the same time when people really start thinking about having kids and planning families. ... God forbid ... the last thing I want right now is to have issues with pregnancy. I’ve had enough issues with this kind of stuff.” – Rose

Because breast and ovarian cancer susceptibility threatens the very parts of the female anatomy that are associated with procreation and nursing children, issues related to family formation are intimately tied to the experience of being a young female *BRCA* mutation carrier. These women often worry about how their plans for or ideals about family formation might be impacted by their positive mutation status (Werner-Lin, 2008). Some participants reported having considered family formation even in their decision about whether or not to undergo *BRCA* mutation testing. Nichelle remembered, “I’ve thought a lot about it prior to even test results...even just getting tested, I hope it doesn’t limit me in having children of my own someday.” Others reported being concerned about their ability to have children almost immediately after receiving positive test results:

[When I found out I was positive,] I was overwhelmed, and really anxious...you think you have all these plans for the future and having a family, and doing the things that you want to do, and for the first time I felt like that was threatened. ...I think [learning about my mutation] created a sense of urgency to have a family. We had already been trying to get pregnant, even before I went through the testing, but I think, for the first time my fertility felt threatened. If I’d gotten a cancer diagnosis, if I’d gone in and had a screening and there was something there – I know that there were women who had gotten treatment for breast cancer and that sometimes they’re no longer able to have children. And, as strange as it is, with myself being adopted, and the complexities that I’ve been dealing with, I don’t know that I would want to go through adoption as a way of creating a family. So, that part, I felt, almost like grief in a way. I don’t know, even though it wasn’t like we weren’t able to get pregnant, we were, but I just felt nervous and scared that we may not be able to have a family.

Jane, 30

Jane's emotional experiences while she was attempting to conceive were significantly impacted by her feeling that her mutation could threaten highly-valued ideals about what the experience of forming a family would be like.

Contemplating how family formation and risk management goals will be accomplished is not an independent process, but rather one that carriers complete with their partners. Almost universally, participants described how current or future partners were or would be involved in these issues. For example, Melanie, one of four participants who were pregnant when interviewed, described her husband's view of the extra considerations required in contemplating having a child who might be a mutation carrier:

I feel bad that I have it, but he just shoos that away right away. He kind of doesn't want to hear it. Things like, now we're having a baby, I'll say "We really need to get life insurance, I'm the one that something might happen to," and he's like, "Stop, I don't want to hear that, be quiet." So it's that kind of thing. Just knowing that you can pass it down, and it's me who is passing it down, but again, he just shoos it off. He's not holding it against me, obviously.

Melanie, 31

Melanie was insistent that her husband's attitude was a relief, and that despite having repeatedly checked in with him about whether he felt unfairly burdened, his message of acceptance and love had been consistent.

Participants were concerned about diverse issues related to family formation, including whether or not to have children, the timing of family formation and completing one's childbearing goals relative to desired risk-reducing surgeries, making decisions about family size, and whether their ability to breastfeed their children would be impacted by plans for surgical risk-reduction. Careful analysis of the data culled from the 40 participants in this study suggested that a framework based on control was useful to conceptualize family formation data. Learning about one's positive mutation status was

experienced by many young women as a threat to their closely-held ideals about how and when family formation will occur in the life course. Shifting family formation plans may be an attempt to regain some sense of control. Becoming more deliberate about family formation forced mutation carriers to be less flexible and to make careful choices about prioritizing family formation or risk-management goals. This decision was manifest in three major domains: timing, family size, and breastfeeding.

Effects of Childbearing and Breastfeeding on Risk and Risk Perception

As women navigate the difficult decisions discussed in this chapter, they do so with the knowledge that the choices they make have significant implications for their risk of developing cancer. Although current research indicates that pregnancy and breastfeeding do *not* increase breast cancer risk in mutation carriers¹⁰, several participants cited a worry that this would be true for them and discussed their feelings about their risk given the fact that they were in their active childbearing years. For Leigh, concern about her cancer risk in the period immediately following childbirth came from knowledge she had gained through reading *BRCA*-related websites, and her experience demonstrates how misinformation can lead women to feel very differently about their risk than they might otherwise feel:

The early onset of the ovarian aspects of this were especially troubling¹¹. ... I'd read on the internet of several anecdotal cases of young women with very young children who developed cancer while breastfeeding or shortly after giving birth, so I was very terrified of my immediate risk at that time.

Leigh, 35

¹⁰ Some of this information come from studies indicating that risk of sporadic breast cancer increases during childbearing; however, this is attributed to a promoting effect of the altered hormonal milieu, not a directly carcinogenic or causative effect (*i.e.*, cancers are already present when the patient gets pregnant, and then get unmasked during the pregnancy) (Jernstrom et al., 2004; Poynter et al., 2010).

¹¹ Age-at-onset for ovarian cancer is younger in mutation carriers (especially *BRCA1*) than in sporadic ovarian cancer, but almost never occurs prior to age 30.

Leigh's sense of being at especially high-risk was amplified because the consequences of a cancer diagnosis for her included the possibilities of leaving her children motherless, or having a negative impact on their childhood as a result of her cancer treatment. Beth discussed how her sense of being at increased risk in the months immediately following childbirth influenced the timeline on which she was choosing to have her RRBM:

Knowing that I want to have surgery, I told him that I need to have surgery before my son is a year old because my sister got diagnosed when her daughter was 11 months old, so I had that ticking time bomb kind of thought...

Beth, 30

For Beth, this sense of increased risk came from her first-hand experience with her sister, not something abstract that she had read. Beth's concern has some basis in fact, since she and her sister carry the exact same mutation, and members of a family are more likely to develop cancer at similar ages. In addition to these general concerns, some participants discussed specific issues related to childbearing and risk that made them feel especially vulnerable.

Infertility-related challenges. Potential cancer susceptibility related to the use of infertility drugs/treatments is an important consideration for a minority of women who carry *BRCA* mutations. Among the participants in this study, only one had had first-hand experience with fertility treatments. Libby was 32 and married when interviewed. Prior to focusing on family formation, Libby had been aware that a *BRCA* mutation was present in her family, but was planning to wait until after she had children to get tested. She and her husband spent several months attempting to conceive a baby without success, and ultimately decided to consult a fertility specialist. Upon learning that a *BRCA* mutation was present in her family, he strongly recommended that she complete her testing sooner because of his concern that using fertility drugs might be dangerous if she were positive.

Upon learning her status, Libby chose to postpone family formation in order to complete RRBM so that she could use fertility drugs without worrying that doing so would increase her breast cancer risk. Libby recalled how she felt about her cancer risk immediately after completing genetic testing:

The more you read, the more freaked out you start to get. I had taken a round of hormones to do an artificial insemination treatment. I just did it because I really wanted to get pregnant. And I was taking a really low dose of hormones, because the doctor knew I had the gene. After that, I felt very vulnerable. I was like, “oh my God,” because it didn’t work. I might have just put myself in a really bad situation. I felt really vulnerable also with the ovarian cancer. Because with the fertility stuff, they track your ovaries so much. I’m constantly getting these ultrasounds of my ovaries and I just felt really vulnerable in that sense.

Libby’s sense of vulnerability derived from her beliefs about how hormonal exposure might increase cancer risk¹², and her ovarian imaging studies, which kept the issue of cancer at the forefront of her awareness. She explained how her fertility treatment had changed after learning that she was *BRCA* positive, and how her use of assisted reproductive technologies influenced her decision-making about RRBM:

I’m still having infertility issues, and they don’t really know what to do with me because I have the mutation. They were like, “We want to give you hormones, but we don’t because we’re afraid. We don’t want you to get breast cancer, or ovarian cancer.” They don’t want to jack me up with hormones and then leave me alone for nine months. They were afraid of a microscopic cancer brewing or something, and then giving me hormones and it just accelerating the whole time I’m pregnant and having a tumor by the time I give birth. They were afraid of that. So, I went to a couple of different doctors and I said, “I’m definitely going to have a mastectomy. So, when should I get it? What would you do? Would you do it after you have kids so that you and breastfeed? Or should I do it now?” And at first, I didn’t even think to do it now because I still know I’m going to have kids. I want to breastfeed and, to me, it seemed like it could wait. I’m only 32. My grandma didn’t get it until she was 38. So, that gives me five years. In my mind, it was going to be a risk in my late 30s. So, what happened was, the doctors all said,

¹² The single existing study that addresses this question concluded that there is no increased cancer risk associated with use of fertility treatments for *BRCA* mutation carriers (Kotsopoulos et al., 2008). This belief (among carriers and providers) seems to come from reasoning by analogy – *i.e.*, estrogen increases the risk of breast cancer (a dramatic oversimplification) so any hormonal elevation must be bad – in the absence of data.

“Look, if you want to be safe, get the mastectomy done now. You won’t be able to breastfeed, but at least you won’t get breast cancer.” And to me, that was like, “Breastfeeding? Alright. I’ve never done it before. I don’t even know the difference. I just don’t want to get cancer.”

Based on her doctor’s well-intentioned but nonetheless arbitrary recommendations, and to mitigate her risk prior to undergoing intense fertility treatments, Libby decided that RRBM prior to fertility-assisted pregnancy was the best route for her. At the time of her interview, she was quickly approaching her RRBM surgical date:

Now, I’ve stopped all treatment. They recommended IVF because they didn’t want to do little doses of hormones over a long period of time. They just kind of wanted to move to the big guns, and do a lot of hormones over a short period of time because your chances of getting pregnant are higher. So, that’s when I thought, let me just get this surgery over with. Then in six months, I’ll start doing IVF treatment. So we put the baby plan on pause to take care of the surgery.

Libby, 32

Although the experience of one individual can certainly not be generalized to all *BRCA*-positive women who encounter fertility issues, Libby’s story does illustrate how fertility difficulties can markedly complicate progression through the young adulthood phase of the life course, as women work to create their families.

Surveillance during pregnancy and breastfeeding. Participants relying on surveillance to manage their risk during their childbearing years frequently reported being very worried about how they would continue effective surveillance while pregnant or breastfeeding. This issue is further complicated by preliminary indications that breastfeeding may be protective for *BRCA1* carriers who breastfeed for a cumulative total of one year; other studies have shown no relationship and there is no association for *BRCA2* carriers (Eitan, Michaelson-Cohen, Levavi, & Beller, 2009). In making her decision about when to try to become pregnant, Melanie, 31, reported that she was concerned with the timing of her pregnancy and ability to continue screening during the

months that she was breastfeeding. This concern was echoed by many other participants. Jane recalled her sense of relief when, upon learning that screening and pregnancy were not compatible, she realized that she had gotten both breast and ovarian screening completed just prior to becoming pregnant:

I ended up incredibly lucky that I had had a mammogram and an MRI and a transvaginal [ultrasound], all prior to getting pregnant, and then we were able to get pregnant. I was really glad because once I was pregnant then it dawned on me that I can't get any screening done during that time.

Jane, 30

Jane's realization of this *after* becoming pregnant suggests that some women may not be receiving sufficient information from their physicians about what screening should be completed before attempting to conceive; if this happens, women may find themselves pregnant after a long gap since the last screening visit, and this may contribute to increased anxiety for the duration of the pregnancy and breastfeeding months.

For some carriers, awareness of the inability to undergo cancer screening during gestation/breastfeeding contributed to childbearing decision-making. Elaine indicated that this led her to have no additional children:

I don't want to take the risk. I hate the idea of being pregnant and not being able to have screening, and then having to choose maybe not to breastfeed because of wanting to have my breast MRI and stuff. I know that I don't want to put myself and my husband through that, so this has kind of made our decision not to have any more children. Why not get [the surgery] now? I know there are risks of osteoporosis and heart disease and stuff like that, but I figure it's less of a threat than the ovarian cancer.

Elaine, 34

Similarly, Leigh stated:

Certainly, since I already had a very nice family, finding out that I had a mutation [was] a factor [in deciding] to not have more, because I have read of all those issues where breast cancer may sometimes be triggered by pregnancy, and you can't screen well during breast feeding, which was always very important to me.

So I think it would have been a lot more stressful to have another child knowing I was a mutation carrier.

Leigh, 35

Jane recalled her frustration at not being able to continue screening during her pregnancy, and the resulting anxiety:

I breast fed my daughter for the first six months, and I think that's something that was very, very, challenging to get doctors [who] are willing to do screenings while you're still lactating. I was able to do a mammogram, but when I met with the radiologist, it was the most unprofessional experience I've ever had. We were arguing with each other, I was in my robe ready to have a mammogram done and she said I couldn't have a mammogram because I was breast feeding and it wasn't going to be a reliable reading because a lot of the breast tissue obviously is so different when you are breast feeding that it could mask a cancer that you wouldn't be able to see on a mammogram. So, I think in the professional community, there seems to be a lot of discrepancy and uncertainty about what to do with lactating women who are high risk for breast cancer.

Jane, 30

Jane's wish for uniformity among medical professionals regarding breast screening for lactating *BRCA*-positive women is a valid request; most doctors will not, in fact, perform regular breast imaging during lactation because images produced are unclear and not very useful; rather, they rely on self and clinical breast exams, and may recommend that women take this extended period of problematic imaging into consideration when deciding how long to breastfeed (Eitan et al., 2009).

Beth recalled how her physician's policy regarding lactation and screening interfered with her plans to undergo RRBM. She stopped breastfeeding earlier than planned so that she could proceed with RRBM, primarily because her older sister had recently been diagnosed with breast cancer and Beth's perceived risk was quite high. She recalled:

I finally stopped it and I called my doctor and I said "OK, I stopped breast feeding today, I want to make an appointment for imaging." And at first, she said, "We want to wait until you have your period, until you've been done breast feeding for

two months before you have imaging.” And I was so mad because I knew I would have to wait about a month between stopping breast feeding and having imaging done, but I was so mad that she wanted me to have two periods because I hadn’t had any periods at all and I didn’t know when I was going to get it, and I didn’t want that to push back the surgery. So I was mad. I called her back after a couple weeks, I’m like, “This is ridiculous, my sister has an aggressive cancer, I don’t care if the imaging isn’t going to be perfect, I’m going to have this surgery no matter what. So, to me it doesn’t matter if the imaging is crystal clear or not ... It’s a screening.” So they finally said OK.

Beth, 30

The experiences of each of these women illustrate how ambiguous expectations about cancer screening during breastfeeding, or a mismatch between a patient’s and doctor’s ideas about how to manage these competing demands, can create anxiety, frustration and anger. If Jane and Beth experienced a deficiency of clear information on this subject from their physicians, it is likely that other women have experienced this as well.

Choices and Life Trajectories

Among young women in the general population, contemplating the timing of major life transitions, including family formation, is a common preoccupation, as they navigate the period during which many components of adulthood (*e.g.*, partner, career, family) are becoming solidified and synthesized (Arnett, 2004). Women consider these decisions both independently and in the context of their ongoing relationships. *BRCA* mutation-positive women in their twenties and thirties are no exception; however, their decisions and the factors they must weigh in making them are complicated because of limits imposed by their mutations. Women who are not *BRCA*-positive face the likely end of fertility potential at the point of natural menopause¹³, which might allow 20-30 years during which biological childbearing can occur. This relatively nebulous “window of

¹³ The current average age at menopause in the US is 51.3 years, with peri-menopause beginning on average at age 47.5 (McKinlay, Brambilla, & Posner, 2008).

opportunity” for having children affords the luxury of casual decision-making, or even choosing *not* to make decisions, *i.e.*, allowing conception and childbearing to happen naturally or without a great deal of planning. In contrast, *BRCA* mutation carriers navigate young adulthood aware that RRSO or ovarian cancer may end their fertility at age 35 or 40. This results in as few as 5-20 years of fertility potential, once they decide to have children.

Werner-Lin (2008) referred to this phenomenon as the “compressed family life cycle,” contending that “salient issues associated with carrying a *BRCA* gene alteration shift at each stage of the life cycle. ... [and] that developmental concerns play a significant role throughout the lifespan when considering genetic risk,” (p. 432). Data from the current study suggest that a more focused way to conceptualize this phenomenon might be to consider how awareness of positive mutation status brings about changes to life *trajectories* (Elzinga & Liefbroer, 2007). In recent years, a focus on life course trajectories rather than the individual or family life cycle has arisen from a recognition that societal changes such as cohabitation, delayed age at first marriage, and alternative family forms have “led to an increased de-standardization and increased complexity of the transition to adulthood (Elzinga & Liefbroer, 2007, p. 226). This part of the life course is no longer best understood as an element of a cycle that is repeated over generations; rather, it is a highly individual set of choices made in the context of the unique set of circumstances in which a given person is situated. Each person, then, must select his or her own paths for career, relationships, and family as they move through the lifecourse, and these paths may diverge, overlap, or change course at various points and for a wide variety of reasons. Knowledge of oneself as *BRCA* mutation-positive would

certainly qualify as a piece of this tapestry, and it is inarguable that possessing this knowledge necessitates decision-making about paths taken in young adulthood. Women who know themselves to be *BRCA*-positive must take this information into consideration as they select, create, and modify their life trajectories.

Existing data suggest that *BRCA*-positive women often consider how the need for and timing of RRSO and/or RRBM will affect their family formation plans (Hoskins et al., 2008; Werner-Lin, 2008). RRSO definitively ends the ability to bear children naturally¹⁴, and RRBM eliminates breastfeeding (the importance of which varied among participants). In addition, several participants were aware that they might die prematurely from *BRCA*-related cancer, as had happened to family members. These women were aware that delaying childbirth might give them less time with those children.

Several participants recalled transitioning from not relating their positive mutation status to concrete ideas about family formation, to a time when these two issues became inextricably linked. This transition often resulted from assembling information about the implications of their mutation from multiple sources, gradually achieving more complete understanding of the myriad ways in which their mutation might alter their life plans. This was described as a lengthy process during which realizations about needing to accommodate life plans to their mutation occurred at various developmental points throughout young adulthood. Alternatively, some participants made one seismic shift from their pre-*BRCA* to their post-*BRCA* lives, reorganizing all of their plans in one fell swoop. In either case, this shift represents a point at which mutation-positive women must contemplate their life trajectories, and those paths might have to be different than

¹⁴ Theoretically, as long as the uterus is intact, it might be possible to use assisted reproductive technologies to implant an embryo; however, use of this technology is not yet widely available or known to *BRCA* mutation carriers.

what was previously planned. For example, a woman who planned to pursue several years of post-graduate education and focus on career development through her twenties and into her thirties might have envisioned herself having her first baby at age 35; when knowledge of her *BRCA* mutation elicits a desire to have RRSO by age 38, her plan may no longer seem workable since it would only give her three years in which to commence and complete childbearing. Therefore, she may have to make changes and/or sacrifices to her educational and career goals in order to accommodate her family formation and risk-management priorities, or alter her family formation goals if she is not willing to change her educational and career plans. Ultimately, these changes create an altered path, or trajectory, through young adulthood.

Messages regarding family formation urgency reached *BRCA*-positive women from multiple sources, creating a complicated backdrop against which decisions were made. For some women, receiving and assimilating these messages conveyed a loss of control or decreased agency with regard to family formation decision-making. For most participants, it was stressful to receive consistent directive messages from medical professionals regarding how they “should” address their mutation-related fertility concerns, but this information did influence their decisions related to timing of pregnancies. Annie learned of her *BRCA* mutation at age 31, while childless and in a casual relationship. Certain that she wanted to have children someday, she felt pressured when the connection between being a *BRCA* mutation carrier and having children finally resonated:

[The genetic counselor] wanted me to meet with an oncologist to talk about preventative chemotherapy measures that were available to me. And if I decided to go that route, she wanted to link me up with some fertility specialists, because of the risks there. That’s when alarm bells really started to go off. I had never

really put together A to B, B to C, to start thinking about it in those terms. You know, at that point, I was 31, single and I'm like, "Oh, Holy cow. Here we go." This is not really the way you picture things working. It was my own sensitivity, being in my '30s and not being married with kids. There's always a little bit of, "Okay. I don't need somebody else telling me to get married and have kids." I want to do it, don't get me wrong. It was kind of that stereotypical, "Oh, my God. I'm turning into that, I'm becoming very sensitive. Is there a huge clock over my head that's ticking even louder now?"

Annie, 32

For Melanie, messages about the urgency of completing her family so that she could proceed with RRSO came from the researchers with whom she interacted in the BI study:

After we were married, we knew that prophylactic surgery, both the ovaries and the breasts are definitely an option for me. And so having kids is obviously affected by that, so we knew that we needed to get pregnant. And people at NIH said, "What are you waiting for, hurry up and start having kids." So we knew that sooner was better than later. I've kind of decided I want to be done by 35. Ovaries'll come out, and then we'll decide about the mastectomy.

Melanie, 30

Melanie's experience also opens the issue of how best to involve partners in the restructuring of family formation plans. This important dynamic was highlighted by numerous other participants, several of whom reported that being mutation positive put "a little more pressure to get your family started. And, indirectly, that puts pressure on the other person, too," (Marjory, 30). Rose had just completed her RRBM when interviewed; at age 30, she and her husband had decided to proceed with this surgery before getting pregnant. She described the impact her mutation status had on her family formation:

It's been hard for my husband and I. This affects things you do in your life, and timing, because these tests are done when you're making all these big decisions about reproduction. I mean that in a very medical kind of way. Because now that I've had the mastectomy I won't breast feed. And, while I'm going through the surgery, we certainly won't be trying to get pregnant ... because I don't want to be put under or taking pain medications while pregnant. I think the timing thing does play a factor, because if it's hard to conceive then we're kind of running out of years. I'll be thirty-one later this year, and I'm thinking of wanting to do the oophorectomy at thirty-five, so it's kind of a compressed window of time. So, timing becomes very urgent in a way, especially if we have any type of problems.

Suddenly everything becomes very compressed. It's just this weird thing where getting testing and doing more mammograms are recommended all around the same time when people really start thinking about having kids and planning families. Maybe if I had made the decision when I was younger... I could have done the surgery five years ago, six years ago, and then it wouldn't have played a role right now. Because, God forbid, it's the last thing I want right now is to have issues with pregnancy. I've had enough issues with this kind of stuff.

Rose clearly felt enormous pressure around having a family. She was fearful that if she did not become pregnant quickly, her available timeline would feel stifling, and she would likely experience a strong sense of failure and/or injustice related to struggling with another complicated reproductive health issue:

People say there's never a good time to have kids, so you have this other added variable. Well, I'd better have kids now, hurry, hurry, quick get it done. And, of course that kind of pressure can also be bad for trying to conceive. That's kind of like the next stage I'll be going through and I really don't know if I can be calm about the whole thing. That I won't feel so pressured.

Rose, 30

Participants reported worrying about their ability to raise children to adulthood before they are affected by a future cancer diagnosis. Serena, age 30, noted the importance of a trend in her family in which most women were diagnosed at a specific age, and how that "deadline" in her head translated to her concerns about family formation:

I think especially now that I have hit thirty, I would like to have them sooner than later. ... I want have kids while I still have the energy to have them. And... if the magic number in our family is age forty-six, I have sixteen years which means I could potentially see a good portion of my son's or daughter's life...if and when I get diagnosed, the hope is that I would be able to have as much time with them as possible, I guess.

Serena, 30

In the aggregate, the experiences described by these women illustrate the phenomenally complicated decision-making landscape surrounding family formation as a mutation carrier. To manage these conflicting demands, participants reported having considered

altering or having actually altered family formation timing, the number of children planned, or the priority they placed on breastfeeding. The remainder of this chapter will be devoted to describing how these decisions were made and, often, negotiated between partners.

Controlling the Timing of Family Formation

Many participants had general ideas about how the timing of pregnancy would be shaped by ideas or recommendations regarding the optimal age for risk-reducing surgery. Even those who were several years away from first pregnancy were aware of the limited time available to have children:

My doctor did say that by my late 30's they probably would want my ovaries to be removed as a precaution with the ovarian cancer, so that's just something to think about as far as the timing. Not that I need to have children now, but just keep that in mind if that's something that I do decide, that there is somewhat of a limited window...essentially a ten year window of opportunity from now.

Julia saw this “window of opportunity” as workable at the time of her interview, since she was in a potentially long-term relationship. However, if this relationship ended, leaving her single in her later twenties, she might be less optimistic. In fact, Julia and her boyfriend had been unable to agree about whether they would have children:

He does not want children and I guess I am still undecided. I'm not opposed to them but I'm not quite sure if that's something that I want in my future. For him, if it happens it happens, but he's not somebody that actively wants to plan or pursue having children. And he was very up front, he said “I [really] don't want kids ... and I don't think that's something that will change.”... I'm still undecided and not quite sure where I fall with that decision. It's hard for me to say...“Kids weren't meant to be.” So we haven't really gone anywhere with it.

Julia, 24

Considering Julia's position as a young, unmarried female mutation carrier further illuminated a couples dynamic discussed in Chapter 7: for those whose childbearing potential is definitively limited by a surgical risk-reduction timeline, establishing and

maintaining a relationship may be dramatically impacted. Having to tell a dating partner that one needs to accelerate relationship and family progression to complete family formation goals by a certain time may detract from young couples' ability to experience natural relationship development, as described by Kristy (see Chapter 7).

Rylan provided an example of how some carriers carefully consider and construct the most workable timing of family formation within their couple relationships. Engaged at the time of her interview, she reported:

We've already had lots of conversations about it. And I know he wants to have kids, for sure, and I've never been as gung-ho about kids as he's been. It's going to make us decide when to have kids, when to stop having kids, when I can have my ovaries removed, which is recommended by age 40, and I'm 34, so it kind of leaves me with literally a biological clock that I have to keep up with. How long are we going to wait to have kids, and how many are we going to have and how quickly will we have them, and when am I going to have my ovaries taken out, if at all, which I really want to do as soon as I can, but we also want kids. And I'm also more open to adopting, and he really isn't as open to that, I think his background and culture and family have a lot to do with that.

Rylan, 34

Because Rylan had already completed her RRBM, her primary risk-management focus was the RRSO she planned for age 40. Her description of her literal biological clock was another powerful illustration of the intensity of this issue, and also illustrated the manner in which some women pulled their partners in close around *BRCA*-related concerns, making decisions at least in part based on what their partners thought or wanted.

Once they have achieved a personal understanding of how their mutation status will shape their family formation decisions, participants began to make specific plans regarding the timing of both childbearing and risk-management interventions. For many, reaching these conclusions was described as the outcome of a process in which they absorbed as much information as they could find (or tolerate) from various sources, and

then identified *the* critical or pivotal piece of information. This was characterized as a gut-level sense of what “felt right” to each individual, *i.e.*, the result of understanding how one feels, rather than what one rationally thinks or what the objective facts or data suggest is the optimal choice.

Reorganizing one’s assumptions and plans about when biological childbearing will occur is a complicated and challenging task. However, doing so effectively is one primary way that women attain a sense of agency regarding the threat of cancer; in the context of a couple relationship, this may also provide an opportunity for building closeness between partners. In choosing which path to take, some participants allowed their plans for risk-management to shape their decision-making about family formation, while others looked to their family formation preferences to guide decision-making about risk-management.

Prioritizing timing of childbearing over risk management. Represented by a notably small group of participants, some women concluded that the need to undergo risk-reducing oophorectomy would not alter their family formation plans. Noelle reported having always wanted to have children at a relatively young age:

I’d love to have like two or four kids, and have them while I’m relatively young. I’m 26 right now and I’d like to have my first child by the time I’m 28. And then as soon as I could physically handle it, I’d start having the surgeries. I’m planning to do both surgeries, and wait until after I’m done with all of my childbearing. [That plan is] not any different than what I wanted before I knew about the mutation. The mutation just makes it more important.

Noelle understood the need to implement a cancer risk management plan and how it would overlap with her family formation desires, but denied any intention of changing her pre-*BRCA* plans. However, her increased awareness about the urgency of completing

her family formation goals had created a difficult dynamic in her relationship with her boyfriend:

I think this whole thing scared him, especially when I know I want to have children by the time I'm 30, or that I want to be pregnant for the first time by the time I'm 30. And I want to start having surgery when I'm 35. Those things scare him to think that there's a deadline. And instead of approaching it with, "OK, well if there's a deadline, let's map it out," he's thinking, "Oh, my God, back off." He doesn't want to even think about it. Where to me it's life and death, sadly to him, it's something that is so upsetting he can't even think about it.

Noelle, 26

Given Noelle's relatively young age, her partner may be able to work through his difficulties with her compressed family formation timeline, permitting them to reach an agreement about how to proceed. However, if this does not occur, Noelle could face the difficult position of looking for a new partner when she is older, and with fewer years remaining prior to undergoing RRSO (planned: age 35).

Like Noelle, Rylan felt that family formation timing decisions should take into account, but not be primarily driven by, RRSO timing. She described other priorities that she and will consider in deciding when to have children and when to undergo RRSO:

I think both of us want to have some time with each other after marriage before we have children. ... we haven't made any decisions now, and I think [that]... would be too premature, anyway. I don't plan that far in advance, really, in any area of my life, and this isn't any different. I think we haven't any idea of what things will be like a year or two or three from now, like financially, whether we'll be ready for kids or not, and that's very important to me. I want us to have a really stable environment for them, and we can't know that right now... I think we'll just have to cross that bridge when we get there, leave things a little up in the air and sort of open-ended as far as my ovarian surgery, but I think that's just the way things just have to be.

Rylan, 34

Having already completed RRBM, Rylan's relatively relaxed attitude about her family formation timeline may be related to her significantly decreased perceived risk of breast cancer; and the absence of ovarian cancer in her family; thus, her overall perceived

cancer risk was likely lower than for participants who felt high ongoing vulnerability. Her experience illustrates a balance of agency (ensuring reduction of personal cancer risk by controlling what happens to her body and when) and communion (making these decisions in the context of a loving and supportive relationship, with thoughtful consideration of her partner's perspective).

Prioritizing risk-management over timing of childbearing. More commonly, participants actively made changes to their family formation timing to accommodate their desire to undergo RRSO. Strategies used included having children earlier than previously planned, and having children in quicker succession (minimizing time between pregnancies). For some, this latter decision was made at the outset of family formation, recognizing the relatively short available time to have multiple children given perceived RRSO deadlines. Several participants who foresaw RRSO in their future felt a sense of urgency to commence or complete family formation, and pinpointed specific deadlines by which childbearing would be complete so they could proceed with risk-reducing surgery:

I'm...concerned about when we're going to have kids...we've already talked about having two kids, and ideally I would like to be done by 35. At this point it's probably going to be [more] like 36 because I really want to have an oophorectomy as soon as possible.

Sophie, 31

For many participants, this sense of urgency was driven by their understanding that ovarian cancer screening techniques are very ineffective, making RRSO the best defense against cancer at that site, as discussed in Chapter 6. Reina, who had not children when interviewed, illustrated this:

[Paul] and I have both had the conversation that our family will be complete before I'm 40 because I do have the desire to have my ovaries removed when I'm 40. Maybe there will be a lot more changes over the next ten years, which I would

love to see, but at this point the screening techniques for ovarian cancer are not as good as for breast cancer.

Reina, 29

Recognizing that decision-making about family formation would be an ongoing process, several participants noted that conversations and negotiations around this topic would likely be a feature in their couple relationships for the foreseeable future:

...Unfortunately, *BRCA* is the gift that keeps on giving and we're going to have to stay on top of a number of things, most specifically my ovaries. And that's going to be a decision that affects him very directly because of the involved choices about reproduction. So I think a lot of the big choices in our lives that we thought would happen either by mistake or with a little bit of forethought, now I'm going to be considering in a totally different light just given my deadline of having my ovaries removed by a certain age.

Pauline, 30

Pauline rightly expects that she and her partner will have to make careful, thoughtful decisions about how and when to achieve their desired family size, and that this will require them to continue to discuss these issues actively rather than relying on chance or more spontaneous choices to determine what they do.

The decision to alter one's family formation plans may require some juggling and even some tough decisions with respect to the other goals that young women wish to accomplish during young adulthood. This nexus is particularly evident in 21st century America, with women increasingly choosing to pursue higher education, be independent longer, and/or marry and have children later (Arnett, 2004). For MaryAnn, a conflict developed surrounding her educational and career goals:

When I was younger I had always assumed that I would settle down in my late 20's, but now that I'm back in school that's kind of been pushed further into the future. We were always waiting for the right time ... so I was thinking early to mid-30's would be good, I didn't have any feeling of rush. Now, knowing that I have a *BRCA* mutation, I've tried imagining the future and the timing of things. They recommend the [RRBM] by 35, and they recommend getting your ovaries out by 40 if not sooner. So then you have to do the math in your head. Let's say I

finish my PhD at 30 or 31, plus a really busy post-doc for two years. So that puts me at 32 to 33 with surgeries looming really shortly thereafter. And it's just not a big window ... I had to start thinking about it more. Maybe I'm going to have to be more flexible; maybe I shouldn't wait. So now being in school and having a family at the same time, I would be open to it. In grad school, no time is an awesome time, but ... maybe writing would be easier because I could be home when I do it. I wouldn't be opposed to having kids in grad school, but I would still try to wait for a better time.

MaryAnn, 26

Changing their plans regarding when to start a family is a complex task for MaryAnn and her fiancé; it requires not only a shift in what MaryAnn had planned for herself professionally, but significant shifts for her fiancé as well. Like MaryAnn, Lynn also noted that the time constraints imposed by her *BRCA* mutation intersected with her educational and career goals:

You get told these statistics, and time tables of suggestions of when you should have your kids by. It was a little bit stressful because it definitely put more of a time restriction on my life. I even joked with my mom, 'I want to go to med school, I won't be [done] until I'm 32. That's going to give me like a three year window to find a husband and to have my babies.'

Lynn, 24

The challenge facing Lynn may be more formidable than MaryAnn's: although younger, Lynn was not in a serious relationship and therefore saw less flexibility in her options for accomplishing her family formation goals. Sadie also remembered that her educational and career goals had been in conflict with family formation plans:

When we first got married, I had gone back to school. Our plan was ... [to] be married for a couple years, I would get out of school, and then we would start having kids. And that changed overnight. Immediately, I started thinking, "Why are we waiting to have kids?" It was always financial ... I would be out of school, we would have two incomes, we'd have more money, we'd be able to provide better. And then I started thinking, "Well, a kid doesn't care how much money you have. They just care that they have parents that love them and all that stuff. And it's never going to be the best time to have a kid. There's always going to be something that you're going to have to change in your life in order to make room for kids. Why don't we just start making changes now?" ... So I approached my

husband and said, “I don’t think we should wait. I think we should just start trying now and see what happens.”

Sadie, 33

These three women demonstrate that some young female *BRCA* mutation carriers choose to pursue education, career and family formation goals simultaneously, a considerable shift from historical notions regarding when and how these major life events would occur. Making this shift will likely have significant positive and negative implications, and create a need for ongoing navigation and balancing of priorities.

Telling one’s partner of the desire to alter the timing of family formation can be a formidable task for young *BRCA* mutation carriers, especially in the context of a new or developing relationship. This is nicely illustrated by Marjory, who described the great care she took in conveying her ideas to her committed partner about when to have children:

I haven’t talked to him about the oophorectomy. He knows we have a limited time to have children, but I don’t think he realizes that I’ll have to have the surgery sooner than later. And so, that is something that will come up. But I don’t want to put too much on him at once. I’m kind of careful when we talk about this, and how much we talk about. I don’t want to freak him out. And so, that particular discussion has not happened. In general, he knows that I’ll get rid of my ovaries at some point, but I don’t think he realizes it could be in five or ten years. So that’s something in the future we’ll probably talk about.

Marjory, 30

Marjory’s experience conveys the perceived high stakes related to negotiating family formation timeline changes. Despite hers being a very solid, happy relationship, she still felt compelled to carefully regulate disclosing the details regarding what her risk-management plans would mean for her fertility potential. This contrasts significantly with several other participants, who invited their partners to be more intimately involved in such issues.

Women who were single when interviewed provided a unique perspective regarding the role they envisioned for future partners relative to the timing of family formation decisions. Lynn, in a casual relationship when interviewed, had given extensive thought to how she might broach this topic:

I will let [my partner] know that [having] this mutation suggests that I have my kids by the time I'm 35, maybe I can stretch it out a couple extra years. I would just tell him, that's the situation. I do want to have kids, and hopefully that person will understand. But I won't know until I get there, and the best thing I can do is just be honest and up front about all that stuff. I'm only thinking I want two, maybe three, so there might be several years where I'm making baby after baby.

Lynn, 24

Lynn's perspective at this point in her personal and relationship development was that the decision-making was her responsibility, while her partner should simply accept the decisions she has made. It is interesting to speculate regarding how Lynn's preconceived notions will actually align with the preferences of the individual with whom she ultimately partners. Her definitive ideas about his role, if unchanged, could create a problematic couple dynamic, or might limit the pool of eligible partners.

Unique circumstances in some participants' lives and relationships have significant bearing on when they will begin or complete their childbearing and undertake RRSO or other fertility-abrogating risk-management options. Kate had two young children and was getting divorced when she learned that she carried a *BRCA* mutation. Although she did not expect at that time to remarry or have additional children, she met a man with whom these options seemed appealing and was planning a life with him. She spoke about how her risk-management timeline added some unique points to navigate as they made decisions about how to move forward together:

Because I am planning on having my ovaries removed when I am 40, our timeline is to have [a baby] sometime in the next three years, because I want to be past

infant stage if there are any effects of menopause that are challenging. Our preference is to be married first, but if it were to happen that a baby came along first, then that would be fine as well. Mostly because of my divorce and him having waited so long to get married in the first place, we're not in a big rush to get married. But at the same time, being that we're both a little older, we don't want a super-prolonged dating relationship before we get married. A year or two and then get married, and what we have planned is that right after we get married, we'll start trying to get pregnant.

Kate, 35

Not surprisingly, choosing to have children sooner than one might have otherwise done leaves some women feeling that they and their partners are taking this significant step forward prematurely. For example, Pauline had

always allowed myself to imagine having children much, much later ... I honestly did not think that we would even have the discussion about having children very seriously before our fifth wedding anniversary. ... And so, that would have put me right at thirty-five. ... But unfortunately, *BRCA* changes a lot of things having to do with our family planning picture. It hasn't necessarily accelerated the desire for us to start a family, which is difficult because you don't want to start before you're ready. But it does put a deadline on our family planning in a way that we didn't really have before. I would want to become a mother for the first time at forty-four ... that sounds like when I'll be ready. But a bummer of *BRCA* is that that's no longer a possibility. So, we just started talking about it, we're still not ready but instead of the five-year window ... we're thinking more like three years before we start ... planning and trying.

Pauline, 30

While this might seem like a reasonable strategy for reducing the perceived time pressure to accomplish family formation goals, it could present other problems if the feeling of being "not ready" persists after children arrive. In addition, some couples may not agree about speeding up their timeline and having children earlier, and there is definite potential for this to cause relationship problems. Though she did not describe this as a serious problem, MaryAnn alluded to some incongruence of opinion between her fiancé and herself:

My fiancé is a little bit more reluctant because he's not ready whatsoever. I don't even think I'm ready, but I mean, who is ever ready to have kids? I don't know. I

still think that he feels that early 30's would be more appropriate. So I feel more pressure than he does right now.

MaryAnn, 26

MaryAnn is certainly not alone in reporting disagreement between partners related to family formation planning. Marie and her husband had planned to wait until they were both finished with graduate school to have a second baby, but, after learning about her mutation, she felt strongly that she wanted to do so immediately. Asked how her husband felt, she stated, "I wouldn't say he's terribly excited. I think he knows what has to be done because of necessity, but it's just so different than what we were originally going to do, so it's scary. It's all just crazy" (Marie, 28, married). Maelie also remembered having had some disagreement with her partner on this issue. She felt strongly that she wanted to have both risk-reducing surgeries as soon as possible, largely because her older sister had died at age 32 after two breast cancers, but still wanted a relatively large family:

We definitely did have kids sooner. We had our master plan of waiting three to five years before we started having children, and we're just coming up on our fourth wedding anniversary and we have three kids already. So it definitely sped that up, and we knew we wanted them, so let's take care of it so we don't have to go through the same fate that my sister and her family did. I had to do a little bit of pushing. My husband would have been fine to wait a couple of years, but I didn't feel comfortable with that.

Maelie, 33

Maelie changed her family formation timeline by having children earlier than planned, and having them close together. Her two older children were born only 11 months apart and in the same calendar year. Having "pushed" her husband to begin childbearing sooner, she reported that they were both thrilled with the family they had created and happy to have been able to put both risk-reducing surgeries behind them while Maelie was still relatively young.

This complex and multifaceted decision is not an easy one to negotiate, and there is no one solution for all couples. Some of these difficulties are due to suboptimal skills in communicating about and processing the demands related to this issue. For some, effective psychotherapeutic care could be helpful in overcoming these challenges. Providing mutation carriers and their partners with access to therapists specifically trained to deal with these issues could pave the way for healthy and adaptive coping, and provide potentially critical support during a stressful decision-making process.

Spacing between children. As previously stated, reducing the time between sequential pregnancies is can be a useful strategy for completing childbearing goals prior to undergoing RRSO. Libby had temporarily put her family formation plans on hold while she completed RRBM, and was anticipating using fertility treatments to successfully conceive. She shared her thoughts about how she and her husband would go about timing multiple pregnancies over the next several years:

[After the first baby,] I would start [fertility treatments] again as soon as possible. I kind of want to get it done as quickly as possible, and then get my ovaries out. Just be free to be done with my ovaries, and then maybe get them out around 38 or something. Some doctors have told me 40, but I don't know. Something around there. Once I get started having children, I want to do it more quickly, but having the mutation didn't push us to start having children sooner.

Libby, 32

Like Libby, Acacia reported having decided early on to have two children quite close together so that she could complete gestation and lactation and move forward with both risk-reducing surgeries:

I think we would have probably spaced another half a year to a year out. We always knew that we wanted to do either zero or two [kids], so once we had our first daughter, we talked about having a second. The ideal situation would have been two-and-a-half to three years apart rather than almost two, but the mutation was kind of a big factor in that, for sure.

Acacia, 30

For some women, the specific decision to have children closer together was made because their mutation was identified after starting a family, so that commencing childbearing earlier was not an option. Thus, Rachel, who felt an extra sense of urgency about having her second baby after learning she carried a *BRCA* mutation because she had required assistance from a fertility specialist to conceive her first baby. She recalled discussing the timing of a second baby with her husband:

When I told him the results, I said, “Well, if we want to have a second one, they want me to have these surgeries before I turn 35. If I’m going to do that, and it’s going to take us three years to get pregnant again, time’s ticking.” So I just sort of presented it that way and said, “I’m going to call the infertility doctors again and see if they’ll take me back,” and he’s like, “Yeah, whatever.” So I did. And we did.

Rachel, 33

Interestingly, Rachel attributed her husband’s seemingly disinterested response to his normal manner of handling difficult topics, and was not at all troubled by his lack of input. She was simply happy that he went along with the new timeline she had suggested.

The mother of one when she learned about her mutation, Marie explained her decision to move her plans for a second baby up by several years, and how that idea was received initially by her partner:

Our whole plan before we knew about this whole genetic thing was, we would probably wait until she was five or six and we would either be finished with the PhD or be closer to being finished. But now I don’t feel that way. I feel like I really want to have another baby. I’ve always wanted to have two kids. As soon as I got those results I told [my husband] that I’m going to stop taking birth control and I really want to have another baby. And he was kind of panicking because he is looking for a job right now ... he is transitioning from finishing school to being in the job market. We’re pretty much completely unstable as far as finances and having anything concrete that you could depend on. It pretty much sounds like a real crazy idea, because basically I would be trying to get pregnant over the summer, continuing teaching, pregnant, and doing my coursework and everything, and doing my research. But from my recent experience of cancer and seeing how life is so short, I think that that’s the most important thing to do. I’m always talking about trying to get pregnant in this window, and it has everything

to do with the mutation, because that decision was made based on the results from the genetic tests.

Marie, 28

For Beth, who unexpectedly became pregnant before she was married, thinking about the timing of a second pregnancy overlapped significantly with her high perceived risk of cancer, which stemmed from her recent experience watching her older sister battle chemotherapy and mastectomy after a breast cancer diagnosis:

I would want to start trying immediately after we get married. I don't think it would take us a long time, but if it takes us a while to get pregnant then we'll have time. Just knowing that I'm going to have to have my ovaries out... just to give us enough time to have a second child. I kind of do feel like I have to hurry up and have an oophorectomy. I just don't want to put it off and put it off and then what if I get ovarian cancer when I'm 35 and I was planning on having an oophorectomy when I'm 35 ½? So I do feel like everything is a little more rushed than I would like.

Beth, 30

Shannon noted how her *BRCA* mutation was one of several factors influencing her and her husband to proceed with a second pregnancy soon after their first:

[The mutation] was definitely not the only factor. It was probably more the fact that it took a little while to conceive my son and I didn't want to go through that waiting again. That was probably the biggest factor. And then, we knew we wanted a second child and we were already living in a home we planned on being in a long time. So, as far as stability goes, we were ready to provide that. And then, having the additional information about the gene mutation was just an added factor to all of that.

Shannon, 31

Finally, Rachel recalled her initial belief that being a mutation carrier should not translate into an accelerated pace of family formation, and how that changed after she and her husband thought carefully about the implications of having had to use fertility drugs to conceive:

It was almost, in a roundabout sort of way, a little bit of denial because I thought, "It's not going to change my life right now." But then it did, because I decided that with all of the information I had been presented, that perhaps we needed to

move our family on the fast track. And I was also referred to a gynecologist oncologist, and he started talking about the risks and what he wanted to do, and the fact that I have PCOS¹⁵ which meant that with both my babies I was on a significant amount of fertility drugs ... there is still no real knowledge as to what being on the fertility drugs does to you. It's great that they got me pregnant, but especially in a *BRCA* patient, is that increasing my risk? And we still don't have the answer to that. But that was definitely something that this gynecological oncologist made me take a step back and think about. So with that in mind, we put the second baby on the fast track and when my daughter was only 51 weeks old, I went back into the fertility program to try to get pregnant a second time. I can tell you I didn't want two kids in 20 months, which is what I had. I wanted time to enjoy it being my daughter and my husband and myself. I wanted time to figure out how you deal with having a baby. But finding out that information, I truly felt like I was a ticking time bomb. I decided that was something that I needed to move along and make it happen a little bit faster than what I had originally intended.

Rachel, 33

Rachel's experience is analogous to women and couples choosing to proceed with family formation before they were ready. Rather than *starting* her family early, Rachel chose to *expand* her family sooner than planned, with clear implications for her relationship with her daughter and her idealized notion about what the early years of parenthood should be like.

For some participants, having children closer together was chosen because it would allow them to be flexible with total family size. For example, Wanda had a clear age in mind at which she wanted to be finished with childbearing and proceed with RRSO. Unsure about whether they wanted to have two children or three, she and her husband chose to have the first two closer together so that there would be enough time remaining before the deadline age to have a third child if they decided to do so:

I think even with the first, we knew the clock was kind of ticking. My mom was 37 when she was diagnosed, so we want to make sure that we get our kids in before that. And then ... between the first and the second, I think we both might have preferred to wait another year, but not knowing if we want to have three, we figured we've got to get them in now. [If I didn't have the mutation], I think we'd

¹⁵ PCOS = poly-cystic ovary syndrome

have one at least, and I don't know if we would have had the other one this soon. I want to have my ovaries removed when I'm like 35, is what I'm thinking now. And each year it gets closer and closer to that 35. I mean, obviously if we end up having a third, two years from now, I would breast feed and wait until I was done with that before I would have my ovaries removed.

Wanda, 32

Other participants reported not having changed their ideas about how closely to space subsequent pregnancies, but appreciating the benefits of having their children closer together as part of their pre-existing plan so that they could move forward with risk-management strategies not conducive to ongoing childbearing. Shannon provided an example of this when she stated:

We did have our two children pretty close together, but it didn't have anything to do with that, per se. We were just ready to have another one. Our first child took a little longer than we had thought, so the second time around, we knew we wanted another one, so we just let it happen whenever it happened, and it happened ... a lot quicker than the first one. But that worked out, too, because then I was able to start thinking more about having surgery after my kids were born.

Shannon, 31

All of these coping strategies illustrate how important time management became in making BRCA-related decisions, how pressured they felt as this process unfolded, and what a relief it was when there was unexpectedly more time available than they had estimated. Happily, participants were often able to identify positive aspects of having altered their family formation plans in response to the demands of their *BRCA* mutations, as Marie noted:

I guess the only positive thing would be maybe that we are going to have another child soon instead of putting it off. I see other friends and couples that put it off because it's not the right time, and then maybe they'll never have a chance to have another child because they put it off too long. That's kind of a blessing. No matter what crazy things are going on in our life, we're going to put family first.

Marie, 28

Independent family formation. The prospect of not finding a desirable partner before one's perceived window of opportunity for childbearing closes may stimulate thoughts of how to get pregnant without a permanent relationship partner. This consideration is perhaps the most powerful manifestation of perceived need to prioritize managing risk over one's preferences for family formation. Only two participants spontaneously discussed this topic, and their perspectives provide a useful contrast. For Isabelle, considering having a child without a partner was a significant departure from her previously held ideals about family formation, but it nonetheless was an acceptable option if she were still unmarried at a certain age:

When I was much younger I always said that I wanted to be married, possibly pregnant by 21, because I just want to have a lot of kids. But after getting to 18, I realized how ridiculous that was. Now, at 22, would I have liked to have been in a relationship and possibly thinking about getting married? Yeah. I want to say by 25 or 26 I would like to, if not be married, at least be in a serious relationship, or moving there. If not by 30, I know I would already either adopt or something, or I don't know about artificial insemination, but at least on the boat to getting a kid on my own.

Isabelle, 22

Serena had also considered having a child on her own, but did not find it particularly appealing:

Artificial insemination or [surrogacy] or something like that [is] a possibility. I don't think I would actually do it though...because for me if I'm going to have kids, I want them to be a part of me and somebody else, somebody that I truly care for and love. So, I think that for me it would be really tough to do the artificial way. I don't think I'd do it.

Serena, 30

It is interesting to speculate how *BRCA*-positive women might view these options as they approach the age at which they feel an RRSO should be done; perhaps the sensation of dwindling time for biological family formation would make independent family formation options such as artificial insemination or surrogacy more attractive.

Considering Isabelle and Serena, it is remarkable that the younger is the one who was open to independent childbearing; perhaps as women approach the age at which that choice must occur, it is a lonelier, scarier experience and therefore less appealing.

Family Size

While changing or renegotiating the timeline on which family formation will occur is common for young women with mutations in *BRCA*, it is not the only strategy participants reported using to complete their families in an expedient manner and proceed to surgery. Many participants also reported having considered how the number of children they wished to have might be influenced by their mutation status. Participants who discussed decisions about family size generally approached this decision in one of two ways: by making their risk-management decisions in part based on their ideals about family size, or by regarding their plans for risk-management as urgent enough to warrant changes to the number of children they planned to have.

Prioritizing family size over risk-management. As is well understood in the *BRCA* community, choosing to have risk-reducing surgery during one's childbearing years has powerful implications for family formation. Young female mutation carriers are typically advised by their physicians to postpone RRSO until they are certain they have had as many children as they desire. For some women, this simplified short-term, risk management decision-making. Rachel recalled:

At that point, I only had one baby, and all of these decisions were being presented to me, but I wasn't ready to make a decision on any of this kind of stuff because I still wanted to try to have a second kid. So it was almost like, "Yeah, I've got the mutation, but I'm not doing anything about it, so carry on. I'll go in for the six month checkup appointments if that's what they feel I need to do but I'm not doing anything." So in a roundabout sort of way, it was almost a little bit of denial, because it's not going to change my life right now.

Rachel's certainty that having either risk-reducing surgery was inappropriate when she learned of her mutation was based on her choice not to have surgery before completion of childbearing¹⁶. Rachel also shared her thoughts about whether additional children would be a part of her future given the difficulties encountered in conceiving her first:

I knew I wasn't done trying to have kids. It was so much work to get my daughter, I had no idea I was going to have a second. But, I knew that I wasn't ready to throw in the towel yet.

Rachel, 33

For some participants, making final decisions about the total number of children desired was the last remaining step before they could proceed with surgery. Wanda described her ongoing discussions with her husband about whether her current pregnancy, their second, would be the last:

We're trying to figure out how many kids we want to have, and we talk about, "Okay, when we're done having kids, what are we going to do with me to make sure that I'm okay." We're pregnant with our second child right now, so we have been talking a little bit here and there about when we are going to decide that this is it, or we want one more? Because that's about the point where we would probably go for the oophorectomy.

Wanda, 32

Like Wanda, Shannon was certain that she would eventually undergo RRSO, but was unclear about the timing because of lingering uncertainty about family size:

The recommendation was before the age of 40. Or if I know that I'm done having kids, then do it at that time. So, it'll definitely be before I'm 40. It's just that we're waiting to make that final decision about having another baby or not.

Shannon, 31

As described in chapter three, Audrey unexpectedly learned that she was *BRCA* mutation-positive through genetic testing of her young son with Fanconi Anemia. He died at age two, following which Audrey and her husband struggled with whether to have

¹⁶ RRBM is an option during this period for women who are willing to forgo the opportunity to breastfeed subsequent children.

additional children, and how many. She shared her memories of delaying RRBM after unexpectedly learning that another baby was on the way:

I was planning on doing the mastectomy after my daughter, but not the oophorectomy, and perhaps have more children. At that point, after my daughter was born, I think we were even almost done. It wasn't even so much about not having any more kids. I really think that we were both getting to the point that we had our daughter, she was healthy, she wasn't a carrier. . . . we can stop it. And then, about the time I got ready to say, "Okay, I'm going to start really getting more serious about this and what do I want to do? And how do I want to fix it?" I found out I was pregnant with my son. So then I was like, okay, well I'm not going to do anything right now. And then, after my son was born, I said, okay, now I'm really going to do something again. And so he's getting ready to turn a year old, and I am six weeks from my prophylactic bilateral mastectomy. God willing, he just decided that he wasn't finished. And gave us another son. And he was healthy – he was a carrier, but healthy. I guess [God] decided we just weren't quite done yet. But, we definitely are now.

Audrey, 28

Audrey was scheduled for RRSO shortly after her interview, as part of the same procedure during which her breast tissue expanders were replaced with permanent implants.

Prioritizing risk-management over family size. Perhaps even more powerful than having one's risk-management decisions influenced by family size, the reverse (*i.e.* having decisions about family size influenced by demands of risk-management) occurs for many women. Many participants experienced a time during which they questioned the appropriateness of having (additional) biological children, in the aftermath of genetic testing. Several participants cited their feelings that having more children would delay surgery, and this was a concern for their own health; others spoke more from a perspective that *not* having more children would permit surgery sooner. Chris wanted another child with her second husband, but he was significantly older than her, and this

desire was not congruent with his life plans. This age disparity added another complicating factor to Chris's decision about whether or not to undergo RRSO:

He said that if he was twenty years younger, he would want to have kids...he's twenty years older than me. I had pushed him to get a vasectomy, and before that we had thought about freezing some sperm, just in case [we] changed [our] mind[s]. But I kind of know that he didn't really want to have kids at his age, he wants to retire...It's kind of hard because I think about having kids. I would like to have had another child. I knew that that wasn't going to probably happen with him...But on the other hand...what if something happened? I was only thirty two. What if I got married again? Would I want another baby? Would I want to start over? Well, probably, yeah. But in the situation that I was in, married, he's twenty years older than me, probably not. But, you just sort of never know, I guess, where life is going to take you.

Chris, 33

Ultimately, Chris and her husband decided that having a child was not the right choice for them, and Chris proceeded with both risk-reducing surgeries. Her clarity about wanting to start over were she to find herself in a different relationship in the future suggests that were that to occur, she may experience regret about having ended her fertility potential *via* RRSO.

Sadie related a unique story about deciding whether to have children and its intersection with risk-reducing surgeries. After deciding to try to have a baby sooner than originally planned because of Sadie's desire to have RRSO, she and her husband encountered significant infertility-related challenges. They tried solutions ranging from medical to homeopathic, and eventually:

We had cut our losses and said, "Okay, let's face the facts, four years, we have never ever gotten pregnant. This is probably it." The idea would be that we would just hang out for a while, save some money and then go down the adoption road.

Soon after making this decision, they realized that life without children might have its advantages. They focused on opportunities for travel, financial flexibility, and greater freedom that having children would not allow, and decided that if they subsequently

changed their minds, they could adopt. Making that decision allowed Sadie to move forward with RRSO at the age of 33, significantly younger than most *BRCA*-positive women. Sadie's sense of fulfillment at having weathered this journey and having emerged with a set of decisions that truly felt like a good fit was remarkable:

Ultimately, that feeling is just about me, really, and about coming to terms with that image that I've always swallowed hook, line and sinker my whole life is that in order to be a selfless person, we have to have kids and procreate and all that stuff. It's been an interesting ride, definitely. But I am much more excited about being able to travel and see the world and spend time with my husband and be with him than I am about bringing in adopted kids from somewhere. And that may change. You know, when we get to be 40 or 50, we may be like, 'oh, that's the real meaning in life, having kids.' And if that happens, we can adopt. There's not a timeframe on that.

Sadie, 33

Although her sense of urgency regarding risk-reducing surgery was not the only factor in her decision not to have children, she clearly viewed the ability to pursue RRSO without regret as a positive outcome of her anguishing about family size.

Trixie provided a final example of deciding *not* to have children because of one's mutation status. Her decision stemmed from her experience watching her oldest sister die from breast cancer at age 32, four years after discovering a malignant tumor while breastfeeding:

I was 20 years old and my sister was diagnosed with breast cancer out of the blue. She had just had a baby and they thought it was an enlarged milk duct. So, still I thought it was nothing. It sounded normal enough, from what my parents explained to me. And then it hit us like a ton of bricks when they actually came back and said it was cancer. We did not see it coming.

Trixie then explained how her ideas about having children shifted toward remaining childless very early in her adulthood, even before she was aware of the *BRCA* mutation in her family:

Even though I didn't know my status, I've known I was high-risk since I was 20 years old, and I really think that has played a role in my not wanting to have kids. I think because of my parents, the guilt that they feel and how sad they feel, I don't want to feel bad. I don't want to pass it along to anybody. I'm glad that my siblings have kids. I think they're great. I love being an aunt, but myself, I just don't know if I could deal with it. And then watching my parents watch their child die... I don't know how people do it. And I think, subconsciously, through this whole process, that is what has made my mind up about kids. And [Mark] is also a very independent person, he's just a take-off-and-go kind of guy. And he's in his mid-40s so he has already passed that phase of his life where people normally have kids, and it just never happened for him. Never had the desire.

Trixie, 27

Trixie's partner choice seems related to her desire to remain childless: she had selected a partner who was old enough to feel certain that he also did not want children. Having created a situation in which her decision to remain childless was not only accepted, but embraced, Trixie completed RRBM at age 27 and was actively considering RRSO when interviewed. Her relief at not having to worry about facing her sister's fate was clear.

Aside from choosing not to have children, several participants reported having less drastically altered their ideas about how many children they would have based on their desires to employ surgical risk-reduction, and some participants who had not yet commenced family formation noted that they had considered this option. For example, Rylan noted that the number of children she and her fiancé ultimately decided to have would be an important consideration in allowing her to move forward with RRSO:

I would love to have twins, that would be great because I do think we want to have two or three kids and the faster we can have them the better – then I could get my ovaries out. So that'll be an issue, number of children, rate of birth, and adoption will be issues we might deal with in the future. We've talked about how it would be great if I could have twins and then have my ovaries out, and then if we decided we wanted more, we could adopt, or try to have one or two and then adopt more if we wanted. I think he wants at least one of his own, and then maybe I can negotiate with him at a later date about how many he really needs. I don't really know how many kids I want right now, so I have no idea if I'm going to like motherhood or not. If I don't, I'll probably just have one.

Rylan, 34

For Rylan, decision-making about family size was important not only because of her timeline for RRSO, but because she and her fiancé differed in their preferences: being from a big family, he preferred to have several children, while Rylan felt she might be happy with only one.

While Rylan was contemplating family size from the perspective of being childless, Jane provided insight into how decisions about family size might be impacted in women who have already begun childbearing. She reported that she and her husband might decide upon a smaller family than initially planned before learning that she was mutation-positive:

When I found out about my mutation, we were already trying to get pregnant. Now we would like to have another child, but beyond that I don't know that we will have more than one more, whereas before we might have considered having more than just two children. Before it was kind of open-ended, let's see where we're at with kids as we have them, and then maybe see if we want to have more. And now it's kind of like, well, we'll just have one more. And I read that the risk of breast cancer with *BRCA2* increases with more pregnancies¹⁷, so that's scary to me. And then the risk of a diagnosis is greatest within two to five years after a pregnancy. And I'm not sure if that's related to hormones, or what the exact reasoning is for that. And then I think about whether or not to have my ovaries removed, and I know that the ideal is to have it before the age of 35, so that kind of all plays into it as well.

Jane, 30

Jane raised several important themes here. First, she described that her flexibility in family formation was reduced by being mutation-positive; rather than making *ad hoc* decisions about childbearing, Jane and her husband felt compelled to agree on total family size *before* they actually reached that goal, so that risk-reduction planning could begin well in advance. Second, she referred to another important dynamic surrounding risk perception and its connection to pregnancy and childbearing. Jane is concerned not only about how multiple pregnancies might increase her cancer risk, but also about

¹⁷ This belief is *not* definitively supported in the medical literature.

increasing the length of time during which she would remain at high risk if she were to continue having children. This will be discussed in greater detail later in this chapter.

Women who had completed childbearing recalled how this issue had played out for them. Having learned that she carried a *BRCA* mutation when she already had three children, Leigh shared her sense that the mutation was only part of her decision to have no more:

If I wanted more, maybe I would have done it anyway, but it was sort of a small factor in favor of not having any more children. I'm pretty sure it wasn't the deciding factor. Because I don't think I could handle any more under any circumstances right now.

Leigh, 35

Similarly, Elaine's knowledge about her mutation pushed her to decide definitively that she wanted no more children:

One of the frustrating things in genetic counseling is that, over and over again, the genetic counselor and every provider says, "just make sure you're done with child bearing before you go ahead and have an oophorectomy." And it's like, "Gosh, I was thinking if I was going to have another kid I was going to wait until my late thirties." They don't want to hear that actually this has made me decide not to have more children. But that's a logical choice that I may not want to risk pregnancy, where your risk is just a little bit elevated, not having screening ... Why risk it when I have three healthy kids? It's even part of the consent, they ask you if you're done with your child bearing. I understand they don't want to take healthy ovaries out of a woman who may not be 100% certain, but this has made me certain. I wish that I didn't have this and I wish that I could contemplate another child but this is a consequence of knowledge of this mutation. I think even before we [found out about the mutation] we were saying, "Okay, we're not going to do this." I think we were feeling like it was going to be too much. I think it would have been too much for my marriage and so even before I got the news, I had given away baby stuff. So I don't want to make it sound like, "Oh, this made the decision." It's probably the nail in the coffin. You know, you can give away baby stuff but you're still fertile.

Elaine, 34

Finally, making decisions about surgical risk reduction led some participants to doubt their pre-existing ideals about desired family size. Beth reported that what had

seemed like an autonomous family size choice now felt like a decision imposed by her *BRCA* mutation:

Before this whole cancer thing I always just wanted two kids, and he just always wanted two kids, so that was great. Now, knowing that I'm going to have to take my ovaries out, and that once I do there's never a chance to have another child, I keep asking him, "Are you sure you don't want to have three kids?" And I don't even know that I want three kids, *per se*, because of the expense and it would just be a lot, but just knowing that once they're out, they're out and there's never another chance to have another kid – I'm sure that until I have that procedure done I'll always ask him, "let's have three, let's have three." It's one thing if I have my ovaries we can both say, "We're done, two kids is great, it's our choice." But by taking the ovaries out it's like it's not really my choice. It's my genes, it's the mutation, it's the risk of cancer, the choice has been taken away from me and it's all about the cancer. It's not about what I want, or what he wants, it's about the cancer.

Beth, 30

This is a powerful conclusion. As *BRCA* mutation carriers make decisions about their families and relationships based on cancer risk management priorities, their frustration over losing autonomy could be a potentially important phenomenon that might be usefully addressed by mental health professionals.

Breastfeeding

Preferred timing of RRBM and the breastfeeding choices women make comprise a parallel set of problems to those just discussed. Weighing recommendations to complete RRBM as early as practicable against the desire to breastfeed – universally accepted as a gold standard for raising healthy children (Bonati & Campi, 2000; Oddy, 2001; Scariati, Grummer-Strawn, & Fein, 1997) - creates a heart-wrenching decision for women who are struggling to make the best health care decisions for themselves and their children. The importance of breastfeeding as a unique and powerful bonding experience with their babies - an experience that some consider irreplaceable - cannot be denied. Worry about being able to provide the healthiest nourishment for their babies without the

ability to breastfeed was also a major concern, and this emerged most strongly from women who did not yet have children; they often idealized breastfeeding and powerfully lamented its loss. Participants typically selected one of three distinct paths: completing all childbearing and breastfeeding, and then proceeding with RRBM; completing RRBM prior to commencing childbearing; or undergoing RRBM in between pregnancies.

Delaying RRBM until childbearing is complete. For some women, the desire to breastfeed clearly outweighed their urgency over mitigating breast cancer risk *via* RRBM; these women did not consider having RRBM until childbearing was complete. This decision seemed quite straightforward to Sophie: “I obviously don’t want to get any surgeries before having children.” Period. No ifs, ands or buts. Others, like Isabelle, who was still several years away from commencing childbearing when interviewed, discussed her wish to allow her breasts to fulfill their purpose before having RRBM:

I do love my breasts, and I love them even more now, and do I really want to get rid of them before I get a chance to enjoy them for what they’re for? ... I always knew that I wanted to have a lot of babies, and I know breastfeeding doesn’t always work for everybody, but that was always something that I wanted for me and my child, and would I be able to deal with the fact that I wouldn’t be able to do that later on in life? That’s also keeping me from rushing into prophylactic mastectomy. It definitely is a worry of mine. It’s like, well, God gave these to me, and I want them to do their job before I have to take them off.

Isabelle, 22

When participants chose to complete childbearing before undergoing RRBM, they often did so with a sense that they were risking their own health to do so. This was illustrated in Chapter 5, where Melanie discussed her awareness of the limitations of breast surveillance during breastfeeding, and how influential that information was in how she would time pregnancies and decide how long to breast feed. Similarly, Shannon

shared her perception of her husband's concern about these issues, and his eagerness for her to finish breastfeeding and move toward surgery:

When I was breastfeeding our second child, he actually asked when I was going to be done with that so I could start making my doctor's appointments. I know it weighs on his mind, too. And that was kind of my thought, was that I was going to finish breastfeeding the kids and then move forward from there.

Shannon, 31

Spacing between children, desire to breastfeed for as long as possible, and ability to screen effectively during childbearing years intertwined in Shannon's decision-making. Many participants reported feeling much more confident about moving forward with RRBM after finishing breastfeeding. Marie, whose change in plans regarding spacing between children was discussed previously, described her confidence that she would be ready for RRBM after breastfeeding her next child:

I think I probably talk about it every day. Because with this whole plan that I made for myself – which is still the same – I had said that if the results came back positive, I would want to have another child immediately. And then, I'm an adamant breast feeder so I really would want to breastfeed for a year or 14 months or whatever, and then get the mastectomy done.

Marie, 28

Rachel was several steps further down the same path; having already breastfed her two children for a satisfactory length of time, she described her readiness to undergo RRBM:

I nursed both of my kids. Breastfed them both, and at the time when I was done, my son started to become a biter. So he actually got the breastfeeding taken away a little earlier than his sister did, because mommy didn't have the patience for that. And once I was done nursing, I was kind of like, "What do I need them for anymore?"

Rachel, 33

Consequently, Rachel was actively moving forward with RRBM at the time of her interview.

Pre-pregnancy RRBM. In contrast, another group of participants preferred completing their RRBM prior to their first pregnancy. Women choosing this option were quite heterogeneous in age and relationship status. They reported that after weighing the pros and cons, their sense of urgency to proceed with RRBM outweighed their desire to breastfeed their children. For some, this was a straightforward decision; others struggled to choose between these two important priorities. Monique was still actively considering pre-pregnancy RRBM when interviewed. With high near-term perceived breast cancer risk due to the very young age at which her maternal aunt was diagnosed, she discussed the pressure she felt from her family members to mitigate this risk by removing her breasts prior to having any children:

A lot of people in family want me to have some surgery and I don't know if I'm ready. You know what I mean, if I have a kid, breast feeding and stuff. So that's the hardest decision, if I want to do this now or afterwards. I mean if I have to, I guess I will do it now, but my doctor said to wait so far.

Monique, 26

Like Monique, Charlotte was considering pre-pregnancy RRBM under significant family pressure:

Previously, I was set on wanting to have children and wanting to be able to breastfeed ... once I was able to do that, I would go ahead and have the mastectomies. Now I'm definitely getting a little stressed and tired of all the testing and wondering how much longer I want to do this. And there's a little bit of pressure from my family, so now I'm considering doing it at a younger age and not necessarily having to breastfeed my children. I've talked about it with my good friend who lost her mom to cancer. It comes down to which risks I want to take – do I want to put myself at increasing risk every day, every year and not being there for my children? Or reducing the risk now being there for my children, but not having that breastfeeding experience?

Charlotte's inclination toward pre-pregnancy RRBM was influenced by her family, her desire for longevity, and escalating "screening fatigue." This latter phenomenon, described in Chapter 6, is driven by cumulative experiences related to periodic breast

surveillance and the anxiety accompanying inconclusive results, repeated imaging and the occasional biopsy; this is commonly observed in young women who choose to rely on surveillance to manage their risk. Continuing to describe her shift in thinking, Charlotte went on to state:

I look at it now as a smart choice, that I'd be reducing the risks. And I'm actually going to go meet with [my cousin's] doctor in a couple of weeks ... just to learn more about why he thinks 27 is the right age for me. So, I think if I were to lean toward that decision, it would just be because it seems like the smart thing to do, to manage my risk. But I think when it came to the point where I had children, I would definitely be pretty emotional that I wouldn't be able to have that experience or provide that to them, because from my point of view, it can be healthy for them with their immune system and things like that. Going into it I think I would know that it would be hard at the time and maybe want to talk to some counselors about it or how to get through that just because I think I know that it would have a pretty big effect on me.

Charlotte, 26

Charlotte's perspective suggests that she understands the gravity of the decision she faces, and is anticipating that the ramifications of pre-pregnancy RRBM might be difficult to deal with when she eventually has children. Her spontaneous identification of the need for psychosocial support suggests an unmet need for this population:

comprehensive and specific psychosocial support from a provider who understands these complicated dynamics and decisions should be available to women in this population not just in the phases in which they are making these decisions, but also in an ongoing fashion as they continue to face challenges (both expected and unanticipated) brought on by decisions they have made in the past.

Pauline also had not yet begun childbearing, and was contemplating pre-pregnancy RRBM. She candidly discussed both her personal desire to breastfeed and the societal pressure she felt to do so:

The loss of the ability to breastfeed is something that has definitely crossed my mind. I guess most women of my education or/and social class tend to breastfeed their babies and tend to do it in a sort of public and very boastful way. I do regret that I'm not ready to have kids immediately because if I were I might have a baby, I might be pregnant right now, just so I could go through the experience of at least breastfeeding one. But that hasn't been enough to cause me to change my views and my time frame that I'm comfortable with about having children. And you can't really breastfeed a child if you are going through chemo or radiation or if you're dead. So the functionality and the purpose of breasts in terms of reproduction, I'm not going to get the experience, and sure, I'm bummed, but that's not going to really rule my decision. It just hard because I think that breastfeeding is very in vogue right now in terms of educated, upper middle class women. [There's a belief that] people who don't breast feed aren't doing what's best for their child. I sort of anticipate the moment when I'm at the play lot five years, ten years from now and a mother I don't know very well says to me, "Oh, you don't breastfeed?" sort of in a sneering way and I say, "Well, no I don't because I had a double mastectomy," and sort of the show-stopping quality of that statement. That'll be something I confront. But I've been assured that women throughout the ages have still been able to bond with their babies even if they've been bottle-fed. If it was not in vogue right now to breastfeed, maybe I wouldn't even think twice about it. But there's just sort of this idea of good moms who eat organic and you know, drive safe cars, and have a graduate degrees, you know, they all breast feed their babies. Well, I can't do all of those, unfortunately. But that also sort of allows a little bit of egalitarianism in the way that I actually wish to parent in the context of my relationship. If I'm not breastfeeding, my husband can also help me feed the baby. That's something that I think is important for both the way that I hope to conduct my marriage and our co-parenting but also something good for [Glenn], that he could have that bonding experience as well.

Pauline, 30

Pauline highlights a neglected aspect of this weighty decision: women who choose pre-pregnancy RRBM may face questions and criticisms from others regarding their risk-management and family formation choices. Pauline's insight that there are also parenting and relationship *benefits* to the choice that she is making is an adaptive way of coping with the doubts she anticipates in response to outsiders' possible reactions to her choice.

While Monique, Charlotte, and Pauline provide excellent examples of women early in the process of deciding to complete RRBM before childbearing, other participants were further along this path. Marjory and her partner had decided

conclusively that mastectomy prior to childbearing was the right choice for them; they had moved on to talking about the length of time they would want to wait after RRBM to attempt to conceive for the first time:

We'll start as soon as we can. So, [early] next year. ... That's one thing that I have to find out about. I've actually been wondering about that. Obviously there's some recovery time. I've kind of wondered, is that putting my body through too much in a short period of time? I know surgery is pretty major. And then, to become pregnant shortly thereafter that, I don't even know what the recommendations are. We pretty much want to start right away. So whatever the recommendation is, I imagine we'll start pretty much right after that.

Marjory, 30

Marjory nicely highlighted another issue that can arise for women who choose RRBM: when they choose to time this surgery prior to childbearing, the available remaining time to bear children before they reach the age at which RRSO is recommended is reduced, particularly if breast reconstruction is done. This may mean that although breast cancer risk perception is reduced post-RRBM, ovarian cancer risk perception may increase slightly, as a consequence of delaying RRSO.

Rose had recently completed her surgery when interviewed. She described the two major components of her decision to do pre-pregnancy RRBM:

The motivation was more two things, and the first one being that my husband and I are thinking of having kids in the next couple years, and there are some issues with monitoring [for] breast cancer and being pregnant. And, I didn't like those risks and I didn't also like the thought of having kids and then what if I got breast cancer when I had a kid or while I was pregnant. And the second one was timing. ... I had a really good, long stretch of time ... that just worked out for work where my entire building was shut down, so I knew that I wouldn't be responsible for anything. And for me that is a good thing and I honestly think I healed so much more quickly. All I had to do was focus on myself. ... And then also, the thought of having to do something like this in the future, what if I do put it off and then have kids, and do it in ten years. I don't want to do it when I have little kids. If I could prevent that why wouldn't I have the surgery now as opposed to later when I may have little kids running around?

Rose, 30

Rylan, who had also completed her RRBM when interviewed, recalled conversations with her fiancé about the tradeoff between RRBM and breastfeeding

When I told him I wanted to get the mastectomy, his first question was, “Well, you won’t be able to breastfeed our kids.” He was kind of concerned, he wasn’t happy about that. And I said, “Well, I don’t think that’s a big deal compared to getting cancer and possibly not being around for them, to bring them up, I think ... breastfeeding is something I’m willing to sacrifice. We can buy breast milk from someone who is lactating, or we can buy the synthetic breast milk. ...” And he said, “Yeah, but you won’t be able to hold them and the whole bonding thing.” And I was like, “Well, no. This kind of sucks and there are some things that aren’t positive about my mutation. And sacrifices will have to be made, but I think it’s worth being able to avoid cancer and probably save my life.” He was more concerned about the breastfeeding than the cosmetic thing. I think the breastfeeding thing is something that I’ve already gotten over. I think he’s accepted it, that it can’t happen now. He doesn’t understand how horrible cancer is to go through and the treatment, but from the things that I told him, he could see that, “Yeah, you’re right. It’s more important to avoid this than worry about being able to breastfeed or not.”

Rylan, 34

This is a powerful example of the remarkable manner in which some women share decisions about their own bodies with their partners, perhaps a unique feature of couples in which the female partner carries a mutation in *BRCA*.

Grace’s story of decision-making about and living with pre-pregnancy RRBM is a similarly powerful one. She related how her ideals about commencing childbearing immediately upon completing the BI study were altered, and how that change resulted in her decision to complete the surgery – without reconstruction – before attempting to conceive:

That last screening totally messed up my time frame. Our plan was to get the last clean bill of health with my last MRI and mammogram, and then we’d go to Ireland, drink a couple Guinness, and get pregnant or hope to get pregnant in those next months, and then have a baby, breast feed and then have prophylactic surgery. Instead, I got the abnormal result and they wanted to re-screen me after our trip to Ireland, which in my mind meant I had to put pregnancy off until I got that clean bill of health. And, getting that abnormal result, thinking that we

probably couldn't start at having a family, or trying, reality really set in that this is not something to just toy with.

Despite her knowing this abnormal test result might portend serious trouble, Grace was still focused on getting a "clean bill of health" (*i.e.*, screening with no abnormalities) and proceeding with childbearing, until

... my husband sat me down and had a little talk with me about how the plan needs to change and that I need to do surgery before having a baby. My initial reaction was to say, "Screw you." I just was not ready for my plans to change. But over the course of 24 hours, I really absorbed what he was saying and why ... It was about wanting to have me around and be healthy, and that outweighs the urgency to have a kid and that feeling of wanting to breast feed and wanting to have that experience. And, so we rearranged everything. We ... did surgery and recovery and then a month or two after surgery, started trying to start a family...

Grace shared her perspective on the impact of altering her plans:

In hindsight, I'm so glad the surgery's behind me before having kids and before going through pregnancy. The more I read about breastfeeding, the more I realize how difficult and challenging and painful all these things are. Even though it makes me sad not to be able to breast feed my baby, at the same time I know I can be a better parent, and even have a better pregnancy not having to worry about, "What is this change? Is this normal, is this cancer, is this something to worry about?"

Three days past her due date when interviewed, Grace was uniquely able to provide insight into her decision to forgo breast reconstruction. She related what it was like to be both pregnant and breastless:

I knew I wouldn't look like a normal pregnant woman because I don't have these large engorged breasts. [Jason is] like, "I actually think I like the breastless look better on pregnancy than with them, because it's just very clean and it really flatters your belly." It was just really sweet hearing him talk about that. He's had no issues with the way that they look or the way that they heal. Because even though they're straight lines they don't heal evenly, the middle heals before the ends and stuff, but he's been totally fine and accepting with it.

In addition to her husband's wonderful support, Grace's realistic perspective about her body, and interactions with other women who reinforced that she had made wise choices,

reinforced her feeling good about her decisions. Asked what it was like to visit her doctor's office and be surrounded by pregnant women with breasts, Grace replied:

To me, my body isn't this great amazing specimen that's on display for everybody else. It's a functional thing, so in some ways having gone through the surgery and having scars and already accepting myself as not as what's perceived as normal, or socially like a normal beautiful body, I've been able to accept the changes with pregnancy a lot easier, I think, than some first-time moms that might freak out about the stretch marks, or the changes. The other really cool thing is I was at one of these support groups, and I had shared how I was really nervous about being pregnant without breasts and how I sort of had to redefine what's feminine anyway, in my mind. I've got long blonde hair and I'm petite, so I've got that going for me, which is very feminine, but you just have to embrace different parts of yourself. And at the end of the group this woman came up to me and said, "I just have to tell you, you were talking about feeling feminine, there's nothing more feminine than carrying a baby." And I never thought about it that way, but it's so true that, yeah, that's what a woman's body is built to do, and what an amazing experience that I get to have that. So, it just made me think about my pregnant body even more in a loving, accepting way than I had previously.

As a final thought about not electing reconstruction, Grace stated:

I couldn't be happier, and [Jason]'s been really good. I mean, I think he would agree that not having to worry about whether there is something there, behind the implant, especially with pregnancy, your body's going through so many changes that, you know, it's like a clean slate. You can just look and see if there's anything there, and feel it so easily. I think with reconstruction I would have had a lot more worry about what's going on that I don't see or feel. He says it's aesthetically pleasing because it's just a very clean straight line, where with implants sometimes you don't really know what you're going to get.

Grace, 30

Grace's story clearly illustrates that couples can and do actively engage and debate with one another about how to accomplish both family formation and risk-reduction goals in the presence of a *BRCA* mutation. Allowing her husband's opinion to be so influential in decision-making about such a personal and intimate decision underscores their powerful and intimate connection: Grace's willingness to allow Jason to so powerfully shape her experience would not have been possible in a relationship that was not firmly based in trust, respect, and genuine love. Jason's ongoing support of Grace and his ability to

consistently reaffirm her decision further exemplify the presence of appropriate communion in this relationship.

RRBM between pregnancies. Some participants considered timing RRBM to occur between pregnancies. Often, this was described as a compromise: it permitted having the breastfeeding experience once, without delaying RRBM to the point of intolerable increases in perceived breast cancer risk. In addition, it offered a way to significantly reduce cancer risk without terminating fertility (as happens with RRSO). Acacia, who was seven months pregnant with her second and final child when interviewed, recalled having strongly considered RRBM between pregnancies, but ultimately deciding to wait:

I'm really happy because with my first daughter, I chose to breastfeed and it's a very wonderful bonding time. If I had the opportunity and I could do it where I felt I was still in kind of the safe age range, I wanted to try to do that. I'm pretty happy with my decision, thinking right now that I don't have any big cancer worries.

For Acacia, this decision was acceptable because she was able to alter her family formation timeline, both by having children sooner and having them closer together, so that she would complete childbearing before she reached what she perceived as the age when her risk would be considerably higher and require surgical risk-reduction. She further stated that in making the decision about the timing of her RRBM, she ultimately

decided maybe it would be better just to bump [the second baby] up about a half year to a year and have everything working and then after that baby was born just kind of do both surgeries, get it all done together. I think we talked about it for a good three months or so, pretty in-depth, before we came up with a plan, but that's ultimately what we decided.

Acacia, 30

In addition to weighing the ability to breastfeed her second baby in this decision, Acacia also embraced the relative convenience of having both surgeries simultaneously, thereby

minimizing the recovery time during which her ability to actively care for her children would be compromised.

Inter-pregnancy RRBMs also create some flexibility with regard to family size and timing of additional children. The decision to have RRBMs after her second pregnancy fit nicely with the ongoing debate between Shannon and her husband about whether and when to have a third child:

He's happy with two, and that doesn't have anything to do with the cancer risk or the surgery. Even before we had any children, he always thought that two would be a good number. I always leaned a little bit more towards three. But we agreed that this wasn't a major issue for us either way. And that we'd still be willing to revisit it after we had the two and go from there. And then, after we had the second one, we both agreed that the surgery is the bigger issue right now and that we could think about a third after that, if I was still thinking about it.

Shannon, 31

Timing her RRBMs at this moment allowed Shannon to keep the option of a third child open, while focusing on mitigating her breast cancer risk. Beth also discussed her decision to proceed with inter-pregnancy RRBMs:

I think not having that pressure will be nice ... realistically I'm sure some of the emotions might come back when I have another baby and I think to myself [that] I won't be able to give my baby this breast milk that everybody says is liquid gold. ... the breast feeding community, they kind of push it and they say if you breast feed your child has better immunities, they won't get sick as much. And I think my son has gotten sick three times in three months even though he has been getting milk. So, I don't know. I think it will be a relief that I don't have to and that I have a reason why I can't. Because now people almost expect you to breastfeed and I can't and this is why I can't. And I think giving my baby formula and them having a mother is better than me trying to keep my breasts, giving them breast milk and possibly getting breast cancer, and them not having a mother.

Beth, 30

Shannon and Beth both spoke from the invaluable perspective of already having one child and knowing what it means to be a mother. This likely contributed to their clearly expressed and powerful concern about the well-being of their children, and their ability to

see RRBM between pregnancies as a way to protect their current and future children as well as themselves.

Chemoprevention

The use of chemoprevention agents¹⁸ as a strategy to mitigate cancer risk is used by a minority of *BRCA* carriers who wish to reduce their risk during their childbearing years without terminating their fertility. Use of oral contraceptive pills (OCPs) (which reduce the risk of ovarian cancer in mutation carriers by about 50%) or the anti-estrogen agent tamoxifen (which reduces the risk of sporadic breast cancer by about 50%; limited data suggest a similar protective effect in mutation carriers) is more effective in reducing risk than screening (which does not actually reduce risk, but rather identifies existing cancers at an earlier, potentially more treatable stage), but less effective than surgical risk-reduction. *BRCA* positive women may consider this strategy either before or after completing childbearing; when the latter is chosen, it is typically a strategy for delaying surgery rather than a substitute for it.

Several participants cited their current or past use of OCPs as useful in mitigating their risk of ovarian cancer during young adulthood. Other medications were viewed with less enthusiasm. Annie recalled believing that chemoprevention was too risky and not the right choice for her:

For me, going down the whole preventative chemotherapy route and fertility and all that that entailed, that was just too much for me. That just wasn't something that I felt would work for me at all. I thought it would be more traumatic, not only emotionally, I don't know why I thought maybe even physically, that would be a little bit too much for me. But, I think the risk, too, of taking any kind of drug and ultimately not being able to have kids, [and what if] something went wrong or, I don't know. There just seemed to be too many variables with that *versus* having a

¹⁸ Chemoprevention is the designation given to the use of medications to reduce the risk of cancer.

preventative surgery...I think just the chance of not having kids as a result of chemoprevention stuff, that to me is a little bit more traumatic.

Annie, 32

Ultimately, Annie chose to have RRBM prior to her first pregnancy rather than taking the chemopreventive option offered by her physicians. In contrast, Grace discussed her plan to employ chemoprevention (OCPs) after she was finished with childbearing, in order to buy a bit more time before RRSO:

My theory is have a baby or two and then get on birth control, which I've never been on, to regulate the estrogen levels. I think that's what's supposed to reduce your risk a little bit for seven to ten years or something. And then have my ovaries out. I'd like to at least put it off until 45, because the issue with getting them out too early is worrying about bone loss and that kind of stuff. And he's on board with whatever. I think he just wants me around. Especially now being a mom.

Grace, 30

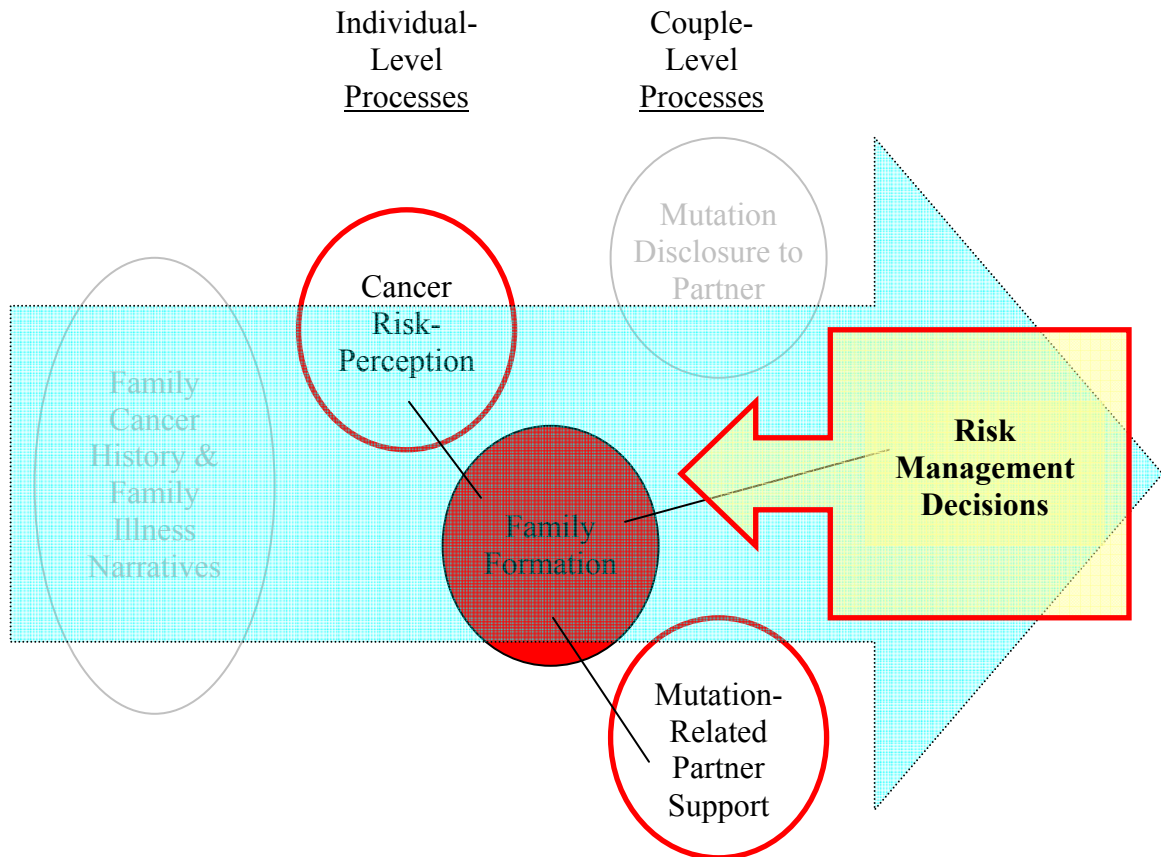
The dual priorities of maintaining her own general health (*e.g.*, delaying the cardiovascular, bone density, and vasomotor problems related to premature menopause) and surviving to be around for her children made chemoprevention at a later stage an appealing choice for Grace.

Chapter Conclusion

Data presented in this chapter powerfully support the notion that women's choices about family formation are intricately linked to perceived risk, partner support, and risk-management decisions, as illustrated in Figure 8. Regarding risk, mutation-positive women contemplate not only the risk that they might someday get cancer, but non-oncologic components of perceived risk—*e.g.*, the fear that they could die and leave their children without mothers, or simply being afraid of death. These additional risks may be just as powerful a motivator to intervention as simply worrying about developing a malignancy, and providers who can recognize these additional elements of risk and make

them part of the conversations they have with women in deciding about risk-management may be better able to counsel mutation carriers in a way that feels comprehensive.

Figure 8: Role of Family Formation in the Model



These data also illustrate several examples of misunderstandings or misinterpretations of risk-related information by carriers. It is clear that most of these are the product of a medical setting in which providers (even those who are up-to-date about the latest research) do not have as much information as they need to make evidence-based recommendations. Consequently, most provider suggestions are based on “best clinical judgment” rather than hard data, and providers often make the same assumptions about what must be true on these topics, as patients do. The most effective and personally

relevant counseling for these women will likely come from a medical team that includes multiple specialties (*i.e.*, gynecological oncologists, endocrinologists, surgeons), and women are unlikely to receive sufficient care from primary care physicians (or even ob/gyn physicians) alone. Therefore, medical teams comprised of a variety of appropriate specialists should be constructed and made available to young female *BRCA* carriers.

With regard to partner support, participants illustrated how their partners (either current or future) had been, or would someday be, involved in the decisions they made about how to manage their risk. Even women who were not in relationships revealed that they considered how their future partners might feel about decisions they were making while single. Mutation carriers involved their partners, to varying degrees and in different ways, in decisions about how to time family formation, how many children to have, and how to balance their desire to breastfeed with their desire to mitigate risk through RRBM.

The major theme that developed in analyzing data related to family formation was that of control: participants exercised control over the various facets of family formation to make way for their preferred method of family formation. There was wide variation in how women prioritized their risk-management and family formation goals, and women were most satisfied with their decisions when they identified and acted on a plan that resulted in minimal sacrifice to either set of goals.

Finally, data presented in this chapter are similar in important ways to previous research about “the disordered body” and how cultural discourses on femininity may cause women to feel inadequate when they perceive that a flaw or lack of ability would prevent them from accomplishing tasks such as childbearing and breastfeeding (Becker, 1997). Becker studied women struggling with infertility and found that they “equated

infertility with the ability to nurture. Their ability to nurture others, and by extension their fertility, were central to their sense of who they were,” (p. 84). Similarly, some *BRCA*-positive women in this study felt that their femininity, their identity, was threatened by the presence of a mutation that might render them less or differently able to bear and nurture children, or that they might have to sacrifice that ability in order to preserve their health. Prior to learning that they were mutation positive, many of these women expected that pregnancy and motherhood would proceed “normally,” in the manner that they had planned and predicted throughout their lives. Having to alter those expectations was jarring and difficult for some, and represented a life disruption. Some viewed themselves as deviating from the cultural ideal, as was illustrated by Pauline in her discussion of being questioned for not breastfeeding when she someday takes her infant to a park, or by Jane when she spoke about having her fertility threatened by her mutation. This sense that they are failing to live up to a cultural or structural expectation because of a limitation imposed by their mutation represents an additional layer of stress for young female mutation carriers.

As has been suggested previously (Werner-Lin, 2008), more research is needed to understand the importance of timing of genetic testing in the life course. Participants in this study demonstrated the very different ways that genetic information is understood, how meaning is made, and how decisions are shaped by awareness of one’s breast and ovarian cancer risk at varying points during young adulthood. Many of the women who were single or in casual, open, or unsteady/impermanent relationships when interviewed demonstrated a high degree of worry about how they would accomplish their family formation goals in what they saw as a limited amount of time. In many ways, the

strategies that are likely to lead to the best results with regard to cancer risk and survivability (early testing and intervention) and those that are likely to lead to the optimal mental health and relationship outcomes during young adulthood (waiting until one is in a significant relationship, and perhaps until after one has children, to test) are in direct opposition to one another. If these very young (*i.e.*, 18-24) women are to continue to be routinely tested for *BRCA* mutations when they are members of families in which mutations are known to exist, appropriate and ongoing psychosocial care is of paramount importance, so that these women may have a knowledgeable, caring professional with whom to speak about their concerns. These providers can also be of great assistance to older mutation carriers and those in relationships, providing both individual- and couple-based therapeutic services as mutation carriers move through the shifting dynamics and changing priorities of young adulthood.

CHAPTER 10: DISCUSSION

Data presented in chapters five through nine demonstrate the complex ways in which cancer risk perception, mutation disclosure in couple relationships, mutation-related partner support, and family formation are connected to each other and to women's decision-making about, and experience with, cancer risk management. Chapter Five provided two illustrative examples of women's journeys through young adulthood as mutation carriers, and outlined the basic theoretical model used to understand these data. In Chapter Six, participants illustrated how their perceived risk of cancer was shaped by what they knew from their family experiences. Specifically, many discussed how family narratives regarding the course and behavior of cancer were integral to forming their own beliefs about whether and when cancer might impact them. Other participants described how a *lack* of knowledge about cancer in their families left them feeling vulnerable and unsure about their own risk. They shared their concerns and uncertainties about how their perceived risk of cancer might impact their decisions about family formation, and how stepping back to consider personal risk in the context of one's own body, previous risk-related experiences, and historical context (*i.e.*, currently available medical knowledge and technology) allowed them to make risk management decisions that facilitated a sense of agency and regaining control over their lives from the mutation. Finally, their stories lend empirical support to the notion that psychosocial and genetic counseling care are needed throughout the mutation-positive experience, even after risk-reducing surgery. Patients' ongoing interactions with these professionals can help mutation carriers stay informed about current research, understand their risk over time, and minimize possible

misunderstandings about risk that may leave them feeling more vulnerable to cancer than they actually are.

Chapter Seven, with its focus on disclosure of positive mutation status within couple relationships, revealed how young women's choices about risk management shape their options for timing of disclosure to partners, especially in dating relationships: women who had already undergone one or more risk-reducing surgeries often felt compelled to disclose early, while those who had not yet undergone surgery had more flexibility in the timing of disclosure. For many participants, the act of disclosure itself was catalyzed by imminent risk management activities and/or the desire to have a partner as a source of mutation-related support. Additionally, women felt compelled to share information about their mutations with partners who might someday become co-parents, because of the obvious implications of *BRCA* mutations for the health of future children. When those already in serious relationships (long-term dating relationships or marriages) learned of their mutation status, disclosure often occurred as part of the normal day-to-day conversations between partners, as male partners were typically involved throughout the genetic testing process. Still, many participants in this latter category reported having had important conversations with their partners about the implications of their mutations on their plans for family formation, if childbearing was not yet complete. Regardless of their relationship status, participants reported that they were most fearful about their actual or potential partners' attitudes/responses regarding mutation-related decisions/interventions yet to come, while those that were *fait accompli* would be more easily accepted by partners.

Mutation-related partner support, the focus of Chapter Eight, emerged as an important component of the successful management of high-risk status for young women. Partners (both current and future) played key roles in women's decision-making about risk management, and in developing strategies to minimize disruption of family formation goals. Additionally, partners were described as advocating for carriers as they moved through the mutation-positive experience, encouraging them to keep up with screening, requesting important information from physicians, and reminding participants about shared priorities that necessitated some mutation-related action (*e.g.*, have a mastectomy so that we can have our family without worrying about breast cancer). A surprising finding from these data was that a substantial subset of participants reported feeling that their relationships with their partners were strengthened as a result of having worked through *BRCA*-related issues together—a phenomenon likely unanticipated by newly-confirmed mutation carriers or their health care providers. Challenges related to inter-partner communication and sexuality emerged as the most prominent stumbling blocks related to partner support; effective psychosocial intervention to assist relationship partners in speaking openly about their feelings and needs would likely be of great benefit to both.

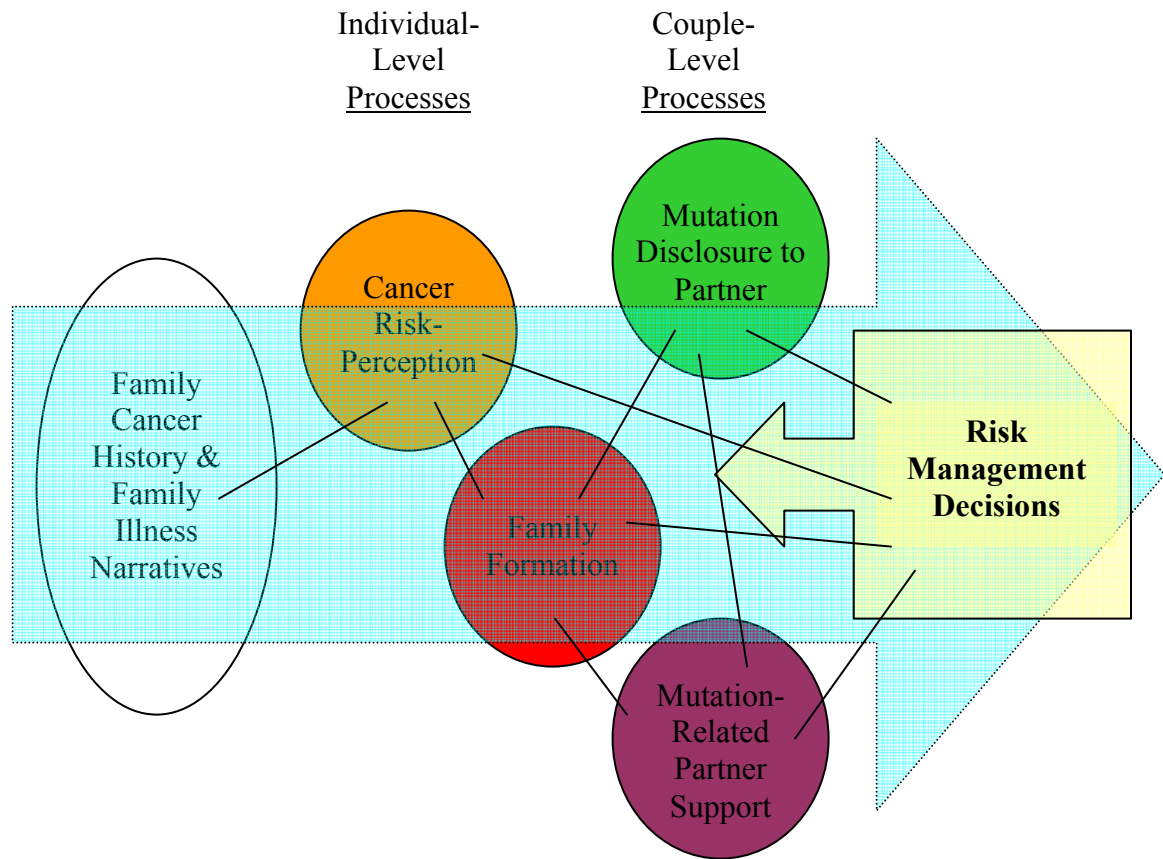
Finally, issues related to family formation were central among all participants, as demonstrated in Chapter Nine. Risk-related information was a frequent topic of these discussions, as women worked to make sense of the factual information and opinions of various providers, peers, and loved ones. The often confusing morass resulting from this process was difficult for some participants to navigate. Ultimately, most women made choices that allowed them to gain some control over their mutation by forging a path that

included their preferred course of family formation, or a close approximation of it. Prioritizing either risk-management or family formation on several different dimensions (timing of family formation, family size, and breastfeeding) was a critical strategy that many participants employed as they moved down this life path, often succeeding in a striking a balance that reconciled these conflicting goals.

As women reconciled all of these issues in deciding how to manage their risk, many of them ultimately decided that risk-reducing surgery was the best course of action; early RRBM was surprisingly common in this group, and my experience working with *BRCA* mutation carriers more broadly is that surgery at young ages is becoming a more and more common choice. Participants who had made this decision seemed to have done so in a way that makes sense if considered in the context of anticipatory loss: finding the possibility of living every day with knowledge of cancer risk and impending loss intolerable, they chose instead to endure a short, intense period of pain and loss (the surgical removal of their breasts and all that comes with that process). Even though they know that they could probably wait until they are done with childbearing and survive with their breasts intact, living with the fear of cancer is not worth that. These brave women choose instead to face something that most would find it difficult to conceive of, and in so doing free themselves from the emotional and physical constraints of their mutation-positive status and gain control over their genetic destiny.

Taken together, these four chapters provide empirical evidence to illustrate the complex set of decisions that *BRCA*-positive young women make, as illustrated in Figure 9. For women who learn in young adulthood that they are mutation-positive, the journey from understanding the meaning and role of cancer in one's family, through

Figure 9: Theoretical Model of Negotiation of Health Risks for BRCA+ Young Women



comprehending the implications of their carrier status for themselves as individuals, to making one’s own decisions about risk management, is not straightforward. Rather, it is a complex and unique set of interrelated compromises and considerations that require these young women to think hard about their relationships, priorities, and risks—and the different ways that those might fit together in the present or in the future—within the context of the relationships and societal structure in which they are embedded.

Theoretical & Research Contributions

As described previously, data from the current study contribute to our current understanding about the lived experiences of young *BRCA* mutation carriers in several important ways. The findings from this study helpfully inform each of the theories used

in its development. These data study sheds light on the depth of the relationship between intra-individual levels on Engel's Hierarchy, illustrating how powerfully cellular-level phenomena can impact a person and his/her broader contexts at all levels. For example, participants' knowledge of themselves as *BRCA*-positive was a powerful component of how they understood themselves, as well as how they thought about their breasts and ovaries and the ways that they might (or might not) use them. Experiences with breast and ovarian screening and knowledge of the ineffectiveness of ovarian screening specifically powerfully shaped women's ideals about the long-term suitability of screening and their decisions about whether and when to undergo risk-reducing surgery. With regard to the interpersonal components of the hierarchy, participants spoke powerfully about their experiences in real or hypothetical couple relationships, and shed new light on our understanding of how these important bonds absorb and adapt to the *BRCA* mutation-positive experience. When participants had negative experiences in couple relationships, they demonstrated how other levels of the hierarchy are impacted (e.g., by relying more on peers or friends for support in the absence of a supportive partner).

With regard to life course perspective, findings from this study powerfully illustrate how life trajectories can be altered effectively to accommodate significant and unexpected health demands. Consistent with feminist perspective, the life course concept of agency emerged as a dominant theme and effectively describes the empowering decision-making processes in which these young women (and their partners) engage to create a path through young adulthood that allows them to meet both relationship and health goals with minimal compromise to either. Further, women in this study often

found new paths that made something positive of their experiences as mutation carriers, such as by creating an educational or artistic product to inform other young carriers, or choosing to work in a field that would allow them to focus on cancer and/or cancer susceptibility. These experiences were universally described as empowering and represent creative ways of sidestepping significant constraints and limitations.

Women's risk-management decisions during young adulthood significantly inform our understanding of anticipatory loss. A striking number of participants had completed or were contemplating at least one risk-reducing surgery at the time of their interview; choosing this during one's twenties or thirties is a courageous decision, and one for which many participants had no model in their families or broader social networks. Despite their knowledge of the potential negative effects of the surgeries themselves (*i.e.*, pain, sexual side-effects, changes to body image and/or sexuality), many participants chose to undergo these procedures because doing so seemed like a better alternative than living with the ongoing anxiety of not knowing whether, when, or at which site a cancer might occur. Although some risk and uncertainty always remains after surgery, it is much less than what would be present if no surgical intervention occurred. Post-surgery, many women reported being able to put their cancer risk in perspective and not allow it to take over so much of their consciousness, time, and energy, freeing them to be more fully present as partners, mothers, daughters, etc.

Stepping back to think about the larger implications of these findings allows consideration of how the lessons learned here might translate to other contexts and phenomena. Mine is a contextual model, which describes process and contributes to our understanding of how individuals make meaning of significant health risk events in their

lives. It provides empirical evidence that elements previously known to be correlated – for example, cancer risk perception and family formation – are connected in unique ways. These data tell us *how* such elements are related. The model goes further to demonstrate the heterogeneity of experience between young *BRCA* mutation carriers, and suggests that other health risk-related phenomena may also be experienced in unique ways by different individuals in varying contexts.

Participants connected the limitations imposed and challenges presented by their mutations to their identities as women quite differently from person to person. For some, the loss of breasts and/or ovaries, changes to body image and sexuality, and diminished freedom to engage in childbearing and nursing of children – to their minds, an important component of how they would be good mothers – in a manner concordant with their prior ideals were experienced as threats to their roles as partners and mothers. Not surprisingly, this view was more often voiced by unmarried women and/or those who had not commenced childbearing when they learned their mutation status. For those who were permanently partnered and/or already had children, the mutation was perceived as less of threat to their femininity, because those roles had already been at least partially fulfilled. Because of the highly feminine nature of hereditary breast and ovarian cancer, it is likely that this situation, more than most other health threats, brought forth such feelings. However, the broader concept of threats to gender-based expectations based on health risk likely cuts across context and gender. For example, both men and women who encounter infertility might feel that this circumstance threatens their ability to fulfill gender-specific roles. Men who face serious health threats that prevent them from working and earning an income to support their families may experience a sense that they

are failing in their role as father/provider. In both cases, the processes by which individuals make decisions about and manage these health challenges might be influenced by the same components described in the model developed here: risk perception, couple dynamics related to disclosure and support, and considerations about forming or maintaining one's family.

My elucidation of how individuals differently prioritize, give value to, and manage the various stresses that come with knowledge of serious health risk is a major contribution of this research. Though the 40 participants described here faced a generic set of challenges as a consequence of being *BRCA* mutation carriers, they made unique choices based on their own circumstances, preferences, and the opinions of those close to them. Understanding this heterogeneity helps to explain the wide variation in how people deal with real lived experiences, which cannot be as effectively captured in quantitative research.

Furthermore, in keeping with the major goals of grounded theory, the model that emerges from this research should help us to understand other disorders or life situations. The product of this research represents a joining of several theories—some major “Big T” theories and some smaller “Little t” theories (Schneberger, Watson, & Pollard, 2007)—to create a new model that holds potential as a mid-range conceptual framework. If it is a useful, portable theory, then other health challenges with analogous threats, in which different elements become more prominent or fade into the background, should be explainable using this theory. Direct expansions of the utility of this model might include genetic counseling or infertility; more indirect applications might be found in considering families' decisions about living in dangerous or unhealthy environments, or

understanding the long-term impact of diethylstilbestrol (DES) on the now adult children of women who gave birth in the mid-20th century¹⁹.

The proposed model could be useful in a general genetic counseling context. Providers might usefully consider the four components of the model in working with individuals, couples, and families contemplating genetic risk. Standard genetic counseling includes a sharp focus on perceived risk and efforts to align it with actual risk; issues related to family formation are also frequent topics of discussion between genetic counselors and their patients. Most of this interaction takes the form of genetic counselors conveying information about risk or family formation options to patients, whose role is to listen, ask questions and become informed. The experience of genetic counseling for patients may be seen as more beneficial if individuals are given an opportunity to speak about their feelings and worries, and be heard. Many women in the current study reported that their genetic counselors were helpful in providing medical information and resources, but were less successful in making them feel understood or helping them figure out how to deal with genetic information. A standard practice of offering referrals to qualified medical family therapists, or other mental health professionals who can work with patients in this capacity on an ongoing basis, might greatly improve the experience of genetic counseling clients who need to talk about their risk in a depth greater than that which can be accommodated in the standard (60-minute) genetic counseling appointment.

¹⁹ DES is a synthetic estrogen prescribed to pregnant women between 1938 and 1971 to prevent miscarriage and premature birth, based on the theory that those problems were caused by a deficiency of naturally produced estrogen. Although its utility was disproven in 1953, DES continued to be prescribed until 1971, when the FDA issued a bulletin alerting doctors that exposure to DES increased female's risk of clear cell adenocarcinoma (CCA), a rare vaginal and cervical cancer, if they were exposed *in utero*. Later, it was discovered that DES daughters are also at risk for other reproductive abnormalities (including infertility) and that DES sons have elevated risk of non-cancerous epididymal cysts (Centers for Disease Control and Prevention, 2010).

Additionally, few genetic counselors have sufficient time to hear their patients' family illness narratives in full, and to either help patients understand how those beliefs intersect with the information being provided in counseling, or to work with them to improve the flow of information and understanding of illness legacies within families (J. Green, Richards, Murton, Statham, & Hallowell, 1997). Genetic counseling experiences in which couple-level processes are sufficiently attended to are even less common (Evans, 2006). Patients are typically left to their own devices to decide how to communicate genetic information to partners (and others), and to identify and meet support needs brought about by genetic risk status. Once again, making appropriate referrals to mental health professionals equipped to work with couples and families in overcoming these challenges would build upon and greatly enhance the genetic counseling experience. This can only happen when genetic counselors understand the systems-nature of their work and recognize that individuals' broader relationship contexts must be attended to therapeutically, both currently and over time.

With regard to infertility, the proposed model can contribute to our understanding of how both men and women process and make meaning of the challenges they encounter in naturally conceiving a biological child. With regard to risk perception, fertility-challenged individuals may experience guilt or blame if they or their partners believe that the cause of their difficulty getting pregnant is/was preventable, e.g., if infertility was the result of a previous sexually transmitted infection, drug or tobacco use. Women who choose to delay childbearing into their thirties and forties may perceive that they are at higher risk of experiencing infertility, and may question previous life choices. Couples may struggle with disclosure when one partner knows him- or herself to be infertile prior

to the start of a relationship, and who then must inform the other partner as the relationship becomes more serious; or, learning about fertility-related challenges in the context of an existing relationship might challenge intact couples to cope with this potentially unexpected and difficult news together. Couple support processes and patterns may be strained as partners struggle to realign themselves around a stressor that can necessitate significant renegotiations of their family formation plans, allocation of resources, and relationships with others outside of the couple unit. Finally, family formation issues are closely tied to a diagnosis of infertility; individuals and couples alike may be forced to consider alternative methods or timing of family formation, and possibly decide not to pursue having children at all.

Moving beyond contexts of genetic medical conditions, a consideration of how the proposed model might apply to families living in or near hazardous environments elicits several interesting possibilities. For example, a low-income parent deciding whether or not to move her/his family to a part of a city near a power plant or sewage treatment facility, or even moving to an area where there is limited access to healthy food or clean air and water, often have valid reasons for doing so. Housing is often less expensive in areas where known or potential health threats are present, and jobs for which a low-income individual is qualified may be more readily available in these areas. An individual and/or parent must consider whether the advantages to her/his budget and ability to provide for their family warrants exposure to possible health risks. While dynamics around couple disclosure may be less salient in this context, access to adequate information from local authorities becomes more salient, and is reflective of how power works to obscure information around health risks – and how that shapes family dynamics.

Also, there are family health implications of a history of slow disclosure that emerges over time, and that different generations must cope with as it evolves. With regard to support, defining and providing couple support are likely to be relevant for those in relationships as they cope with the stressors inherent in living with environmental risk, or working to improve the family financial situation so that living in such an environment is no longer necessary. Family formation issues may arise when environmental health threats jeopardize one's ability to have a healthy pregnancy, or when living in an extremely toxic environment results in severe and/or unexpected birth defects or complications. Individuals making these choices, and professionals addressing issues of low-income families in their research and policy work, might find the proposed model useful in conceptualizing families' decision-making processes related to geographic location and housing.

Lastly, families in which pregnant women were treated with DES to prevent miscarriage and premature birth (a treatment strategy employed in the US between 1938 and 1971, but now long since abandoned) may be better understood when examined using the proposed model presented here. Women given DES while pregnant are at moderately increased risk of breast cancer. "DES sons," or boys and men who were exposed to DES in the womb, are at risk of non-cancerous growths on the testicles, and possibly other genital abnormalities. "DES daughters" face more frequent and more serious risks, including reproductive tract abnormalities, clear cell adenocarcinoma (CCA, a rare vaginal and cervical cancer), ectopic pregnancy, premature delivery, and infertility. Although efforts were made to locate and inform women who had been prescribed DES after its health risks became known, many individuals are still unaware

that they are in this category (Centers for Disease Control and Prevention, 2010). Consequently, people are still learning that health problems they are encountering in adulthood are the result of drugs given to their mothers during pregnancy, and now aging mothers are learning or continuing to cope with the knowledge that drugs given to them during their pregnancy jeopardized the health of their children, creating complicated disclosure dynamics on multiple levels. These facts become part of ever-evolving narratives as intergenerational histories unfold over time within families. Individuals, couples, and families affected by DES move through changing phases of DES-related risk over successive generations, and information is still emerging regarding how the third generation (grandchildren of women prescribed DES) might be affected. Individuals must contemplate and decide how to make sense of these potential health risks, and reconcile their feelings about being exposed to this risk by a medical community that might be perceived as having failed to protect them. When cancer or fertility-related challenges are the result of DES exposure, individuals must go through disclosure processes similar to those described previously in this chapter, with the same kinds of variations depending on context. Couples' support processes may require realignment to accommodate the challenges presented by this health phenomenon, and partners are likely to work together to become educated about how DES exposure might shape family formation.

Overall, the model proposed here presents a new way of understanding how issues of family and health may be integrated, and illuminates contexts previously unexamined. While four additional contexts have been suggested here, it is likely that many other relevant situations exist. Additional research utilizing the proposed model

will help to refine it over time, and future quantitative investigations can clarify the directionality and magnitude of relationships between model elements.

Practical Implications

Findings from the current study have several implications at the policy level, for providers who work with *BRCA*-positive women, and for mutation carriers themselves. For physicians and genetic counselors alike, a general orientation toward improving cancer survivability requires considering how these data might inform their ability to provide effective intervention. If mutation-positive women are to survive longer, they must be active *early* in the life course in working to mitigate their risk, and genetic testing at a young age is the essential first step. Providers ordering genetic testing for young women would be better able to educate and serve that population if they more fully understood both the complicated psychosocial implications of the *BRCA*-positive experience and the importance of initiating appropriate interventions while carriers are in their twenties. Understanding the unique challenges to self-perception and relationships for young mutation carriers would greatly facilitate providers' ongoing interactions with women in this population. These well-informed providers would be better able to attend to carriers' continuing need for new and updated information, and to provide informed guidance and resources related to risk, sexuality, pregnancy and breastfeeding, and/or surgery as carriers meet these different challenges along the mutation-positive path.

Further, a heightened awareness of how easily carriers can misinterpret information, receive inaccurate data, or simply not have access to appropriate resources to become educated about their risks and options, should incentivize providers to implement a medical strategy in which communication among providers and specialties is the norm,

and patients have access to the full range of doctors (*e.g.*, gynecological oncologists, reproductive endocrinologists, ob/gyns), nurses, genetic counselors, and mental health professionals, *working as a team*, to make decisions with and in the best interest of patients and their loved ones.

Genetic counseling practice may also be informed by understanding the importance of a comprehensive understanding of family history for accurate risk assessment and risk perception. This was demonstrated by those participants who felt uninformed about their risk because of a lack of information about their family health history. While genetic counselors already focus on obtaining an accurate family history for quantitative risk assessment, an increased awareness of how the lived, experienced family narrative shapes and influences the beliefs, attitudes and decision-making of young mutation carriers could broaden and enrich their approach to high-risk women. Listening fully to the stories young women tell about how they experienced cancer and risk in their families provides an invaluable opportunity to reinforce that which is factually correct, and clarify inaccurate information, as women struggle to formulate a meaningful, functional understanding of their own personal risk and management options.

When referral to other mental health professionals – especially medical family therapists – is appropriate, therapists might be optimally effective if they strive to have an intergenerational conversation in which the full range of individuals touched by a family's cancer narrative can discuss how they understand and make meaning of this information. A willingness on the part of families (perhaps assisted by providers) to have healthy, non-threatening conversations with *minor* children about cancer in the family,

and what it means for them as a group and as individuals, could help to make *BRCA* a less intimidating threat, engender a sense of competence in handling mutation test results, and create a next generation of potential carriers who are well-informed and prepared to face challenges at this complicated stage of the life course.

For any provider working with this population, the insights provided in this study could be helpful in increasing competence and confidence that they can empathize with young mutation carriers' struggles. A therapeutic interaction between a *BRCA* mutation carrier and a genetic counselor, medical family therapist, physician, or nurse might occur when the provider can say to the patient, "I truly understand what you're going through, and what difficult choices you're facing. There is no easy answer. And, I know of some examples of how other women in your situation have coped. Let me share that knowledge with you." A genuine sense that one is understood in the medical environment might be a powerful experience for many young carriers.

With regard to policy, health insurance issues are of vital importance for women contemplating testing and coping with the implications of a positive result. Though the passage of the Genetic Information Nondiscrimination Act (GINA) in 2008 was a major step forward in protecting individuals with known susceptibility to health risks from discrimination with respect to health insurance and employment, significant challenges remain relative to implementing similar protection for life insurance, and in helping the public become aware of their rights and feel confident that they are truly protected. Further, if health insurers were required to cover not only costs related to genetic testing and medically-indicated risk-management strategies, but also the costs of mental health

care (including couple and family therapy) mutation carriers' needs could be more effectively met.

Insurance coverage for assisted reproductive technology for carriers who wish to utilize these services would also greatly benefit this population. This is an intimately personal choice, and it is unfortunate that it is often decided instead by an insurance company's willingness to pay for the procedure. A striking number of participants in the current study, as well as many other women I have encountered in my work with this community more broadly, have stated that they would be interested in pursuing prenatal genetic diagnosis (PGD) to ensure that they would have mutation-negative children, but that the high cost of the procedure would likely prohibit them from doing so. In the long run, insurance companies would likely spend less money paying for PGD than they would paying for genetic testing, several years of screening, risk-reducing surgeries, and/or cancer treatments when babies born with unknown mutation status reach young adulthood. Ideally, women and their partners would be able to make the choice that works best for them, not forced to do what best meets the needs of their insurance provider.

Finally, my results have implications for the mutation carriers themselves. The predominantly positive narratives provided by these participants undercut preconceptions that living with knowledge of oneself as *BRCA*-positive during young adulthood is “undoable.” Making data such as these available to women contemplating genetic testing, and to women who are newly-confirmed as carriers, might provide a roadmap for how to tackle *BRCA* at this stage of the life course. Many participants noted a perceived lack of access to accurate, high-quality, personally relevant information and also the opportunity

for social comparison at various points along their journey as mutation carriers, especially in the days and weeks immediately following their receipt of results. Because it is difficult to connect each newly-tested carrier with another young woman in a similar relationship and family context who lives near her, who could offer face-to-face support and interaction, providing salient information derived from the current study might be the next best thing. Knowing that others have experienced similar fears, worries, and challenges, and understanding how they have overcome those, would be empowering for some newly-aware carriers, and they should have access to this type of resource. The feeling that one is alone in the mutation-positive experience is troublesome, and not uncommon – but it does not have to be so.

Limitations and Future Directions

The primary limitation of this study, as with all qualitative research, is that I was the sole researcher, conducting all 40 interviews and interpreting the data independently. My personal biases inherently shaped how the interviews were administered (*e.g.*, empathy for participants, unplanned follow-up questions, knowledge of context) and how “emergent” themes were identified and organized. Therefore, it is likely that other researchers might generate different responses from the participants, and might interpret the data differently, as well. However, because careful measures were taken to ensure the trustworthiness and quality of the data (*e.g.*, theoretical sampling, peer review and debriefing, member checking), as described in Chapter 3, the product of this research is no less valid.

A second important limitation is the use of women’s largely retrospective narratives of the periods during which they were actively contemplating issues related to

their mutation-positive statuses. The extent to which this may limit or bias the findings varies from participant to participant, as some women were newly tested and in the active phase of working through such issues. It is possible that women's memories may have become distorted over time, or that important phenomena were not recalled. Perhaps some women were focused on either the positive or negative aspects of their experience, and unconsciously misrepresented the memories related to the other category. However, because women's narratives about their experiences as mutation-positive are what operationally shape their current adaptation and the meaning they assign to their status, and because women's stories and the way they subjectively recall their experience inform both their behavior and identity, these perspectives are still extremely valuable, despite the potential for factual distortion. That is to say, what they think they recall may be functionally more important than what actually occurred. Producing a record of women's unique narratives and perspectives is consistent with the principles of both feminist thought and grounded theory methodology.

Another generic limitation of qualitative research is that findings cannot be generalized to a broader population from which respondents were drawn, in this case, to all young female *BRCA* mutation carriers. Recruitment methods were unique to this study, and they yielded a particular group of 40 women who produced these particular results. Furthermore, the issue of volunteerism creates a limitation in that women self-selected into this study; participants recruited through FORCE and snowball sampling initiated contact with me to indicate their desire to participate (as opposed to participants recruited from the BI study, to whom I reached out directly based upon my prior research which targeted these women (Hoskins et al., 2008; Peters et al., 2006)). The FORCE

recruits tended to be younger and more likely to have recently learned their mutation status. A subset of the BI participants are part of a prospective NCI study which has been active for more than 30 years. These families had a long history of participation in multiple different NCI-sponsored research projects over time, and awareness of the family's mutation status dates back more than 10 years for many of them. These families commonly had many affected family members and had made extensive use of risk-reducing surgery before these management strategies gained widespread acceptance. Thus, the population described here is heterogeneous in terms of age and longevity of knowledge of mutation status, but relatively homogenous with regard to race, socioeconomic status, level of education, etc. Therefore, these findings provide little insight regarding how individuals who differ on these characteristics might experience themselves and their lives as mutation carriers. Future qualitative studies should focus on these other subgroups within the *BRCA*-positive population in order to increase our understanding of what kind of variation might exist.

A possible major deficit of this study is the absence of a wider range of negative experiences. This could, at least theoretically, imply that negative experiences were rare or uncommon. More likely, this may be another manifestation of volunteerism in which women with positive experiences were more likely to put themselves forward as study participants. Although some participants did described having struggled in a specific area after learning about their mutations, such narratives were rare. Instead, most participants were either still in the depths of the early months of their experiences and did not yet have either perceived successes or failures to report, or they had successfully come to some sort of peace with their situation over time. Anecdotally, one *potential* participant,

who happened to be the sister of another participant, indicated that she was dealing with some very difficult issues. In conducting her screening interview, I felt confident that her narrative would enrich the heterogeneity of experience among the group. Between her screening interview and her in-depth interview, she wrote to explain that she had decided not to participate in the study because she believed that doing so, and re-visiting her struggles in depth, would be too painful. Despite my emphasizing to her that her experience was very valuable and might be helpful to others coping with similar challenges, and despite my reassurance that we did not have to discuss things she wished to leave out, she remained firm in her decision not to participate. It is certainly possible that other young women who read the study announcement and were eligible to participate, chose not to do so for similar reasons. In order to reach women whose experiences with *BRCA* have been negative, recruitment efforts for future studies could seek referrals from genetic counselors and medical family therapists who may encounter such individuals in their work. A carefully worded study invitation targeted to members of this group and delivered by a trusted healthcare provider may be effective in encouraging them to participate in a future interview study more focused on issues pertinent to them.

Future qualitative studies could triangulate sources by including female mutation carriers' partners, family members, and healthcare providers to better understand women's experiences and relationships. Data triangulation could be folded into the current data to include findings from focus groups and Colored Eco-Genetic Relationship Maps (CEGRMS) (Peters et al., 2006) for participants who have been included in those other types of data collection in this and other studies to further enrich our understanding

of their experience. A study focusing on intra-family interactions, in which perhaps three women from each of ten families were interviewed both separately and as a group about their experiences, could provide fascinating insight into family communication, interpretations of family narratives, and variations in information processing and decision-making in the context of a shared family history. The voices of young *male* mutation carriers (and untested males in HBOC families) are still silent in existing literature, and a study similar to this one investigating how they experience their mutation status (either known or potential) would likely be both fascinating and informative. Men from mutation-positive families are statistically just as likely as women to be mutation carriers (due to *BRCA1/2*'s autosomal dominant mode of inheritance), they are at increased risks of certain specific malignancies (*e.g.*, cancers of the male breast, pancreas, and prostate), and they can transmit the mutated *BRCA* allele to their children, both male and female. Finally, once qualitative data have been used to form a comprehensive understanding of the full range of experiences for members of this population, future quantitative studies might formally investigate the strength and direction of relationships between variables and components of the proposed model in order to more confidently recommend interventions and/or changes to policy.

Conclusion

The current study explored how young female carriers of *BRCA1/2* mutations coped with the normative challenges of young adulthood and navigated the constraints imposed by their cancer risk. Specifically, these data illustrated how breast and ovarian cancer risk and risk perception shaped and were shaped by young women's interactions in partner relationships, their decision-making about family formation, and their handling

of risk-reduction strategies and decisions. My findings comprise a unique contribution to the literature on the diverse ways that women experience this challenging health situation in various developmental and relationship contexts.

Participants' experiences suggest that some mutation-related support services were not readily available, hard to find, and occasionally less than authoritative relative to their continued needs over time. There is a shortage of mental health professionals who are conversant with (let alone authoritative about) the facts and management issues related to *BRCA1/2* and hereditary breast/ovarian cancer. This reality compromises providers' ability to help mutation carriers deal with their syndrome-related consequences for interpersonal relationships. These results identify the need for remedies aimed at correcting information and experience-related deficiencies among providers, to enable their providing more comprehensive and ongoing services to individuals, couples and families affected by this and other hereditary cancer syndromes.

More research, both quantitative and qualitative, is needed to explore how people of all backgrounds experience and describe their perceived genetic risk of cancer. The women in the current study have demonstrated that, when given the opportunity to fully participate in a research project that solicits and values individual narratives and perspectives, the experience can be empowering and enlightening for all involved. For many participants, the interview afforded them the opportunity to think about their story in a new way or make connections that they had not previously discovered. At the end of her interview, Marie told me that "your questions really hit on everything that has been impacted for me, but it was cool to talk about all of it together – I haven't done that before." Others simply appreciated the opportunity to talk through their experience and

know that they were not alone, like Reina, who stated, “it’s very cathartic...and it’s a relief to know that other women experience similar issues.” Many women reported that participating in this study was a positive and generative experience, one they were happy to share in the hope that doing so might help others similarly affected. Rachel stated that she was happy to do “anything that I can do to maybe help not have somebody go through the same way I had to go through it.” The opportunity to share their stories, make sense of them, feel like part of a community, and have their experiences validated as meaningful and important was a powerful experience for these courageous women. I am forever indebted to each of my participants for sharing their stories with me. I hope that I have conveyed the gravity of their common challenge as well as the diversity of their solutions in a way that will provide comfort and insight to mutation carriers not yet identified and to the providers who are committed to delivering the best possible care to women at increased genetic risk of cancer. More importantly, I hope that I have captured their uniqueness, their beauty, and their strength.

APPENDICES

Appendix A: Historical & Biological Background of *BRCA1/2* Mutations

The Human Genome Project, undertaken in 1990 by the National Center for Human Genome Research (now National Human Genome Research Institute) and the US Department of Energy ("Human Genome Project Information," 2005), revolutionized our understanding of human genetics. Mutations in the genes *BRCA1* and *BRCA2* were implicated in hereditary breast/ovarian cancer in 1994 and 1995, respectively (Miki et al., 1994; Wooster et al., 1995). *BRCA1* is located on the long arm of chromosome 17, while *BRCA2* is located on the long arm of chromosome 13. More than 1,640 and 1,890 distinct mutations have been reported in *BRCA1* and *BRCA2*, of which 845 and 765, respectively, are classified as disease-causing (Lindor, McMaster, Lindor, & Greene, 2008; NHGRI, 2007). Although the two syndromes share important fundamental similarities (e.g., the striking predisposition to both breast and ovarian cancer), there are unique characteristics of each that make the two orders dissimilar clinically. For example, the risk of developing ovarian cancer is significantly greater in *BRCA1* than in *BRCA2*. The increased risks of male breast, prostate and pancreatic cancer are strongest in *BRCA2* carriers. *BRCA1*-related breast cancer is predominantly estrogen-receptor negative, and has unique histologic features, while *BRCA2*-related breast cancer more closely resembles sporadic breast cancer (Lindor et al., 2008). Both genes are related to DNA repair, transcription regulation, and cell cycle control (Boulton, 2006; Boyd, 2003; Boyd et al., 2000; Brody & Biesecker, 1998; Ding et al., 2004; Prat et al., 2005).

BRCA1/2 mutations predispose carriers to a 50-87% lifetime risk of breast cancer (Antoniou et al., 2003; Bredart, Autier, Audisio, & Geraghty, 1998; Brody & Biesecker,

1998; Brose et al., 2002; Cappelli et al., 2001; Easton, Ford, & Bishop, 1995; Narod et al., 1998; Prat et al., 2005; Struewing et al., 1997; Suthers, 2007). Ovarian cancer risk among mutation carriers varies depending on which mutation is present; *BRCA1* mutations are associated with 20-60% lifetime risk of ovarian cancer, while the ovarian cancer risk conveyed by *BRCA2* mutations ranges from 10-40% (Antoniou et al., 2003; Claus et al., 1996; M. C. King et al., 2003; Pavelka et al., 2007; Risch et al., 2001; Struewing et al., 1997; Whittemore, Gong, & Itnyre, 1997). More recent research has resulted in narrower risk estimates of 65%-82% for breast cancer and 39%-54% for ovarian cancer in *BRCA1* carriers (Antoniou et al., 2003; M. C. King et al., 2003), and 45% for breast cancer and 11-30% for ovarian cancer in *BRCA2* carriers (Antoniou et al., 2003; Boyd, 2003). There is some evidence of an ovarian cancer cluster region on exon 11 of the *BRCA2* gene; mutations occurring in this region are likely associated with higher risk of ovarian cancer than other *BRCA2* mutations (Boyd, 2003; Gayther et al., 1997; Gayther et al., 1995). These levels of risk are considerably higher than those present in the general population, about 12.8% for breast cancer and 1.5% for ovarian cancer (DevCan, 2005; Fay, 2004; Fay et al., 2003). Hereditary cancers also tend to occur at a younger age than their sporadic counterparts. For example, the mean age of onset is 54 years for ovarian cancer in *BRCA1* carriers (compared with a mean age of 63 for sporadic ovarian cancer) (Boyd et al., 2000; Lakhani et al., 2004), and *BRCA1/2*-linked breast cancer typically occurs before the age of 50 (Narod et al., 2002). Carriers' risk level begins to increase at approximately age 25, necessitating medical screening starting early in the life course (Meijers-Heijboer et al., 2001). Overall, diagnosis may occur in the 40s for ovarian cancer (*BRCA1*, later for *BRCA2*) and in the 30s and 40s for breast

cancer (Boyd et al., 2000; Moslehi et al., 2000; Pavelka et al., 2007). There do not appear to be significant differences in histology of tissue removed for risk-reduction from mutation carriers versus non-carriers (Casey et al., 2000). In women who do develop cancers, survival rates are similar to what is observed in age-matched patients diagnosed with sporadic (rather than hereditary) cancers, a ten-year survival rate of approximately 50% (Meijers-Heijboer et al., 2001).

As previously stated, mutations in *BRCA1* and *BRCA2* make up a small but notable proportion of breast and ovarian cancer cases. Shortly after the mutations were identified, they were thought to explain about seven in 100 cases of breast cancer in women with a family history (Ford et al., 1995) and approximately 10-15% of all ovarian cancer cases (Pal et al., 2005; Risch et al., 2001), with more of these resulting from *BRCA1* (about 5.7%) than *BRCA2* (about 3.8%) (Boyd, 2003; Claus et al., 1996; Pavelka et al., 2007; Prat et al., 2005). A more recent study of 1220 women up to age 55 who were diagnosed with breast cancer (unselected for family history) reported that 3.8% had a mutation in either *BRCA1* or *BRCA2* (Southey et al., 1999). However, the proportion of cancer cases in *young* women attributable to mutations in *BRCA1/2* paints a very different picture. A UK study found *BRCA1/2* mutations in 5.9% of breast cancer patients 35 and younger, and in 4.1% in those aged 36 to 45 (Peto et al., 1999). Finally, *BRCA1/2* mutations are thought to explain “approximately 33% of [breast cancer] cases age 20-29 years,” a finding highly relevant to the proposed study (Claus et al., 1996, p. 2318).

The prevalence of the mutations in the general population is quite small; for example, the gene frequency of *BRCA1* as been estimated to be between 0.0006 (Ford et al., 1995) and 0.002 (Struewing et al., 1995); population prevalence for these genes are

difficult to determine because widespread screening is impractical from both logistical and financial perspectives (Brody & Biesecker, 1998). These numbers translate to an estimated overall *BRCA* carrier frequency of about 1 in 800, but it is significantly higher—about 1 in 40—in the Ashkenazi Jewish population due to the prevalence of three unique founder mutations, two in *BRCA1* and one in *BRCA2* (Boyd, 2003).

Both *BRCA1* and *BRCA2* are highly but incompletely penetrant: most, but not all, individuals who inherit a deleterious mutation will develop cancer if no risk-reducing actions are taken. Whether or not cancer develops in the presence of a *BRCA1/2* mutation is presumed to depend on the specific mutation that is present (Surbone, 2001), and on both gene-gene and gene-environment interactions over the lifecourse (Boyd, 2003; Eisen et al., 2000; Pavelka et al., 2007). Environmental influences include use of oral contraceptives and surgical sterilization (Narod et al., 1998; Narod et al., 2001; Whittemore et al., 2004), tubal ligation (Narod et al., 2001), and parity (Narod et al., 1995).

Breast and ovarian cancer are quite different with regard to ease of detection and efficacy of screening. Breast screening is widely practiced, with women in the US currently advised by their healthcare providers and through a wide variety of public service announcements to perform monthly breast self-exams (ACS, 2007). In addition, a clinical breast exam is a standard component of most annual Ob/Gyn and primary care visits (Day, 2008). Therefore, the majority of women who participate in regular medical care receive at least this level of breast cancer screening. However, ovarian cancer screening is much more difficult and less common, and is not a standard component of annual medical examinations aside from a standard pelvic exam (which has little if any

value related to early detection of ovarian cancer). Currently, the “best” available screening methods, serum CA-125 test and transvaginal ultrasound sonography (Pavelka et al., 2007) are generally regarded as ineffective, but are nonetheless offered to women known to be at high risk of developing ovarian cancer (Prat et al., 2005). The recognizable symptoms of ovarian cancer include pelvic/abdominal pain, urinary urgency/frequency, bloating, and early satiety occurring more than twelve days per month, all occurring for less than one year (Andersen et al., 2008); interestingly, gynecological symptoms are reported least frequently (Goff, Mandel, Melancon, & Muntz, 2004). Researchers have recommended that medical professionals make *BRCA*-positive women aware of these symptoms (Pavelka et al., 2007), but they may be easily overlooked since they may be confused with pre-menstrual symptoms or other minor and transient health concerns.

In addition to breast and ovarian cancer, there has been some research indicating that mutations in *BRCA1/2* may predispose carriers to malignancies including male breast cancer, colorectal cancer (Ford et al., 1994; Thompson & Easton, 2002), prostate cancer (BCLC, 1999; Ford et al., 1994; Sigurdsson et al., 1997; Struewing et al., 1997), pancreatic cancer, gallbladder and bile duct cancer, stomach cancer, and malignant melanoma (BCLC, 1999). However, the level of risk of these cancers is significantly lower than the risks of breast and ovarian cancer associated with mutations in *BRCA1/2*, and for most (with the exception of prostate cancer for male carriers), normal population screening guidelines are recommended (Burke et al., 1997).

Both *BRCA1* and *BRCA2* demonstrate an autosomal dominant inheritance pattern. Autosomal dominant mutations are passed with equal frequency from and to males and

females; that is, males and females are equally likely to inherit a mutation in *BRCA1/2*, and mutations may be inherited from either mothers or fathers. This fundamental characteristic of autosomal dominant Mendelian genetics is widely misunderstood in the context of hereditary breast/ovarian cancer, which many erroneously consider to be a “female disease,” thereby overlooking the important role of men in these families, both as carriers/transmitters of the mutations and as persons at risk of specific syndromic malignancies. In addition, only one mutation is needed in order for risk status to be achieved; therefore, individuals need only inherit one mutation, from one parent, in order to be considered “*BRCA*-positive.” Finally, an autosomal dominant inheritance pattern means that each offspring of an affected individual will have a fifty percent chance of inheriting a parent’s mutation. In other words, a parent with a mutation would statistically be expected to pass that mutation on to half of her/his children.

In addition to these two identified mutations, many scientists believe that there are other forms of “inherited breast cancer” that are not yet fully understood. Efforts are underway to identify additional genetic mutations related to increased susceptibility to breast/ovarian cancer (e.g., *BRCA3*), but results thus far have not been definitive (P. Smith et al., 2006; Thompson & Easton, 2002). If additional genes are identified, the proportion of breast and ovarian cancers explained by genetic predisposition will continue to increase (Brody & Biesecker, 1998; Burke, Press, & Pinsky, 1999). At present and given the current state of knowledge about this hereditary cancer syndrome, it is difficult to ascertain what proportion of breast and ovarian cancer cases are due to genetic predisposition, or the general population carrier frequency of *BRCA1/2* (Brody & Biesecker, 1998). However, as more and more individuals are tested and their long-term

health statuses and decisions are followed, our knowledge on this subject will continue to increase.

Appendix B: Medical & Psychosocial Issues Related to *BRCA1/2* Testing

Identification of HBOC families and mutation-positive individuals. In order to fully understand the experiences of young *BRCA1/2* mutation carriers, we must first understand the process by which they learn of their increased risk status. Researchers have identified several factors which suggest the presence of hereditary breast/ovarian cancer (HBOC) within families, including young age of cancer onset (i.e., pre-menopausal or <60), the presence of bilateral breast cancer, and individuals affected in multiple generations (Brody & Biesecker, 1998; Decruyenaere et al., 2000; Prat et al., 2005). The official recommendation statement from the American Society of Clinical Oncology (ASCO) reads:

ASCO recommends that genetic testing be offered when (1) the individual has personal or family history features suggestive of a genetic cancer susceptibility condition, (2) the test can be adequately interpreted, and (3) the results will aid in diagnosis or influence the medical or surgical management of the patient or family members at hereditary risk of cancer. ASCO recommends that genetic testing only be done in the setting of pre- and post-test counseling, which should include discussion of possible risks and benefits of cancer early detection and prevention modalities, (ASCO, 2003).

Although these guidelines are helpful, they are open to interpretation (e.g., there is variability in what different healthcare professionals consider “suggestive of cancer susceptibility” with regard to family cancer history), and therefore some ambiguity may exist in decision-making about testing.

Submission of a biological sample for genetic testing requires a blood or skin sample, from which individual cells are then extracted. If a *BRCA1/2* mutation is suspected within a family, an affected member (one who has been diagnosed with breast or ovarian cancer) is usually tested first. This test requires “full sequencing,” or a complete review of the more than 3,500 distinct mutations and sequence variations that

have been identified in *BRCA1* and *BRCA2* (Van Riper & McKinnon, 2004). Of these, approximately 845 and 765, respectively, are known to be deleterious, or definitely associated with increased risk of breast/ovarian cancer; the others are missense mutations or variants of unknown significance, for which the level of cancer risk is not fully understood (NHGRI, 2007). Full sequencing is no small undertaking. The sheer number of possible mutations, together with their scattered distribution throughout the coding regions of the genes (Boyd, 2003; Prat et al., 2005) makes this a technically difficult and time-consuming process; hence, it is quite expensive, with the current retail cost of sequencing ranging from about \$375 for single site testing to \$3,000 for full sequencing of both genes (NCI, 2002; Van Riper & McKinnon, 2004). This test may or may not be covered by health insurance, and some insurers require that an individual be already affected by cancer to qualify for reimbursement. Therefore, access to pre-symptomatic testing may be limited to those with excellent coverage or who are able to pay for the test privately. Adding to this complicated dynamic, one company, Myriad Genetics, holds the US patents (as well as patents in at least 16 other countries) on the genetic test for *BRCA1/2*, (Balter, 2001). Therefore, all tests of US patients must be submitted to Myriad, which can select the price of the test relatively independently.

Proband testing. There are three possible outcomes of genetic testing of a proband (the first person in a family to be tested) in an HBOC family. First, testing may identify the presence of a known pathogenic mutation; this occurs in less than half of the tests performed in white women, and with a significantly lower frequency in tests of women from racial/ethnic minorities, most notably African-Americans (Boyd, 2003). In cases with this testing outcome, genetic counselors and physicians who work with HBOC

families would be able to understand and explain a patient's (and family members') cancer risk, because risk information has been determined from other families with mutations in the same gene (Palma et al., 2006). However, even with the identification of a known mutation, it is impossible to know whether or not a person will develop cancer, at which site(s) a cancer or cancers may develop, or when (NCI, 2002).

A second possible outcome of an initial genetic test within a family is the identification of a variant of unknown significance, a mutation in *BRCA1/2* that has not been previously identified or for which the associated cancer risks are not yet known (Palma et al., 2006). This result is sometimes referred to as an ambiguous result and is more likely to occur when testing women from racial/ethnic minorities. In this situation, it is much more difficult to know whether the identified mutation is associated with increased cancer risk; all humans have multiple genetic mutations that occur naturally, and most of them are not deleterious (NCI, 2002). An ambiguous result *may be* suggestive of increased risk, but precise risk information is difficult to determine because the mutation has not previously been encountered.

Finally, proband testing may yield an uninformative result when no *BRCA1/2* mutation is identified; ambiguity exists because there is a possibility that a mutation exists in another cancer susceptibility gene (not *BRCA1/2*), or that there is an undetected mutation in *BRCA1/2*. Individuals and families are left in a difficult situation, as risk management decisions must be made without specific guidelines (O'Neill et al., 2006).

Subsequent testing of other family members. If an initial genetic test within a family results in the identification of a known mutation, testing of other family members is fairly straightforward. Because the location of the “family mutation” on the

chromosome is known, subsequent tests within the same bloodline can focus on that exact location to determine whether or not the mutation is present, rather than examining all of the potential *BRCA1/2* mutation sites; hence, the level of potential ambiguity is reduced. This is referred to as *site-specific testing*. Individuals who undergo testing in this context will be able to more easily interpret their test results as either positive or negative; when a test comes back negative in this case, it is considered a “true negative” or “real negative,” (Palma et al., 2006). Because the prevalence of *BRCA1* and *BRCA2* mutations in the general population is so low, it is extremely unlikely that an individual would test negative for a family mutation and actually carry another, different *BRCA* mutation (NCI, 2002). Individuals who are true negatives can assume that their risk of breast/ovarian cancer is the same as the risk of the general population (NCI, 2002; Palma et al., 2006). In certain ethnic populations in which the prevalence of *BRCA* mutations is significantly higher (e.g., individuals of Ashkenazi Jewish heritage), and in which only a small number of specific mutations have been linked to cancer risk (in the Ashkenazim, for example, there are 3 such “founder” mutations), the detection of mutations in both parental lineages is more likely to occur.

If an initial genetic test within a family produces an uninformative or ambiguous result, subsequent family members may also be tested for the variant of unknown significance. Full screening may also be used in order to ascertain whether a known mutation is also present, especially if there are other affected family members. Multiple detections of the same variant of unknown significance in members of the same family and across multiple families will contribute to knowledge within the field regarding the

implications of the variant, as time passes and the health outcomes of those individuals can be followed by researchers.

Finally, if the initial genetic test of an affected member of a family results in a negative result (i.e., no mutation or variant is detected), it may be considered impractical to test other family members unless they are also affected. However, members of HBOC families who do not have a mutation in *BRCA1/2* can still develop breast/ovarian cancer since they are at standard population risk; therefore, one affected family member testing negative in a family with multiple affected individuals does not mean that no mutation is present, and further testing of other affected family members may be advisable.

Limitations of testing. Several characteristics of genetic testing for *BRCA1/2* limit its utility and appeal to some individuals considering testing. Perhaps most importantly, because of the uncertainties inherent in cancer risk figures and the limited spectrum of proven cancer prevention strategies, even an informative genetic test (i.e., a true positive or true negative) leaves many lingering questions (Bredart et al., 1998), as it indicates only the *probability*, rather than certainty, that an individual will develop cancer (Van Riper & McKinnon, 2004). Recent data have raised the possibility that there may still be some residual familial risk of breast cancer even among mutation-negative women from mutation-positive families (Katki, Gail, & Greene, 2007). There are no guarantees about whether, when, or in what form cancer may develop. There are no risk-reduction or treatment strategies that are 100% effective; even surgical prophylaxis does not completely mitigate one's risk of cancer. Therefore, regardless of the outcome of one's genetic test, some level of uncertainty remains (Van Riper & McKinnon, 2004).

Another significant limitation is the fact that genetic tests may miss mutations that truly exist, especially when those mutations are characterized by deletions or rearrangements of large segments of DNA (Srivastava, McKinnon, & Wood, 2001). In both situations, individuals may receive negative or uninformative test results, resulting in inappropriate, insufficient, and/or uninformed medical advice. Negative genetic test results are fully informative only when a known mutation has previously been identified within a family (Van Riper & McKinnon, 2004).

Because genetic conditions are inherently a family issue, the decision of one individual to undergo testing has implications for other family members (Bottorff et al., 2000). It is difficult to ascertain the extent to which pressure from concerned family members influences testing decisions by others in the family (Murphy, 1999). For example, adult children may pressure one of their parents or siblings to get tested if they are known to have had a syndrome-related cancer, or out of a desire to share their own testing experience with other family members out of a sense of togetherness. However, family members must also be aware that it is very possible for positive tests to yield unwanted information including, for example, identifying an individual who has chosen not to be tested as an obligate carrier (e.g., a woman with a maternal family history of breast/ovarian cancer and an untested mother, who chooses to undergo testing herself for a known family mutation and learns she that is positive. Mendel's Laws require that the untested mother also carry the mutation. Thus, the mother who had chosen not to be tested, and who may not want to know her mutation status, will nonetheless learn that she is a carrier) (Bottorff et al., 2000). These complicated family issues are often not fully

considered in the period of time leading up to a genetic test, but they are important factors in the process.

A final and important limitation of genetic testing for *BRCA1/2* is that, because (1) mutated genes are rare in the general population, (2) the genes are very large, and (3) the number of reported mutations is large (i.e., more than 845 in *BRCA1* alone), their identification in a random individual from the general population is impractical. Consequently, a cancer-affected person from a family with multiple cases of breast and/or ovarian cancer, is generally the preferred starting point for testing. If a specific disease-causing mutation is identified, that that specific mutation is sought in DNA samples of other family members. But even with this strategy, the majority of multiple-case families do not have a detectable mutation (Murphy, 1999, p. 236). Nonetheless, members of such families remain at increased cancer risk simply on the basis of their family history, and must be managed with that knowledge in mind.

Genetic testing is most likely to yield an informative result when an affected person is the first tested within a family, because that test can detect, in part, whether a cancer is associated with a hereditary syndrome. If an unaffected person is the first tested and the result is negative, there is no way of knowing whether the negative outcome is due to the absence of a mutation in the family or simply that the individual is a non-carrier of a still-undetected family mutation. Therefore, in order for a family to begin the process of genetic testing for multiple individuals, an affected member must usually be willing to be the first to be tested. Although many cancer-affected individuals are often highly motivated to undergo genetic testing (Meiser, 2005), those who are not can inhibit or delay testing for other family members.

Risks of testing. The literature elucidates several risks that come along with choosing to undergo genetic testing for *BRCA1/2*. Historically, concerns related to potential discrimination in employment and health/life insurance have been cited as reasons to be wary of genetic testing (Bredart et al., 1998), despite the fact that documented instances of such behavior are extraordinarily rare. At the institutional level, steps are being taken to mitigate these risks, including the recent passage of the Genetics Information Non-Discrimination Act (GINA) on May 21, 2008 (HR 493). GINA prevents employers and insurers from making decisions about individuals' job or insurance status based on genetic information; it will take effect in May 2009 for insurance issues and November 2009 for employer issues. Noticeably missing from GINA are protections related to life, disability, and long-term care insurance, any or all of which could be salient concerns for those living with increased genetic risk of cancer (NHGRI, 2008). Although GINA is an important step forward in protecting individuals and families from genetic discrimination, it may not fully alleviate the fear and apprehension that some potential testers experience with regard to their genetic information. Additionally, GINA does not (and no government act or policy could) protect individuals from discrimination that occurs on the interpersonal level, such as, for example, being discriminated against as a potential romantic partner because of another person's fear of the implications of a mutation. Therefore, individuals considering testing must carefully consider how possessing information about their genetic makeup and lifetime health risks may impact their experiences and interactions in the workplace, in the healthcare system, and in interpersonal relationships.

Another important risk of testing is that women who receive a negative or uninformative result could be lulled into a false sense of security regarding their risk of cancer, believing that because they do not have a deleterious mutation in *BRCA1/2*, they are not at risk of breast/ovarian cancer. This is certainly not the case: non-carriers are still subject to population risk of cancer (Bredart et al., 1998). Finally, there are significant risks related to negative psychological and emotional reactions to testing, including anxiety, depression, guilt, grief, and relational issues. These are discussed in detail in this appendix.

At-Risk Populations

As previously stated, the general population prevalence of mutations in *BRCA1/2* is thought to be relatively low, estimated at between 1/800 and 1/1000 (Brody & Biesecker, 1998); more recently, narrower estimates closer to 1/300 have been frequently reported (Boyd, 2003). However, there are some groups for which the frequency of these mutations is much higher, and these groups have been the focus of much intense examination. Among Ashkenazi Jews, three ‘founder mutations,’ two in *BRCA1* and one in *BRCA2*, have been identified, carried by as many as 1 in 40 members of this population (Boyd, 2003). Those of Ashkenazi Jewish descent in the United States have been studied fairly intensely, and are considered a very high-risk group. Among Ashkenazi Jews, about 40% of those with ovarian cancer and 12-30% of those with breast cancer are also *BRCA1/2*-positive (Boyd, 2003; Struewing et al., 1997). Founder mutations have also been identified in the Icelandic population, with a gene frequency of one in 173 (Brody & Biesecker, 1998). Genetic testing is more frequently advised for members of these populations, and may be informative even when a family mutation has

not been identified (Brody & Biesecker, 1998). Even for probands, these tests typically require only site-specific testing for the mutation(s) for which the population is at-risk.

Psychosocial Issues Related to Genetic Testing

Ethical issues. Genetic testing brings with it several ethical issues, and some are more or less relevant in the context of HBOC families. At the broadest level, ethical issues related to testing for *BRCA1/2* lie in the communication of risks rather than certainties, an unavoidable consequence of incomplete penetrance of the genes, knowledge about the presence or absence of a *BRCA* mutation is individual, predictive, and probabilistic (Bayeritz, 1998). Such information is, by nature, easily manipulated and may be misunderstood by those who receive cursory or incomplete genetic education (Knoppers & Chadwick, 1994). This occurs in the context of rising rates of breast cancer among young women, mostly due to improvements in detection (Surbone, 2001). However, one result of such improvements has been a cultural perception of breast cancer as an epidemic (S. King & Schottenfeld, 1996) and as one of the most prominent and symbolic issues in modern women's health (Surbone, 2001).

There are several ethical issues related to the availability of testing and the manner in which testing is performed. One of the most basic is a basic respect for patient autonomy, which dictates that those considering genetic testing be fully informed about the impact that the test outcome may have on their lives. Healthcare professionals interacting with patients in the pre-testing phase must ascertain that each individual is undergoing testing to satisfy his or her own needs, and not simply to please other family members or healthcare providers (Kash, Dabney, Holland, Osborne, & Miller, 2000).

Many in the fields of genetics and genetic counseling argue that because breast cancer and, to a lesser extent, ovarian cancer are conditions for which screening, prevention, and treatment exist, genetic testing for *BRCA1/2* is highly advantageous and should be encouraged for those who are potentially at-risk. However, it is important to consider personal and situational factors that may influence individuals' and families' reception of information about cancer risk, as well as potential emotional reactions; for example, individuals with generally high levels of anxiety may be more significantly disturbed or debilitated by news of a positive genetic test result (Lerman, Rimer, & Engstrom, 1991). Individuals have different "desires for information and control in the healthcare setting," (Lerman et al., 1991, p. 1279), and these desires are likely to influence willingness to be tested and possess information about test results, as well as ability to cope with the result. Coping in the context of any result is largely facilitated by proper genetic counseling and education prior to and after testing, which can address both anticipated and actual emotional reactions and can assess and reconcile inaccurate risk perception (Dorval et al., 2000). However, not all individuals undergoing genetic testing for *BRCA1/2* have access to or are required to undergo genetic counseling, as it is not yet a required prerequisite to testing in all contexts.

The familial nature of genetic information brings with it a multitude of ethical issues, including informed consent, individuals' rights to know or not know their own genetic information in light of family members' preferences, confidentiality and privacy issues, and responsibility to future generations (Costalas et al., 2003; R. Green & Thomas, 1997; Juengst, 1999; Van Riper & McKinnon, 2004). For example, informed consent issues can arise when multiple members of a family undergo testing at the same

time and careful, individualized genetic education is not provided to all members before and after testing. Some individuals have even reported that they received mutation test results over the telephone (Van Riper & McKinnon, 2004). Further, informed consent is not always conceptualized as an ongoing process meant to maximize patient autonomy; rather, many view it as a brief communication of the technical and medical aspects of the testing process. Although this form of consenting may facilitate access to genetic counseling in areas where in-person services are unavailable and is a reasonable solution to the currently inadequate number of genetic counselors in the US (Jenkins et al., 2007), a downside is that it may fail to fully disclose the potential social and ethical implications (ASCO, 1996).

Regarding individuals' rights to know or not know, there is some debate over a patient's right to self-determination and the 'right not to know' (ASCO, 1996; Shaw, 1987). This ties in with feelings of responsibility to future generations, in that some seek genetic information not because of a personal desire to know one's mutation status, but because it is useful to others, e.g., children (Rhodes, 1988). Other problems can arise when, for example, members of different generations do not agree on whether or not to pursue testing. For example, a member of a younger generation may choose to be tested based on knowledge that a relative other than a parent is a mutation carrier, even though the parent of the individual has chosen not to be tested. If the tested individual receives a positive result, the parent is also then known to be positive. The tested individual would then have information about the parent's genetic status that the parent her/himself did not wish to have. Similarly, there is debate regarding, for example, whether the daughter of a cancer patient has a right to know her mother's mutation status, or whether a member of a

high-risk family has the right to be informed of her/his risk by a health professional if appropriate family members refuse to share information (Surbone, 2001).

Once genetic information is obtained via testing, there are commonly questions regarding with whom, when, and how to share that information. If a tested individual dies before sharing mutation information with her/his relatives, should that information be available to those family members after the death? Or is that information considered private and inaccessible? More rarely, information that was not being sought can be obtained via genetic testing for known mutations, such as chromosomal abnormalities or non-paternity, and such information can have serious implications for both individuals and families (Kash et al., 2000). Clearly, such situations have the potential to create complex ethical and family dilemmas.

Decision to test. The decision to undergo genetic testing for *BRCA1/2* is a personal one and can be difficult. Within HBOC families, there exists variation regarding individual decisions to test; it is certainly true that not all at-risk individuals choose to undergo testing (Brody & Biesecker, 1998). Further, while surveys of the general population indicate that interest in genetic testing for hypothetical cancer susceptibility genes is high (Croyle & Lerman, 1993), research regarding interest in testing among those with personal and/or family histories appears to be quite complex. Several demographic variables have been associated with the decision to undergo testing for mutations in *BRCA1/2*. Older individuals have been shown in some studies to be more likely to undergo testing than younger individuals (Biesecker et al., 2000; Foster et al., 2004), while other studies have supported the opposite as true (Lerman, Daly, Masny, & Balshem, 1994; Pasacreta, 2003). Regarding gender, women are more likely to choose

testing than men (Pasacreta, 2003), especially married women; married men are more likely to test than unmarried men (Biesecker et al., 2000). Being a parent also appears to be related to choosing to undergo testing (Pasacreta, 2003). Some researchers have explained these associations as related to motivation to test for the sake of one's children; older and married individuals are more likely to have adult children who may also wish to be tested, and so testing these individuals is especially informative with regard to other family members (Biesecker et al., 2000).

Ethnicity appears to be associated with likelihood of testing, in that Ashkenazi Jewish women are more likely to choose to undergo testing than Caucasian women (due to the high incidence of mutations in that population, as discussed previously), who are in turn more likely to test than African American women (Pasacreta, 2003). One's own breast cancer status appears to be unrelated to the decision to undergo testing or to follow through with receiving results afterward (see *variation in decision to receive results*) (Armstrong et al., 2000; Biesecker et al., 2000; Lerman et al., 1996).

In addition to these demographic variables, research on a variety of hereditary cancer syndromes has identified several personality, psychological, family, and relationship characteristics now believed to be associated with choosing to undergo genetic testing. These include a tendency to cope by seeking information, a belief that one has a high level of control over health, low tolerance for uncertainty (Brody & Biesecker, 1998; Croyle, Dutson, Tran, & Sun, 1995), high family cohesion (Biesecker et al., 2000), higher levels of cancer worry (Durfy, Bowen, McTiernan, Sporleder, & Burke, 1999; Foster et al., 2004), high perceived risk of cancer (Glanz, Grove, Lerman, Gotay, & LeMarchand, 1999; Press, Yasui, Reynolds, Durfy, & Burke, 2001; Struewing et al.,

1995), high actual risk of cancer (Glanz et al., 1999; Lipkus, Iden, Terrenoire, & Feaganes, 1999), and a belief that learning about one's cancer risk is important (Armstrong et al., 2000). Another study reported that women who believe that undergoing regular mammograms could be beneficial to health and could give at-risk women control over health were more likely to undergo testing (Tambor, Rimer, & Strigo, 1997). This is consistent with another group's conceptualization of genetic testing as "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, p. 792) and with other research supporting a connection between positive views of medical screening as an important predictor of testing interest (Press et al., 2001).

Brody & Biesecker (1998) reported that individuals who are optimistic are more likely to choose to undergo testing, but this finding contradicts another study which found that the reverse was true (Lerman et al., 1997). Reported intention to undergo testing at baseline was related to a greater likelihood of testing over time in multiple studies (Lerman et al., 1997; Pasacreta, 2003), suggesting that once an individual enters the healthcare environment in the context of an appointment with a genetic counselor or other health professional who can facilitate testing, the patient has already made a decision regarding testing. Perception of susceptibility to cancer and higher perceived risk of cancer are both reported to be key predictors of interest in genetic testing (Pasacreta, 2003). Other researchers have reported that those who perceive a large number of personal benefits as a result of testing, accompanied by few personal costs, are more likely to choose to be tested (Cappelli et al., 2001), while those with lower levels of cancer-specific distress are less likely to choose to undergo testing (Lerman et al., 1997).

Those who self-refer for genetic counseling are more likely to undergo testing than those who are referred by physicians (Schwartz et al., 2000).

Multiple studies have investigated the impact of psychological distress and knowledge of genetics and breast cancer risk factors on intention to undergo testing and have not discovered a significant relationship (Pasacreta, 2003). Other factors that have been hypothesized to be related to the decision to test but in research appear to be unrelated include “family conflict, family expressivity, depression, spirituality, and self-esteem levels,” (Biesecker et al., 2000, p. 261).

One of the most significant predictors of interest in genetic testing for *BRCA1/2* is perceived susceptibility to breast/ovarian cancer, whereby women who more strongly believe that they are susceptible to cancer are more likely to report that they are interested in genetic testing (Durfy et al., 1999; Pasacreta, 2003; Schwartz et al., 2000). However, other researchers have found no relationship between these two variables in studies of racial differences in interest in genetic testing that focus primarily on African-American women (Lerman et al., 1999; Lipkus et al., 1999).

Variation in decision to receive results. It is interesting to note that not all individuals who choose to undergo genetic testing for hereditary cancer will actually receive their results. Some individuals choose to delay receipt of test results or change their mind about wanting to know about their mutation status after submitting a biological sample. One study of individuals from hereditary breast cancer families found that only 43% of members eligible for testing elected to receive their results; those who did so were more likely to be female, have already had cancer, have higher levels of education, and have health insurance (Lerman et al., 1996); there was a positive

correlation between receipt of test results and knowledge about *BRCA* testing, high perceived benefits of testing, and higher levels of cancer worry and distress (Lerman et al., 1996; 1997).

According to the limited research that has investigated genetic testing and spirituality, it appears that an inverse relationship exists between spirituality and likelihood of receiving test results, and that this relationship is mediated by perceived risk of breast cancer. In a study of 290 untested female breast cancer patients with a >20% chance of carrying a breast cancer mutation in the Washington, DC, area, those who self-identified as “highly spiritual” were significantly less likely to receive test results than those who did not identify as highly spiritual. However, this relationship only held true for women with low perceived breast cancer risk; there was no significant difference between highly spiritual and non-spiritual women in likelihood of receiving test results when perceived breast cancer risk was high (Schwartz et al., 2000).

Regarding family factors, previous research has demonstrated that women are more likely to choose to undergo testing if a family mutation has been identified (Pasacreta, 2003). When family history of cancer was conceptualized as “presence of first-degree relatives with breast or ovarian cancer,” there was no impact on the likelihood of undergoing testing (Biesecker et al., 2000, p. 258). However, another study demonstrated a positive association between the number of first-degree relatives affected with breast cancer and the likelihood of requesting test results after submitting a sample for testing, but that this association did not hold true for the number of relatives affected by ovarian cancer (Lerman et al., 1996). When a group of women with one first-degree relative affected by breast cancer was compared with a group of women with one first-

degree relative with ovarian cancer or more than one first-degree relative affected by breast cancer, the latter group had a significantly higher rate of testing (47% v. 68%) (Lerman et al., 1997). However, a similar study comparing similar groups did not detect a difference in the rate of requests for genetic test results (Schwartz et al., 2000).

Perceived barriers to testing. A number of important barriers to testing have been identified in recent research, and decrease the likelihood that an at-risk individual will elect to undergo testing (Pasacreta, 2003). These include apprehension about the result, effort involved in making multiple trips to a genetics clinic, and taking time away from work and family to undergo testing (Foster et al., 2004). Some women predict, prior to testing, that they will experience an increase in anxiety level (Pasacreta, 2003) or intense negative emotions (Bredart et al., 1998; Patenaude et al., 1996) as a result of a positive test result, and these are considered barriers to testing. Some individuals are stressed by other medical problems, and feel that this inhibits them from pursuing genetic testing for *BRCA* (Patenaude et al., 1996). Finally, some research participants have identified fear of potential discrimination (e.g., obtaining or maintaining health insurance, employment) as a barrier, in addition to concern about potential negative impact on interpersonal relationships (Bredart et al., 1998; Patenaude et al., 1996).

Motivation & perceived benefits of testing. Motivations to undergo genetic testing for *BRCA1/2* are multiple and varied. In various studies, perceived benefits of testing have been shown to have a positive relationship with intention to test, or no relationship at all (Pasacreta, 2003). Many women report that their primary reason for testing is simply to know for sure whether or not they carry a mutation in *BRCA1* or *BRCA2* (Lodder et al., 1999) and become knowledgeable about one's own level of risk

(Thompson et al., 2002), to inform risk management strategies and lifestyle changes (d'Agincourt-Canning, 2005; Thompson et al., 2002), to provide relief from the burden of uncertainty (Lerman et al., 1996), or to provide reassurance in the case of a negative result (Lerman & Croyle, 1996; Pasacreta, 2003; Patenaude et al., 1996).

In general, individuals with children are more likely to choose to undergo genetic testing (Meijers-Heijboer et al., 2000). Multiple studies report that a common motivation to test is for the sake of children or to learn about children's risk (Burgess & d'Agincourt-Canning, 2001; Foster et al., 2002; J. Green et al., 1997; Lodder et al., 1999; H. T. Lynch et al., 1997; Patenaude et al., 1996; Tercyak, Peshkin, Brogan et al., 2007); this motivation is cited by as many as 90% of women with children (Pasacreta, 2003). Similarly, about one-third of women who do not yet have children or are considering having more children cite knowing risks for future children or planning important life decisions (e.g., future childbearing) as important motivators for genetic testing (Foster et al., 2002; Lodder et al., 1999; Pasacreta, 2003). Some women cite the desire to allow cascade testing to proceed (i.e., testing generation-by-generation from the top down) (K. E. Smith, West, Croyle, & Botkin, 1999) as a motivation to pursue testing (Tercyak, Peshkin, Brogan et al., 2007). More generally, many individuals report that the desire to help family members is a large component of what motivates them to undergo testing (Geller, Doksum, Bernhardt, & Metz, 1999; Vernon et al., 1999), or that they feel a responsibility to get tested out of commitment to family members (Burgess & d'Agincourt-Canning, 2001; Hallowell et al., 2003).

Many women choose to undergo testing to legitimate their perceived need for more intense cancer screening than is accessible to members of the general population

(Foster et al., 2004; Pasacreta, 2003), or to more fully understand the necessity of intensive screening (Lodder et al., 1999; Pasacreta, 2003). That is, women are motivated to undergo testing so that they can use potentially positive results to demonstrate to their physicians why they should be screened more often or more intensely than a doctor might otherwise recommend. By the same token, women who test for this reason can legitimately alleviate their concerns about screening if their genetic test result is a true negative. Similarly, a large proportion of women in HBOC families report that they choose to undergo genetic testing in order to make decisions about risk-reducing surgery (Lodder et al., 1999; Patenaude et al., 1996).

Young women frequently cite a desire to prepare for the future and alleviate uncertainty as important motivations to test (Decruyenaere, Evers-Kiebooms, & Van den Berghe, 1993; Foster et al., 2002; Messien, Mastromaura, Kiely, McNamara, & Myers, 1991; Tibben et al., 1994; Van Asperen et al., 2002). However, due to the inability of genetic testing for *BRCA1/2* to provide definitive information about lifetime cancer risk, and due to the relatively high frequency of uninformative tests, uncertainty may not be alleviated for women in many situations (Croyle & Lerman, 1993). This ongoing uncertainty may play a key role in the coping process of individuals, couples, and families, as feelings of uncertainty add difficulty to the already arduous task of threat appraisal and choosing of coping response (Lazarus & Folkman, 1984). This is especially true in the case of an uninformative test result, because it offers no new information to direct behaviors (Baum, Freidman, & Zakowski, 1997).

Timing of genetic testing. Several researchers have investigated the timing of decision-making regarding genetic testing within the context of learning about the

possibility of genetic risk. Individuals may choose to pursue testing within several weeks, or may wait many years to be tested. A 1999 Dutch study found that the majority of 118 individuals at 25% or 50% risk of having inherited a mutation in *BRCA1* or *BRCA2* chose to undergo testing within two months or receiving their test results (Lodder et al., 1999); however, this was likely a highly motivated population given their willingness to participate in the research study.

Generally, once individuals make the decision to pursue genetic education and counseling, which precede testing, they will more often than not choose to be tested; that is, it is relatively uncommon for an individual to go through the genetic education and counseling process and then decide not to be tested, although this does occur in some cases (Botkin et al., 1996; Lerman et al., 1996). For example, 135 of 172 (78.5%), individuals who underwent education and counseling sessions in the context of a study of HBOC families enrolled in a research protocol at the National Cancer Institute (Struewing et al., 1995) chose to go through with testing (Biesecker et al., 2000). There are also some individuals who follow through with testing but choose not to learn their mutation status (Pasacreta, 2003).

Family factors. In the existing research, there are some reports of individuals undergoing genetic testing for *BRCA1/2* not because they truly wanted to, but because they felt obligated or coerced by their families. For example, some male research participants have reported that they felt obligated to undergo testing because female relatives wanted information regarding their own health, and as males they had no right to stand in the way of the pursuit of information about a hereditary syndrome that has its most profound effect on females (Dudok de Wit et al., 1996).

Psychological distress through the testing process. There has been a significant amount of research regarding the psychological and emotional effects of genetic testing for a wide variety of medical conditions, including HBOC. Various studies have demonstrated that women undergoing genetic testing for *BRCA1/2* experience distress prior to testing, while they are awaiting results, and after they have learned their mutation status, and have investigated the implications of choosing not to be tested despite membership in a high-risk family. Since many women undergoing testing are actively experiencing or have recently experienced cancer-related issues within their families, it can be difficult to tease apart the distress that women feel as a result of the genetic testing process from that which they experience as a consequence of their membership in a family with many cases of cancer, which may include bereavement over ill or lost loved ones and fear of developing the disease oneself (Foster et al., 2002; Lobb et al., 2004). It has been suggested that women in hereditary cancer families may need to openly “discuss their feelings of loss and anxiety about the future first, to enable them to focus on the genetic issues,” (Lobb et al., 2004, p. 325).

Psychological distress is also an important consideration in the context of adherence to screening recommendations for high-risk women (Lerman et al., 1993), and research illuminates the association between high levels of distress and attempts to avoid potentially disturbing cancer-related experiences such as screening (Bredart et al., 1998), and women may refuse or avoid mammography, clinical breast examination, and breast self-examination. This is in keeping with research on other hereditary cancer syndromes, such as colon cancer, in which research has identified screening avoidance as an attempt to mitigate distress associated with fear of cancer (Lerman & Croyle, 1996). While it is

interesting to assess psychological functioning on a number of different dimensions at unique points in the testing process, it is also necessary to consider changes across time, as both general and cancer-specific distress at baseline (prior to genetic education/testing) are highly predictive of levels of each later in the process (Esplen et al., 2004; Pasacrete, 2003).

Distress prior to testing. Here, the term “prior to testing” refers to the period from the point at which an individual learns about the presence or possible presence of a hereditary cancer syndrome in her family to the point when she submits a biological sample for genetic testing. It includes the decision-making process, which varies in length and complexity from person to person, as discussed previously. At this stage of the testing process, women are said to be at high risk of carrying a mutation, and are *potentially* at increased genetic risk of cancer.

It is important to note that levels of distress that are extremely low are not necessarily the most adaptive in this context, as they may indicate that an individual is engaging in denial/avoidance behavior rather than adequate mental preparation for the test; rather, somewhat higher levels of distress may be seen as normative among members of this population, as they reflect preparatory worrying during the pre-test preparation period (Dudok de Wit, Tibben, Duivenvoorden et al., 1997). The emotional issues raised by the experience of living in a hereditary breast and ovarian cancer family make the presence of some anxiety a part of the normative experience for members of this population, as they struggle to integrate fear of the illness, the threat of premature death, and the loss and grief experienced during other family members’ cancer struggles with their attempts to proceed normally through the life-course (Lobb et al., 2004). Of course,

very high levels of distress are potentially problematic when they interfere with an individual's ability to manage everyday life.

Multiple researchers have noted that general distress may be higher among women who have experienced multiple cancers within the family prior to undergoing genetic testing (Erblich, Bovbjerg, & Valdimarsdottir, 2000; Lerman et al., 1997; Lloyd et al., 1996). However, research comparing women approaching genetic testing with those who have one first-degree relative affected by breast cancer but are not involved in the genetic testing process (Lerman et al., 1997) and with members of the general population (Croyle, Smith, Botkin, Baty, & Nash, 1997) found no significant differences in either comparison on measures of depression and anxiety, but have noted that although mean levels of distress are similar, women in the pre-testing phase display more variability in distress levels (Lerman, Kash, & Stefanek, 1994; Lodder et al., 1999). Similarly, a study published before the identification of *BRCA1/2* investigating psychological functioning of women with family histories of breast cancer found that fully 27% of them experienced levels of psychological distress high enough to warrant counseling (Kash et al., 1992). That study also found that psychological distress among women in high-risk families was associated with a greater number of perceived barriers to screening, having fewer social supports, and low social desirability (Kash et al., 1992). Relevant outcomes of higher levels of distress included poorer adherence to recommended screening (breast self-exam, mammography) (Lerman et al., 1993) and poor attendance for clinical breast examination (Kash et al., 1992), both of which could have dangerous long-term health implications for high-risk women.

When distress is present among members of this population, it often appears to be cancer-specific rather than general, and occurs with increased frequency just prior to receiving a genetic test result (Croyle et al., 1997; Foster et al., 2002; Kash et al., 1992; Lerman et al., 1997). Primary symptoms of cancer-specific distress in healthy women at high risk of cancer are heightened levels of awareness about cancer, intrusive thoughts, and avoidance (Zakowski et al., 1997). Studies have demonstrated that in the weeks prior to genetic testing for *BRCA1/2*, young women report higher levels of cancer-specific distress than older women (Foster et al., 2002; Lerman et al., 1993; Lloyd et al., 1996; Lodder et al., 1999), and that younger women worry about developing cancer more often and find this distress more disturbing than do older women (Foster et al., 2002). Concern about problems as a result of positive carrier status and probable consideration of risk-reducing surgery as a cancer prevention strategy upon discovery of positive carrier status have been identified as predictors of cancer-specific distress prior to receipt of test results among women approaching genetic testing for *BRCA1/2* (Lodder et al., 1999). The frequency of these beliefs among the study population was quite high: one-third anticipated that they would experience an increase in personal problems as a result of a positive test result, close to half reported that they intended to undergo risk-reducing mastectomy and/or oophorectomy if they found out they carried a mutation in *BRCA1/2*. Awareness of the potentially serious consequences of HBOC was also identified as a predictor of cancer-specific distress in this study. Similarly, it has been suggested that for women approaching genetic testing, it is not the testing process *per se* that causes distress, but rather that feelings of distress are a function of acknowledged risk (A. W. Smith et al., 2008).

In a Dutch study of 85 unaffected women at risk of carrying a mutation in *BRCA1/2*, general distress, measured by levels of anxiety and depression, was predicted by “pessimistic personality and not being inclined to express one’s emotions” (Lodder et al., 1999, p. 910). Having many affected relatives who experienced cancer onset before the age of 40, having young children and being under age 40 also predicted increased levels of anxiety, but not depression. Women who knew or had known relatives with metastatic breast/ovarian cancer displayed significantly higher levels of depression than women who had not known such relatives (Lodder et al., 1999). Other risk factors for cancer-specific distress include the disease having had a greater negative impact on one’s life prior to testing, and expecting relief in the event of a negative test result (Dudok de Wit, Tibben, Duivenvoorden et al., 1997).

Because the pre-testing phase of the testing process begins as soon as an individual is aware of the potential presence of a cancer-related genetic mutation, some individuals spend many years in this stage, and it may begin well before an individual is old enough to pursue testing. To explore the experience of women in this situation, a Canadian study investigated the psychological implications for adolescent daughters of women with breast cancer, comparing them to adolescent females without breast cancer histories. Although the two groups did not differ in overall psychological functioning, the daughters of affected women reported significantly more worries about their future health and their risk of breast cancer, and significantly more of them saw themselves as vulnerable to breast cancer (Cappelli et al., 2005). This study demonstrates that the cancer specific distress experience by young women in high-risk families may begin as

early as adolescence, and raises additional questions about the long-term implications of experiencing this type of distress for such a large portion of one's life.

An Israeli study used the Brief Symptoms Inventory (Derogatis & Spencer, 1982) and Impact of Events Scale (Horowitz et al., 1979) to measure both general and cancer-specific anxiety and distress among women with at least one first-degree relative with breast cancer. The 230 participants fell into three categories: 176 healthy women whose mothers had been affected by breast cancer, 34 healthy women whose sisters had been affected by breast cancer, and 20 healthy women whose mothers and sisters had both been affected by breast cancer. The women completed questionnaires during a one-day educational conference for healthy relatives of breast cancer patients; results of this study must be considered in this context, as the salience of breast cancer issues was likely higher given the concurrent involvement in this educational program. Results indicated that all of the subjects experienced high levels of psychological distress; over half of the participants fit into the very high psychological distress group, and these participants displayed significantly higher scores on the IES. Women in the highly-distressed group were also significantly less likely to be married, supporting the notion of the distress-protective effect of having a partner. Women who had both a mother and a sister affected by breast cancer demonstrated consistently high IES scores across subscales, possibly related to even greater threat of inheriting breast cancer and increased difficulty in denying this threat compared with women with only one affected relative (Baider et al., 1999). This high level of threat combined with the salience of breast cancer during a cancer educational conference likely explains these phenomenally high distress levels,

which are much higher than those observed among a similar population attending a cancer prevention center (Kash et al., 1992).

A British study of 158 women assessed at the time of genetic counseling for hereditary breast/ovarian cancer risk and three months later indicated that 30.6% of participants met the criteria for psychological distress at baseline, and 26.4% met the criteria for psychological distress at follow-up, a statistically non-significant difference (Hopwood et al., 1998). At follow-up, 13.3% of the women were diagnosable with psychiatric disorders, including depression, anxiety, panic disorder, and adjustment disorder. Interestingly, none of these women reported concerns about cancer risk as the precipitating factors in their psychiatric problems; rather, they cited “work-related stress, financial problems and illness in the family (other than breast cancer)...relationship problems with partners...and blocked or unresolved grief...these problems were long-standing and pre-dated the risk assessment experience,” (Hopwood et al., 1998, p. 405). These observations support the notion that women may benefit from increased guidance regarding assimilating risk information into family interaction and dealing with the impact of breast cancer in the family (Hopwood et al., 1998).

In a broader study of individuals approaching testing for a range of late-onset, autosomal dominant, heritable disorders (e.g., Huntington’s Disease), a group of Dutch researchers identified predictors of pre-test distress including experience with the relevant disease in close family, the perception that the relatives’ disease had a significant impact on their lives, the expectation that a positive test result would have a significant negative impact on their own lives, and the expectation of relief as a result of a negative test result (Dudok de Wit, Tibben, Duivenvoorden et al., 1997). Results from the Intrusion and

Avoidance subscales of the IES also indicate that potential testers who expected an increase in distress and/or personal problems as a result of the test displayed higher levels of avoidance (i.e., increased frequency of consciously avoiding thoughts about breast/ovarian cancer), while those who could more readily envision and anticipate life as mutation carriers had higher levels of intrusion (i.e., increased frequency of thinking about breast/ovarian cancer when one does not want to) (Dudok de Wit, Tibben, Duivenvoorden et al., 1997). This has been confirmed in subsequent research, which has found that the majority of women approaching genetic testing for *BRCA1/2* experience some intrusive and avoidant thoughts about developing cancer (Foster et al., 2002; Lodder et al., 1999; Reichelt, Dahl, Heimdal, & Moller, 1999), but that overall depression and anxiety scores are relatively low (Tercyak, Lerman et al., 2001).

Distress for those who decline testing. As previously stated, not all women who are at significant risk of carrying a mutation in *BRCA1/2* choose to undergo testing. Because they often end their involvement in the genetics/research component of the healthcare system after choosing not to be tested, very little research exists regarding the psychological and emotional well-being of members of this population. A study of individuals who either declined testing altogether or submitted biological specimens for genetic testing but declined receipt of their results found that there were significant increases in depression at six month follow-up, especially in those who had high-levels of stress symptoms prior to testing; in fact, “those who declined genetic testing were about eight times more likely to become depressed than those who were tested and found to be non-carriers,” (Lerman et al., 1998, p. 1653). In addition, there is evidence that

individuals in high-risk families who decline testing may continue to experience higher than normal levels of anxiety (Lerman et al., 2002).

Not all individuals who decline testing will remain permanently in that state. A variety of life-cycle triggers occurring across the lifespan may provide motivation to revisit the possibility of genetic testing, including a new illness in a previously tested individual, an impending marriage or birth, entrance of members of the younger generation into adulthood, or identification of additional mutation carriers within a family (Martin & Wilikofsky, 2004).

Distress while awaiting results. The period of time from submission of a biological sample for genetic testing to receipt of results may be several weeks to several months, depending on whether the test involves full sequencing. A typical timeframe is about six to eight weeks (Lodder et al., 1999). Because women often undergo testing in a research context, a good deal of data has been collected regarding levels of psychological distress during this phase of the genetic testing process. Overall, levels of psychological distress seem to be comparable to that present in the general population, with only about 7% of subjects in various studies reporting depression scores that are considered “borderline” or higher (Pasacrete, 2003). Some studies have concluded that women awaiting test results display higher than average levels of distress during this period (Audrain et al., 1997), and researchers have identified elevated intrusion and avoidance scores (Lodder et al., 1999) as well as some impairment in role and sexual functioning as the major issues identified in distressed women in this phase of testing (Pasacrete, 2003). Psychological distress prior to submission of a biological sample is highly predictive of

psychological distress while awaiting results (Pasacrete, 2003), as are several of the other predictors of distress listed in the previous section (Lodder et al., 1999; Pasacrete, 2003).

Distress following disclosure. Breast and ovarian cancer “pre-vivorship” (the experience of living with increased genetic risk of cancer) (FORCE, 2005) is still a relatively new and unexplored situation, as it has only been a bit longer than a decade since the *BRCA1/2* mutations were discovered and testing is still not commonplace. The experience of being a “mutation carrier,” therefore, is not yet fully understood, especially by those outside of the cancer and cancer family communities (Dagan & Gil, 2004). Individuals who receive positive mutation test results are plunged into this stressful situation and may deal with it in very different ways.

Immediate psychological distress reactions to the testing process can have important implications for long-term well-being because they may interfere with comprehension of risk information during genetic counseling (Janis & Mann, 1977) and in the weeks and months following receipt of test results (Lerman & Croyle, 1996). Both acute and chronic psychological distress may influence comprehension of risk information after the formal genetic testing process, and may impact medical decision-making as well; research indicates that highly anxious women continue to overestimate their risk even after genetic counseling; researchers note that “this raises concerns about the appropriateness of medical decisions, such as risk-reducing surgery, that may be based on erroneous beliefs concerning personal risk,” (Lerman & Croyle, 1996, p. 191). High levels of anxiety and distress also decrease adherence to clinical- and self-breast exams (Kash et al., 1992; Lerman & Croyle, 1996) and to mammography screening (Lerman & Croyle, 1996; Lerman et al., 1993).

In general, levels of distress among women receiving results from genetic tests for *BRCA1/2* mutations are similar to those of a normal population; however, there is significant variability in distress levels (Botkin et al., 1996; Lodder et al., 1999). Not surprisingly, some researchers have identified higher levels of psychological distress among those who receive positive test results, especially immediately following receipt of test results (Croyle et al., 1997; Lodder et al., 2001; K. E. Smith et al., 1999; Tercyak, Lerman et al., 2001); in one study, fully 27% of women identified as high risk were distressed enough to warrant counseling (Kash et al., 1992). Another study noted that positive test results were associated with transient distress as measured by the IES (A. W. Smith et al., 2008). While consistently high levels of stress are uncommon, many women have reported that they experience acute negative emotional reactions such as “anxiety, depression, shock, surprise and feelings of neglect” (Appleton et al., 2000, p. 515) immediately following receipt of their mutation test results, as well as in the wake of trigger events such as receiving an appointment letter for a regular screening, waiting for a past-due appointment, having a screening test performed, and receiving the results of screening tests (Appleton et al., 2000; Valdimarsdottir et al., 1995). Heightened sensitivity to breast cancer cues, including changes in one’s own breast tissue, media reports, and family illnesses, prompts anxiety and/or avoidance reactions in many high-risk women. All of this may become a part of a cycle of emotional and psychological highs and lows that occur over time and may coincide with the annual screening cycle (Appleton et al., 2000). Consistently high levels of distress have also been reported by women in some studies, and it is usually associated with high levels of baseline distress, just as low levels of baseline distress are associated with consistently low levels of

distress throughout the testing process (A. W. Smith et al., 2008). This further supports the notion that the testing process itself is not the source of stress for women in HBOC families, but rather that one's response to the notion of being potentially "at-risk" occurs prior to testing and remains relatively constant over time.

Most of the research that exists on the psychological impact of learning one's *BRCA1/2* mutation status has followed women for up to one year after result disclosure, representing only short-term follow-up (Appleton et al., 2000; Lerman et al., 1996). The bulk of this research finds that psychological problems as a result of *BRCA1/2* testing are less frequent and less problematic than anticipated, and most individuals display levels of psychological disturbance within general population norms (Bredart et al., 1998; Lerman et al., 2002; Pasacreta, 2003). In fact, studies of both carriers and non-carriers have identified *decreases* in general distress of approximately 20% within one to two weeks after result disclosure, as compared with pre-test levels, with even greater reductions in distress at six-month follow-up (Croyle et al., 1997), and reductions in depression and functional impairment one month after result disclosure (Lerman et al., 1996). These findings may reflect the sense of relief that comes with having information about one's mutation status, after a potentially lengthy period of wondering. However, distress related to uncertainty may re-emerge as high-risk women begin to face life as confirmed mutation carriers and must come to grips with a continuing sense of uncertainty and ambiguity about whether, when, and where cancer will develop (Lerman & Croyle, 1996).

Some researchers have identified levels of psychiatric morbidity in the post-genetic testing population that are higher than population norms (e.g., Hopwood et al.,

1998); interestingly, in the Hopwood study, women reported that the distress they were experiencing was not related to their genetic testing results, but rather to more universal stressors such as marriage, work, children, etc., common stressors for pre-menopausal adult women. Of those referred for psychological help, very few reported cancer concerns as their primary issue. Therapeutic work focused on issues of unresolved grief, loss, and relationship problems, often related to their positive mutation status. The researchers concluded that “a minority of women may need help with the impact of breast cancer in the family,” (Hopwood et al., 1998, p. 402). It has been proposed by some that those with higher levels of intrusive thoughts may be more psychologically vulnerable following receipt of test results (Lerman et al., 1998); this is in keeping with research demonstrating that individuals who underestimated the emotional impact of result receipt prior to testing are more likely to experience distress in the six months following receipt of results (Dorval et al., 2000). The limited research on long-term psychological effects of genetic testing suggests that a minority of individuals continue to experience cancer-related distress more than two years after completion of the testing process (Lloyd et al., 1996).

Significant increases in cancer-specific distress, including sadness, guilt, and anger, in the period immediately following receipt of test results have been noted in several research studies (Brody & Biesecker, 1998; H. T. Lynch et al., 1997). Test result is one of the most reliable predictors of post-test psychological functioning (Esplen et al., 2004), although individuals may experience distress in the wake of either a positive or negative result. Significant differences were identified between overall levels of both cancer-specific and general distress when comparing carriers and non-carriers (Bredart et al., 1998; Croyle et al., 1997), as well as in reductions in both types of distress over time,

with non-carriers finding more relief (Lerman et al., 1998; Lerman et al., 1996; K. E. Smith et al., 1999). It has been noted that the decrease in distress inherent in a negative test result, due to relief of worry about increased risk, is not afforded to mutation-positive women, who may also be more likely to experience an increase in distress (Esplen et al., 2004).

Several studies have investigated at-risk individuals' ability at pre-test to anticipate levels of post-test psychological distress. This is thought to be important because those who make an accurate appraisal of post-test emotional experience may be less likely to encounter unexpected difficulties after they undergo testing, and in turn may be more compliant with screening and prevention recommendations (Lerman et al., 1993). Studies on anticipated reactions consistently point to anxiety and uncertainty about if and when cancer will develop as emotions that individuals expect to experience after testing (Bredart et al., 1998; Lerman & Croyle, 1996). In addition, those who have first-degree relatives who have experienced ovarian cancer are more likely to anticipate anxiety (77%), depression (80%), and impaired quality of life in reaction to a positive mutation test result (Bredart et al., 1998), likely because they have witnessed the devastating experience of that cancer in a close loved one. Expectation of a negative test result has also been identified as a predictor of post-test psychological distress (Esplen et al., 2004). This is in keeping with research on stressful events suggesting that negative or catastrophic events are met with a greater distress response when they are unexpected (Lazarus & Folkman, 1984; Ortony, Clore, & Collins, 1988).

One study identified significant differences between those affected and unaffected by cancer regarding how they thought they would react emotionally to their testing

outcomes, whereby unaffected individuals overestimated their emotional reactions and affected individuals underestimated these responses. Only 30% of unaffected individuals underestimated their emotional responses (Dorval et al., 2000). Investigators in this study also noted a positive association between one's ability to accurately anticipate emotional response to mutation test result and one's post-disclosure psychological adjustment, with underestimation associated with greatly increased psychological distress at six-month follow-up.

Family cancer experience has also been repeatedly identified as an important factor influencing psychological distress in the genetic testing process for *BRCA1/2*. Those who have been exposed to more cases of cancer within the family are often more psychologically distressed by the process of genetic testing and by a positive result. The amount of social support one has access to, both within and outside of the family, is also thought to influence psychological coping (Esplen et al., 2004).

The stress experienced as a result of learning that one is mutation-positive may continue in the long-term and may be present even after preventive, risk-reducing measures are taken. In a Dutch study comparing women who had undergone RRSO with women who chose screening to manage ovarian cancer risk, 10% of women in the RRSO group reported symptoms consistent with the presence of post-traumatic stress disorder (PTSD), with breast and ovarian cancer risk as an underlying factor (Madalinska et al., 2005). This suggests that for some women, even the most extreme and effective risk management strategies will not be sufficient to reduce cancer-specific distress.

Researchers have hypothesized that an individual's age may play an important role in determining one's psychological distress in response to test results. There are

several factors that may influence both older and younger women's reactions to a positive mutation test result. Younger women may experience more psychological distress because they are forced to consider the threat of cancer over a longer period of time. Additionally, concerns about body image may be more salient for a younger population. Distress in this group may be mitigated by the fact that they have a greater period of time remaining to create their families while their cancer risk is still relatively low, as opposed to women in middle age who may be facing the more imminent threat of cancer and/or being encouraged to undergo risk-reducing surgery even though they may not have achieved their ideal family size. Generational differences in the mutation-positive experience are also significant. For older women receiving positive mutation test results, a cancer diagnosis may be equated with death, due to the fact that historically, treatments for breast and ovarian cancer were not as good as they are now and a generation ago, death due to cancer was a reasonable expectation. However, for young women who have come of age in an era of improving medical technology and availability of excellent treatment and improving survivability, breast and even ovarian cancer is more likely viewed as a challenge that can be overcome, and so attitudes are formed in light of that perception (d'Agincourt-Canning, 2005). Resources that are related to and vary with age, such as marital status, education level, income, and access to health insurance may also play a role in women's level of psychological distress as a result of genetic testing (Botkin et al., 1996).

As a whole, this literature suggests that some individuals may experience a relatively brief (several weeks to several months) crisis response immediately after learning of their positive mutation status, but that the majority return to relatively normal

levels of psychological functioning in the weeks and months following disclosure (Pasacreta, 2003). The reduction of uncertainty and doubt facilitated by the completion of the genetic testing process is a clear psychological benefit for many women who choose to undergo testing, and may be present regardless of test outcome (Bredart et al., 1998). However, the specific types of psychological disturbance experienced by women in high-risk families varies, and may include anxiety, depression, intrusive thoughts, guilt, and family stress.

In the research context, women often report that they expect to experience anxiety, depression, and impaired quality of life in the aftermath of a positive genetic test (Bredart et al., 1998). However, these are not the only reactions that occur; rather, emotional, psychological, and relational experiences following positive mutation test results are quite varied. Specifically, researchers have examined anxiety, grief, depression, anger, guilt, functional health status, family stress, and family communication.

Anxiety. Psychological responses to testing that take the form of anxiety may manifest in many different ways, including “hypervigilance, intrusive thoughts...sleep disturbances, confusion and somatic symptoms, and persistent worry about the future,” (Lerman & Croyle, 1996, p. 193) and are relatively common among women with significant family histories of breast/ovarian cancer (Baider et al., 1999) and among those who receive positive *BRCA1/2* mutation-test results (Croyle et al., 1997). The relationship between testing and anxiety is among the most frequently studied of psychosocial issues in *BRCA*-related research; researchers have long acknowledged that individuals and families undergoing screening may experience this type of psychological

distress (Coughlin et al., 1999). Understanding of the dynamics surrounding anxiety and mutation test result disclosure is important because anxiety has been shown consistently to reduce cancer risk comprehension due to interference with information processing abilities (Janis & Mann, 1977; Lerman et al., 1995); this, in turn, may result in failure to adhere to screening or risk-reduction recommendations as women attempt to keep their anxiety low by avoiding cancer-related experiences, including screening (Lerman et al., 1996).

In longitudinal studies, baseline levels of anxiety are reliable predictors of post-test anxiety, with carriers experiencing higher levels of distress than non-carriers (Croyle et al., 1997). Significant frequencies of anxiety have been identified in various *BRCA1/2* study populations (Esplen et al., 2004). In one study, a reduction in anxiety among non-carriers was detected at one- to three-weeks post-disclosure, while levels of anxiety increased slightly among carriers (Lodder et al., 2001). Another study identified reductions in anxiety across *all* participants, regardless of mutation status, suggesting that part of the anxiety that is experienced during the pre-test and awaiting-results phases maybe related to uncertainty, which is relieved by receipt of test results even if they are positive (Croyle et al., 1997). However, researchers note that learning of a positive test result may represent a shift from uncertainty about whether or not one has the mutation to uncertainty about whether or when cancer will develop, rather than a relief from uncertainty altogether (Murphy, 1999), and this transition may elicit an anxiety response in some individuals (H. T. Lynch et al., 1997).

Family history of cancer and the extent to which individuals have personally witnessed the cancer experience(s) of a loved one or loved ones has been studied with

regard to women at high risk of breast cancer (Gilbar & Borovik, 1998); it may also be an important factor in determining their psychological distress response to learning their mutation status (d'Agincourt-Canning, 2005). In part, this may reflect the experiences of those who were diagnosed with and treated for breast and ovarian cancer prior to the availability of successful treatments, at a time when a diagnosis was often legitimately equated with death (Rosenbaum & Roos, 2000). Even in the face of modern scientific reality, such powerful experiences may be important components of individuals' processing of genetic risk information (d'Agincourt-Canning, 2005).

A Swedish study of 87 women undergoing presymptomatic genetic testing for *BRCA1/2* mutations concluded that as a whole, the group experienced a significant decrease in anxiety mean scores over time, regardless of mutation test result, with anxiety levels similar to population norms before testing and one-year post-test, but spiking in the weeks and months immediately after the test and receipt of results (Arver, Haegermark, Platten, Lindblom, & Brandberg, 2004).

Several factors have been associated with increased experience of anxiety after receipt of mutation test results. A Canadian study of 157 women at high familial risk of ovarian cancer concluded that women who scored above the cutoff for clinical anxiety were "significantly more worried about their daughter's future, were more likely to have altered plans for the future, reported greater problems with work, were more worried about being at risk of cancer, reported greater problems with their partner, perceived their partner as less supportive, and had a trend to be younger," (Robinson et al., 1997, p. 201). This suggests that anxiety is related to a variety of other problems that may be present in

the period following testing, and that reductions in anxiety may have further benefits in eliminating these other symptoms and problems.

It has been noted that anxiety is more common among those who had previously made unalterable life decisions based on an incorrect assumption that they were at increased risk (e.g., women who have undergone risk-reducing surgery prior to testing, only to find out that they do not carry the mutation), and among those whose distress levels prior to testing were higher than average (Lerman & Croyle, 1996). One study notes that mutation carriers who harbor an intense preoccupation with cancer “may experience the same emotional spectrum of cancer survivors—a sense of personal vulnerability and only tenuous longevity,” (Bredart et al., 1998, p. 176).

One of the most commonly reported symptoms of anxiety among women in HBOC families is *intrusive thoughts, usually measured by the IES*. One study found that in the weeks following result disclosure, both carriers and non-carriers experienced a diminution of both intrusive thoughts and avoidant behaviors (Lodder et al., 2001); this suggests that the root of these intrusive thoughts and avoidant behaviors might not be just the phenomenon of being a potential mutation carrier, but also the *uncertainty* associated with the pre-test experience. Other research on group intervention has demonstrated that women who are able to discuss their feelings of uncertainty, fear, guilt grief, etc. in an open, authentic manner in a group environment experienced a significant reduction in their experience of intrusive thoughts (Esplen et al., 2004).

Grief. Grief is a relatively common response to discovery of one’s positive *BRCA1/2*-mutation status, and may occur most intensely in the first six months after receipt of test results, which can be thought of as a loss experience (Ringdal, Jordhoy,

Ringdal, & Kaasa, 2001; Surtees & Wainwright, 1999). In discovering their positive mutation status, even when it has been suspected previously, women are not only forced to face a potentially complicated set of risks and decisions, they may also experience a re-emergence of previously resolved feelings of grief related to the illness or loss of loved ones, previous cancer experiences, etc. (Bottorff et al., 2000; Clarke, Butler, & Esplen, 2008; Hopwood et al., 1998; Lobb et al., 2004), and this merging of past and present cancer-related experiences can be particularly painful (Bonanno & Kaltman, 2001). Women may also experience a sense of loss along with the receipt of a positive mutation test result, which may be related to their sense of security and peace of mind being threatened, loss of hopefulness about the future, and/or loss of a sense of immortality. Women who receive negative mutation results for *BRCA1/2* have also reported a sense of loss in their sense of connection to a deceased family member or members, as if the intangible bond that they once shared over cancer was broken by the discovery of their non-high-risk status (Esplen et al., 2004).

Depression. Depression may take the form of sad mood, helplessness, hopelessness, and reduced energy and activity levels; when depression continues long-term, results such as drastic increase or loss of appetite, insurmountable fatigue, etc. may be present (Lerman & Croyle, 1996). While clinical levels of depression are not significantly more common among members of HBOC families than the general population, about one-third of individuals in one study experienced an acute period of sadness and crying (H. T. Lynch et al., 1997). Lodder's 2001 study indicates that levels of depression after genetic testing for *BRCA1/2* decrease for non-carriers, and increase slightly for carriers. Another study noted significantly greater reductions in depression

and role impairment in non-carriers as compared to carriers when comparing baseline scores to those at one-month post-disclosure; findings also indicate that there are differences in the frequency of clinical depression at one-month post-disclosure, with carriers demonstrating higher rates of depression than non-carriers (14% versus 8%) (Lerman et al., 1996). In longer-term follow-up, only non-carriers demonstrated significant reductions from pre-test to post-test rates of depression, while rates of depression among carriers actually increased over six months (Lerman et al., 1998). Interestingly, one British study found a reversal of these trends regarding depression, whereby non-carriers demonstrated a significant increase in frequency of depression while carriers demonstrated a non-significant decrease (Wagner et al., 2000); a similar finding was observed in a Swedish study, although depression scores for non-carriers in that study were still within the normative range (Arver et al., 2004).

One study suggests that family experience with ovarian cancer may be significantly related to individuals' experience of depression in the post-testing phase. A notable majority of women with ovarian cancer-affected relatives report, prior to genetic testing, that they believe they will experience depression if they test positive for a mutation in *BRCA1*, and almost three quarters of such women under age 45 who underwent testing for *BRCA1* reported feeling depressed six weeks after they were tested (Lerman & Croyle, 1996). This finding may be related to the gravity of ovarian cancer and the relative frequency with which it takes the lives of those affected.

A Canadian study of 157 women at high familial risk of ovarian cancer found that 31.4% scored above the clinical depression cutoff point on the Center for Epidemiologic Studies Depression Scale (CES-D) (Radloff, 1977). Classifying participants into two

groups based on this cutoff point revealed significant differences between the two groups: those among the depressed group were “younger, currently more depressed over the death of a relative with ovarian cancer, more worried about being at increased risk of developing cancer, and more likely to have altered plans for the future [mostly about having children] as well as a result of their perceived risk level,” (Robinson et al., 1997, p. 200). The women who scored above the depression threshold were also more worried about future risk for their daughters than those in the non-depressed group (Robinson et al., 1997). These findings support the notion that young women experiencing genetic testing are particularly vulnerable.

Depression is not observed only among individuals who discover that they are mutation carriers; interestingly, increased depression was also present in 12% of non-carriers six weeks after testing in a kindred of 32 relatives tested for a *BRCA1* mutation, although these levels were not above the threshold for diagnosis (H. T. Lynch et al., 1993). As previously discussed, this psychological disturbance may be related to regret about irreversible surgical decisions made on the assumption of high-risk status, or may be related to feelings of guilt; this will be discussed further later in this appendix.

The availability of social support has been associated with decreases in depression over time among women who receive positive *BRCA1/2* test results (Esplen et al., 2004). Participation in a supportive-expressive therapy group focusing specifically on difficult issues associated with positive mutation status, such as guilt, fear, grief, etc., was associated with improvements in depression symptoms, and this improvement is thought to be largely due to the availability of social support associated with participation in the group (Esplen et al., 2004).

Anger. Anger is another relatively common response to receipt of a positive *BRCA1/2* mutation-test result (Croyle et al., 1997). Individuals may experience a sort of “why me?” reaction to the news (Bredart et al., 1998). Anger may also result from the afore-mentioned transition from uncertainty about whether or not one possesses a *BRCA1/2* mutation to uncertainty about whether and when cancer will develop, as an element of hope is dashed in discovering that one has not escaped the mutation (H. T. Lynch et al., 1997).

Guilt. Feelings of guilt and responsibility are not uncommon among high-risk cancer families to begin with (Brody & Biesecker, 1998), and the testing process and identification of a mutation in additional family members can bring these feelings to the surface (Croyle et al., 1997; H. T. Lynch et al., 1997). Guilt reactions are likely to take different forms depending on the outcome of a genetic test. In those found to be mutation carriers, feelings of guilt may be related to having passed a *BRCA1/2* gene on to one’s children, and worrying about the future health and well-being (Bredart et al., 1998; Lerman & Croyle, 1996). Individuals who receive negative mutation test results may experience “survivor guilt,” comprised of potentially invasive thoughts and questions about why they should be spared from the experience of carrying the mutation when their loved ones are enduring such stress (Lerman & Croyle, 1996). Researchers have also noted that this guilt manifested after the receipt of test results may play a role in reproductive decision-making (Frets, Verhage, & Niermeijer, 1991), discussed in further detail in a subsequent section.

Functional health status. When the results of a genetic test for *BRCA1/2* are at the forefront of one’s mind to the extent that they limit her ability to function normally, an

individual may experience changes in “role functioning, sexual functioning, social functioning, and sleep,” (Lerman & Croyle, 1996, p. 194). This type of disturbance has been observed in as many as 30% of women at risk of breast cancer, and is attributed to prominent concerns about personal risk status (Kash et al., 1992).

Family stress. The potential for psychological and emotional disturbance as an outcome of genetic testing is not limited to the individual; stress may also be experienced at the couple and family levels. Higher than average levels of cancer-related stress and anxiety are likely to be present in these families even before testing as the members struggle with experiences of breast and ovarian cancer over time (Brody & Biesecker, 1998). In the midst of dealing with those stressors, learning about *BRCA1/2* mutations in one’s family may necessitate new levels and types of communication, perhaps between extended family members who are typically not in touch; this may bring about conflict and confrontation, as relatives attempt to integrate new information and engage in collaborative information gathering and decision-making. This may be complicated by, for example, a lack of consensus regarding who should be informed about the presence of a mutation, or who should undergo testing and when. This new dialogue also has the potential to bring together family members who were previously disengaged (Bottorff et al., 2000). Families may experience an increase in conflict and/or a decrease in cohesion as potential disagreements arise over testing decisions (Brody & Biesecker, 1998) and communicating results to younger family members, individuals and entities outside the family, etc. (Bottorff et al., 2000).

Individuals within families may also vary in their experience of psychological distress relative to other family members, especially when there is variability in test

outcomes; for example, one sibling who tests positive among a group of siblings who test negative may experience more psychological distress related to feelings of isolation, or one who tests negative in a group of positives may experience high levels of survivor's guilt (Esplen et al., 2004). A study specifically investigating the impact of sibling test result on reactions to *BRCA1* test results found that

men who received their [positive] results first and noncarrier men whose siblings all tested positive experienced adverse short-term psychologic reactions...carrier women experience elevated distress shortly after receiving their results...[and] women who learned their positive carrier status first report the highest levels of distress, (A. W. Smith et al., 2008, p. 389).

These findings represent important test result/family context interactions, with significant differences related to gender. Relative to other hereditary cancer syndromes, HBOC may present more gender-specific issues and bring the issue of gender to the forefront of family relationships, since women are primarily affected by the increased risk (Pasacreta, 2003). Existing gender issues within families may be magnified by the presence of a *BRCA1/2* mutation.

Researchers have noted that the experiences of genetic testing and result disclosure do not take place in isolation, but rather occur “within the context of the cancer histories, testing experiences and results of [one’s] relatives. Individual reactions to learning a relative’s mutation status may bear a relationship to the psychological sequelae associated with [one’s] own testing,” (Pasacreta, 2003, p. 617).

Family communication. Once an individual has completed the genetic counseling and testing process, it is not atypical for him or her to take some time to come to terms with his/her own risk before taking on the challenge of making decisions about communicating mutation information to other family members (Forrest et al., 2003);

research indicates that carriers wait an average of four months before disclosing information about their carrier status to adolescent and adult offspring (Patenaude et al., 2006), and typically view the ages of 19-25 as the ideal time to inform offspring about their carrier status (Segal et al., 2004). The burden of this task varies with the number of other tested family members, the novelty of the issue of HBOC within the family, and the general level of knowledge about *BRCA1/2*-related issues among family members. Furthermore, family roles, traditions, and dynamics will determine the extent to which one is expected and/or allowed to share information or may choose to keep it private (Forrest et al., 2003; Foster et al., 2004; J. Green et al., 1997).

Researchers have noted various ways in which family context shapes the manner in which a family communicates about cancer. Individuals' decisions to pursue genetic testing in light of new information can "destabilize family expectations and norms with other members feeling pulled in different directions, in terms of his/her own needs [and] the family's style of coping," (Clarke et al., 2008). The task of communicating genetic information to potentially affected members of a family may serve to bring together previously disengaged relatives, as genetically-linked but emotionally distant siblings, cousins, etc., trade information about the presence of disease in order to "more [precisely] estimate...one's own risk," (Forrest et al., 2003, p. 323). In contrast, decisions regarding testing can incite new conflict in between involved family members, adding relationship stress to an already stressful situation (McDaniel, 2005). Dynamics around family communication are complicated by the fact that genetic information about one member of a family has implications for multiple other family members, especially when the family is dealing with a first member to be tested; for example, if an individual who is the first

tested among her/his family is identified as a mutation carrier, a parent is automatically implicated as a carrier, and all of the individual's siblings and children are suddenly at 50% risk of also carrying mutations (Costalas et al., 2003). Therefore, those making decisions about genetic testing and information cannot ever act truly independently, as genetic information is inherently also family information.

In general, a large proportion of individuals who undergo testing for *BRCA1/2* disclose their results to first-degree relatives within the weeks and months immediately following their own receipt of results. A 2006 study of 273 women who had undergone testing for *BRCA1/2* in Boston revealed that those who received positive results disclosed to their mothers at a rate of 92% within four months, and at a rate of 81% to fathers within the same period; factors impacting disclosure of mutation status to parents included participant age (with younger women more likely to disclose) and father's cancer status. Comparable rates of disclosure were identified for sisters (90%), brothers (78%), daughters 18 or older (97%), and sons 18 or older (90%) (Patenaude et al., 2008). Brothers were more likely to be informed if the *BRCA1/2* mutation had been inherited by the participant from the father. Not surprisingly, telling children was related to children's age (with adult children more likely to be informed than pre-adolescents) and gender (with daughters more likely to be informed than sons).

Prior research indicates that individuals undergoing genetic testing for *BRCA1/2* have more trouble communicating a positive result than a negative one, likely due to higher levels of distress for both the speaker and listener during such an interaction (Costalas et al., 2003). Carriers also experience more difficulty in explaining mutation results to adult siblings than to adult children; this may reflect a cohort effect, in that

adult children would tend to be younger and more fluent in discussing issues related to genetics due to increased accessibility of genetic information in the form of TV, internet, and other outlets. In addition, women are more likely to be told about the presence of a *BRCA1/2* mutation in the family than are men, likely because they are at much greater personal risk of developing cancer than are men and because it may be more difficult to explain issues related to breast/ovarian health to male relatives (Costalas et al., 2003; Julian-Reynier et al., 2003).

A recent study of the process of disclosure of positive *BRCA1/2* mutation status to offspring highlighted some of the pertinent issues of the proposed study (Clarke et al., 2008). Women expressed unique concerns related to disclosing to young adult daughters, especially with regard to the possibility that daughters may face discrimination “including securing adequate insurance and finding a life partner. The concern that the daughter might be viewed as ‘damaged goods’” (Clarke et al., 2008, p. 799) was also described. Also present in this group were concerns about balancing a daughter’s right to know about her risk with a desire to protect her from difficult information and the unhappiness that might come along with it. Some women described a process whereby they attempted to gauge whether or not their offspring even *wanted* to know about the mutation, based on offspring’s words and actions over time (e.g., not following up on own genetic testing). They experienced discomfort and felt dishonest when they chose to withhold or delay information (even though this was based on positive intentions), and feared that they might disclose inadvertently, which might have a negative impact. Ultimately, mothers’ decisions about informing their daughters of their own carrier status were influenced by their perceptions that their daughters would be capable of managing

the psychological issues that might accompany the news (Clarke et al., 2008). The experience of disclosure for women in this study was often highly emotional, though some participants described it instead as a straightforward conveyance of honest information relatively free of emotionality, sometimes purposefully so as to communicate that the presence of the mutation was not to be interpreted by the daughter as a big deal. Perhaps most interestingly, women in the study described instances of being “caught off guard” by children’s questions, and as a result giving their children inaccurate information because they were not prepared to deliver the truth. It is interesting to consider, in these cases, how future communication about the mutation and its presence within the families will be received by daughters who realize that they were previously misled. Finally, some women in the study described a sense that by disclosing to their children, they were relinquishing control over how the genetic information should be managed; they told of struggles reconciling themselves to their children’s decisions when children chose paths that were not what the mothers would have desired, and to ongoing fears about the potential consequences of test results on their children (Clarke et al., 2008). Certainly, the real and varied concerns and experiences of women in the Clarke *et al.* study shed light on how issues family issues may impact the process of disclosure of positive *BRCA1/2* mutation status by mothers to their young adult daughters, changing the relational context in which women in the population of interest deal with their own testing and risk management processes.

A study of individuals in HBOC and Huntington’s Disease (HD) families in Denmark indicates that individuals often find the presence of a health professional (i.e., genetic counselor, physician, etc.) helpful during initial communication of information

about a mutation to other family members, because the professional can help reduce the uncertainty associated with risk information and answer family members' questions that the tested individual may not be equipped to answer (Forrest et al., 2003). They also observed a phenomenon whereby older individuals, especially females, within a family were perceived as responsible for communicating risk information to members of younger generations, even if the younger generation were adults; this is in keeping with other research on mutation disclosure (Clarke et al., 2008). There are both research-based and anecdotal reports of frustrating experiences in which family members experience a great deal of frustration when the person or persons seen as "responsible" for and having the authority to communicate risk information to a younger member fails to do so and, for example, "participants felt that they could neither pass on information to relatives who needed to know...nor persuade those with authority...to do so," (Forrest et al., 2003, p. 321).

Researchers have noticed two distinct communication styles relevant to the sharing of genetic information in families. *Pragmatists* go about the business of discussing the family mutation in an active, practical fashion, while *prevaricators* seek opportunities within the context of daily life to bring up the subject. Consequently, it often takes much longer for prevaricators to complete the process of informing family members about a mutation, because they may wait weeks, months, even years for the "right moment" to present itself (Forrest et al., 2003). Given these two communication styles, it is easy to understand how families can encounter problems around the issue of communication; when both of these styles are present within the same family, there is likely to be disagreement regarding when, how, by whom, and to whom genetic risk

information should be communicated. The Danish study revealed that “individuals who wished to pass on information but felt they must defer to a relative with more authority...either had to keep quiet and shoulder the additional emotional burden or force the issue by disregarding authority or ‘blackmailing’ the prevaricating relative,” (Forrest et al., 2003, p. 322).

As discussed elsewhere in this paper, the ability to share genetic information with children to aid in their medical decision making is one of the most frequently cited motivations for genetic testing (Lerman et al., 1996; H. T. Lynch et al., 1997). However, the process by which this information is shared is not necessarily simple or straightforward. Information about mutations and cancer risk is often given to the younger generation at a time that is determined to be maximally helpful in terms of the receiver’s ability to take action to reduce risk and at which intervention is practical; in other words, for those in HBOC families, children might be told about the presence of a *BRCA1/2* mutation in the family during their mid-twenties, when they may realistically need to begin surveillance and prevention measures. Furthermore, most health professionals will not administer, and professional societies generally recommend against, genetic testing for mutations associated with late-onset diseases (including *BRCA1/2*), until a minor has reached at least the age of consent, as there is negligible risk during the childhood and adolescent years and the potential medical and psychosocial benefits are not applicable until adulthood (ASCO, 2003; ASHG & ACMG, 1995; Kodish, 1999; Sharpe, 1993; Tercyak, Peshkin, DeMarco et al., 2007; Wertz, Fanos, & Reilly, 1994). For women in HBOC families, preventive measures (i.e., frequent screening, chemoprevention) are generally not recommended until an at-risk female

reaches the age of 25 (Burke et al., 1997; M. B. Daly et al., 2006), although some parents have reported that they hoped lifestyle modifications during childhood (e.g., healthier diet, increased physical activity) might decrease long-term cancer risk for children in HBOC families (Tercyak, Peshkin, DeMarco et al., 2007). In addition to these factors, the sense that children are old enough to understand the information is another oft-cited factor in decision-making about when to inform children about the presence of a mutation (Forrest et al., 2003).

A study conducted at the University of Chicago Cancer Risk Clinic investigated communication about the presence of *BRCA1/2* mutations to children under the age of 25 within families by analyzing 42 qualitative interviews with carriers who had a child under the age of 25 at the time of genetic testing. Fifty-five percent of participants discussed the hereditary risk of cancer in general terms (i.e., not necessarily explicitly disclosing their own mutation status), and approximately half informed their children of their positive carrier status (Bradbury et al., 2007). This is consistent with other research suggesting that about half of mothers who undergo testing for *BRCA1/2* disclose their results to their minor children within one month after testing (Tercyak, Hughes et al., 2001; Tercyak, Peshkin, DeMarco, Brogan, & Lerman, 2002). Conversations often comprise a gradual process of disclosure, in which parents informed their children about the presence of a genetic mutation in the family and then about their personal risk and mutation status over time. Disclosure was associated with “older child age, female parent sex, parent history of [risk-reducing] surgery...and less formal parent education,” (Bradbury et al., 2007, p. 3707), while child’s sex and parent’s history of cancer were not associated with disclosure (Bradbury et al., 2007; Tercyak, Hughes et al., 2001). The authors posit that

the association between disclosure and history of risk-reducing surgery may indicate that some parents choose to disclose their mutation in an attempt to explain their surgery decisions to their children, although this finding did not stand out from among the interview data collected.

In one study of parental disclosure of *BRCA1/2* presence in families to children, disclosure to children of all ages was generally handled independently by the mutation-carrying parent or with a spouse, with a minority (about 5%) reporting that a genetic counselor had been their most important supporter during the decision-making process; however, 21% reported that a genetic counselor was involved in the decision, and 14% reported the involvement of a physician (Bradbury et al., 2007). There was a good deal of variation with regard to the length of time that parents waited before disclosing information to their children, with 30% of parents indicating that they had delayed disclosure to at least one of their children; length of delay varied from several months to six years and was chosen for the following reasons: waiting for a child to get older, giving self time to adjust to the information or to decide how to use the information personally, and waiting to share the information in person (i.e., in cases in which the child did not live with the parent). Interestingly, although the mean age of children of this set of mutation carriers who were disclosed to was close to 18, “two children younger than 10 were told of their parent’s *BRCA* mutation or the hereditary risk of cancer, one as young as 7,” (Bradbury et al., 2007, p. 3707).

When parents disclosed to children under 25, they often felt that the children did not fully comprehend the gravity of the information with which they were being presented, with older child age associated with increased parental perceptions of

understanding. Parents reported emotional responses among offspring including concern/anxiety, crying, and fear; however, parents' perceptions of children's emotional responses were not associated with offspring age or sex. Most parents reported no effect (65%) or a positive effect (22%) on the parent-child relationship as a result of disclosing mutation information, with a small number of parents reporting that their own feelings of guilt post-disclosure resulted in some tension in the parent-child relationship (Bradbury et al., 2007).

Certainly, there are situations in which an individual may decide not to share information with others, either keeping information to themselves entirely or disclosing only to certain family members. *Declining to tell* may be the result of an attempt to protect family members who are seen as vulnerable to discrimination or who may not be able to handle information about a mutation. In other cases, it may be the result of difficulty transcending physical and/or emotional distance or conflicts that existed before knowledge of the mutation was available. Finally, some individuals may decline to tell because they believe that certain family members do not "need" to know (e.g., an elderly uncle with no children may not be considered at-risk within an HBOC family) (Forrest et al., 2003). In sum, the motivations for withholding information from certain family members are varied and not necessarily negative.

Interestingly, despite this clearly varied experience in the process of telling family members about the presence of a mutation, it appears to be a moot point for some individuals. Many of the participants in the Danish communication study described having "always known" about the presence of a hereditary disease in their family, or that

they had come to realize this over time as more and more members of the family were diagnosed (Forrest et al., 2003).

Long-term implications. The body of research that follows women beyond one-year post-disclosure to assess their psychological functioning is quite small. One British qualitative study used telephone focus groups to discuss these issues with women who had known about their positive mutation status for at least two years. Participants' responses revealed that beyond the first year post-disclosure, carriers continue to experience acute bouts of

anxiety, depression, shock, surprise and feelings of neglect... triggered by risk-related events, such as being initially informed about their risk (by a health professional), receiving an appointment letter, waiting for an overdue appointment, having a mammography or biopsy and receiving the results (even if they showed no signs of breast cancer), (Appleton et al., 2000, p. 515).

The group also described a sense of being very sensitive to breast cancer cues, such as detecting changes in their own breast tissue, hearing a story about breast cancer in the media, or learning of a new diagnosis within their families. In response to these cues, they engaged in behaviors such as increased frequency of breast self exam, seeking out or taking greater interest in media reports, feeling anxious, or purposely trying to avoid the cues. The women described an ongoing cycle of emotional and cognitive fluctuations that coincide with their annual screening cycle and that is relatively consistent from year to year (Appleton et al., 2000). This is in keeping with previous research on women with familial risk of breast cancer who have reported that they experience distress just prior to routine mammography, and that this distress is relieved by a normal result (Valdimarsdottir et al., 1995).

The need for genetic counseling. In communicating about risk to patients in a healthcare system, it is widely acknowledged that the manner in which risk information is framed has significant bearing on how it is heard and the actions that will arise from it (Sarfati, Howden-Chapman, Woodward, & Salmond, 1998). Many researchers and practitioners have noted that the provision of appropriate, competent, in-person genetic counseling before, during, and after genetic testing and education may effectively serve to mitigate the risk of adverse psychological reactions (Cull et al., 1999; Lerman, Daly et al., 1994; Lerman et al., 1996; Offit & Brown, 1994). This includes becoming fully informed about the benefits and limitations of receiving genetic testing results, including the current limitations on options for prevention and detection of cancer (Geller et al., 1997; Lerman & Croyle, 1996). This is further complicated by the ever-changing nature of genetic risk information, and health professionals and patients alike must constantly sift through new information to fully understand the current knowledge about risk (Bottorff et al., 2000). As a result of these factors, even when high-quality services are provided, there can be short-term negative effects on psychological functioning as a result of a positive *BRCA1/2* mutation test result (Croyle et al., 1997).

What is also important in the context of genetic counseling about hereditary cancer risk is the appropriate inclusion of family members or, at minimum, a discussion with the patient of whether and how other members of the family system will be informed of their risk. Healthcare professionals must recognize the psychosocial support needs of all members of the family system, including those who test negative for a mutation and may experience guilt, confusion, worry, etc. (Bleiker & Aaronson, 2000).

Appendix C: Overview of Reproductive Options for *BRCA* Mutation Carriers

Adoption. Couples in which a partner carries a *BRCA1/2* mutation may face unique challenges related to the adoption process if they choose to pursue that method of family formation. Should they elect to adopt through a public agency, they may be met with hesitation in placing a child in a family in which the risk of future cancer is significantly higher than average, should this information be made known; this dilemma may be similar to what is faced by some cancer survivors who try to adopt (Schover, 1999). In private adoption, in which birth mothers are often allowed to personally select adoptive parents, it has been suggested that birth mothers may be deterred by a personal and/or family history of cancer, regardless of the medical facts or what measures are being taken to reduce one's own risk (Schover, 1999), and may be legitimately concerned about the potential for adoptive parents to develop cancer in the future, which would certainly impact care of and relationships with the adopted child.

Third-party reproduction. Third-party reproduction includes any method in which an individual outside of the couple is biologically involved in the creation or gestation of a child. Relevant options for couples in which the female partner is *BRCA1/2*-positive include traditional surrogacy (a surrogate is inseminated with the intended father's sperm), gestational surrogacy (a surrogate is implanted with an embryo to which she is genetically unrelated), oocyte (egg) donation, or some combination of these (Schover, 1999). When gametes are donated, that may be accomplished anonymously (donor and recipient never know each other's identities), through a known donor (donor and recipient agree to meet as part of the donation process and possibly beyond), or through directed donation (donor and recipient already know each other, i.e.,

a relative or friend is the donor) (Rosen, 2005). In addition to the logistical stress of using this strategy, couples who select third-party reproduction may face some societal stigma (Braverman & Corson, 1995; Cahill, 1996). They also report fears that they will have trouble bonding successfully with the child(ren) produced via these strategies, or that the child(ren) will have unexpected psychological or health problems. Further, some parents are concerned about their ability to talk openly with their child(ren) in the future about the fact that they were created through third-party reproduction, and fear that the children themselves will feel stigmatized by this knowledge (Rosen, 2005). Third-party reproduction is still the method of choice for some of these families for several reasons. One partner, or perhaps both, will still have a genetic relationship with the child. Parents are able to exert some control over the selection of the donor (as opposed to the level of control possible in the case of adoption) and over their child's prenatal care. Finally, parents are often able to choose this option and keep it a secret from their child (should they choose to do so), which is not always possible with adoption (Rosen, 2005). Use of this option also presents psychological and emotional challenges for mutation-positive women, who must come to terms with what may be a loss of a lifelong desire to accomplish pregnancy, childbirth, and breastfeeding (Schover, 1999).

Reproductive technology. Currently, a limited number of interventions are available to reduce the risk that a *BRCA1/2* mutation carrier who wishes to carry her own biological pregnancy will pass her mutation to offspring; specifically, parents may utilize Pre-implantation Genetic Diagnosis (PGD) or Prenatal Genetic Diagnosis (PND).

Pre-implantation genetic diagnosis. The use of PGD by families with *BRCA1/2* mutations is feasible, but as yet not widely available. In this procedure, egg and sperm

cells are extracted from parents and brought together in a laboratory setting through a process called in vitro fertilization (IVF). When fertilized embryos reach 8 cells (approximately 3 days after fertilization), one cell from each viable embryo is extracted and tested for the mutation of interest. Only embryos that test negative for the mutation are then selected and implanted, resulting in a child (or children) that is (or are) mutation-free. Mutation-positive and any other unused embryos are either stored, destroyed, or donated to research. PGD was developed to prevent pregnancies affected with serious, life-threatening genetic diseases, and has been used more widely for families with serious (often fatal), high-penetrance recessive (e.g., Cystic Fibrosis), dominant (e.g., Huntington Disease), and X-linked disorders (e.g., Fragile X). Its use in the setting of cancer predisposition syndromes has thus far been rather limited; a 2006 literature review identified only 55 cases of PGD and PND combined in this context, taking place at only 13 centers; it has not yet become common practice (Offit et al., 2006) and is not universally available to couples in which one partner is *BRCA1/2*-positive.

One major advantage of using PGD as opposed to other techniques, such as amniocentesis or PND (which can test for a mutation after pregnancy has been achieved naturally) is that it does not require parents to consider abortion in the case of a positive mutation test (Harmon, 2006). Another advantage is that, in cases where infertility is also an issue and IVF is needed regardless of the use of PGD, the cost of IVF treatment may be covered by insurance (Simpson, Carson, & Cisneros, 2005), which can partially mitigate the very high total cost of this procedure.

The availability of PGD for testing for *BRCA1/2* has come to the forefront of late, largely because the current cohort of interested women are the first generation to reach

reproductive age after their mothers developed cancer and were identified as mutation-positive (Harmon, 2006). In light of this increased demand, some physicians and researchers have argued in favor of making PGD for late-onset disorders more widely available, arguing that availability would have a positive effect on mutation carriers of childbearing age because it can reduce fatalistic attitudes and make individuals more optimistic about starting a family (Harmon, 2006). In fact, some mutation carriers who proceeded with natural conception on the advice of physicians who did not recommend the use of PGD have reported feeling angry that they were not given the option to use available medical technology to reduce their children's cancer risk; some speculate that physicians' personal values stop them from informing patients about PGD, even when it may be desirable to the patient and allow them to stop a family legacy of illness and death by not passing the harmful mutation to their children (Harmon, 2006). Other mutation carriers take offense at the idea that new children are only desirable or selected for family membership if they "pass a test" by not carrying a *BRCA1/2* mutation, implying that they themselves and others like them would not have existed had the test been utilized by their parents (Harmon, 2006).

Since the use of PGD for BRCA mutation carriers was approved in the UK in 2006, there has been some new research regarding the attitudes of patients about the procedure. One study found that 75% of the 53 mutation-positive women surveyed believed that it was acceptable to offer PGD to prevent the transmission of *BRCA1/2* mutations. Regarding personal use, 37.5% of respondents who had already completed their families stated that they would have considered using PGD had it been available to them previously; only 14% of women contemplating a future pregnancy stated that they

would consider using PGD. Many of the women who stated that they would not personally consider using PGD to prevent having a child with a *BRCA1/2* mutation wrote emphatically about their opinions, citing the value of their own lives, the fact that a *BRCA1/2* mutation is not a death sentence, and fear about the future of scientific technology. As a group, the women viewed PGD as more acceptable than PND (discussed later in this appendix), but expressed concerns about undergoing IVF to accomplish PGD, about the risk of misdiagnosis, about the low success rate associated with IVF, and about the inconvenience of going through the procedure (Menon et al., 2007).

Although we did not ask specifically about the use of reproductive technology in our pilot study interviews, the topic did come up spontaneously in conversations with some of our participants. Our data suggest that some carriers would be open to considering this procedure, but only if IVF were warranted for a separate reason and PGD could be accomplished as a sort of “add-on” procedure; in most cases, natural conception was still preferred (unpublished data).

Prenatal genetic diagnosis. The other interventional option for couples in which one partner carries a *BRCA1/2* mutation is to utilize prenatal genetic diagnosis (PND). In this process, cells from a growing fetus are extracted during the pregnancy, via amniocentesis or chorionic villus sampling (CVS) (Offit et al., 2006). Amniocentesis is the collection and analysis of amniotic fluid, and typically occurs at 16 to 20 weeks gestation. CVS is the removal and analysis of a small piece of the placenta and is usually performed at 10 to 12 weeks gestation. Both procedures pose some level of risk to both the mother and the developing fetus and are significantly uncomfortable for the mother

(Offit et al., 2006). Extracted materials are tested for the mutation; a positive test indicates that the child carries the mutation. Couples then have an option regarding continuation or termination of the pregnancy if they have a strong desire to avoid bearing children who are *BRCA1/2* mutation carriers. Research suggests that rates of termination in the case of an identification of an abnormality are low; rather, PND is often used to reassure parents that their baby will not carry a mutation, or to prepare them for future health concerns if the baby does carry the mutation. Still, it is certainly plausible that some patients will choose to terminate pregnancy upon receiving a positive prenatal genetic test for *BRCA1/2*.

A small number of studies have investigated individuals' attitudes regarding termination of pregnancy based on the presence of *BRCA1/2* mutations discovered via PND. A Dutch study asked 78 subjects, all members of families with identified *BRCA1/2* mutations, if they found termination of a pregnancy for this reason acceptable for themselves, for female and male fetuses. Responses of mutation-positive and mutation-negative individuals were compared. Among carriers, none found this option acceptable, regardless of the sex of the fetus; among non-carriers, a minority found termination acceptable for female (14%) and male (10%) fetuses. The authors posit that

...the stronger reluctance in mutation carriers than in non-mutation carriers towards terminating a pregnancy of a mutation carrier boy or girl may have several reasons. Firstly, mutation carriers may be more acutely aware of the burdensome emotional implications of terminating a pregnancy because of *BRCA1/BRCA2* carriership than non-mutation carriers. Secondly, they may perceive terminating the pregnancy of a mutation carrier child as incompatible with their own existence (Lodder et al., 2000, p. 883).

Similar findings were reported in a British study of adults with another hereditary cancer syndrome, familial adenomatous polyposis (FAP), which predisposes carriers to

colorectal cancer . While almost all (93%) believed that prenatal diagnosis should be available to affected families, a smaller percentage (64%) stated that they would personally use it, and only 24% would consider terminating an affected pregnancy (Whitelaw, Northover, & Hodgson, 1996).

In clinical practice, demand for PND for *BRCA1/2* mutations has been low and is expected to remain so for the foreseeable future, largely because the diseases associated with these mutations are survivable and carriers experience decades of healthy life before the onset of disease, and because of the heavy emotional burden associated with making a decision about termination or continuation of an existing pregnancy (Lodder et al., 2000).

Legal and ethical issues. Laws and policies regarding the use these reproductive technologies to test for susceptibility to late-onset, incompletely penetrant conditions vary from country to country. For example, in the UK, the use of PGD for late-onset, low-penetrance genes like BRCA was approved in 2006 (Menon et al., 2007), while in Italy it has been completely banned (Harmon, 2006). US legislators have been surprisingly silent about the use of PGD for detecting genetic mutations such as these, having created no regulations thus far; hence, PGD is being used for this purpose at several facilities in major US cities (Harmon, 2006). The Code of Medical Ethics of the American Medical Association (AMA) states that the use of PGD is acceptable in some cases, but warns that “selection to avoid a genetic disease may not always be appropriate, depending on factors such as the severity of the disease, the probability of its occurrence, the age at onset, and the time of gestation at which selection would occur,” (American Medical Association Council on Ethical and Judicial Affairs, 1994). This sentiment is echoed by the American Society of Reproductive Medicine and by medical ethics

societies in Europe (Offit et al., 2006). It seems clear that the use of preimplantation genetic diagnosis and prenatal diagnosis could easily be seen by some as inappropriate under the terms of these statements.

Using PGD or PND to prevent the transmission of *BRCA1/2* mutations raises a host of ethical issues. Most basically, there is the question of whether aborting a fetus or discarding an embryo due to the presence of a *BRCA1/2* mutation is acceptable. This is both a broad ethical question and an individual moral one. Given that hereditary breast and ovarian cancer are generally late-onset, that the genes are incompletely penetrant, and that treatments are improving with time, there is certainly a significant chance that as a result of the use of PGD/PND, fetuses will be aborted or embryos discarded that may have never developed cancer, or for whom highly effective treatment would have been available by the time they reached the age of risk (Lancaster, Wiseman, & Berchuck, 1996; Schover, 1999). At the other end of the spectrum is a question about “whether parents who give birth to a genetically damaged child, when they had the chance to use prenatal diagnosis, will be stigmatized or even denied health insurance benefits [in the future]” (Schover, 1999, p. 56). In a world where genetic technology is advancing faster than physicians, patients, and society at large can keep up with it, we must question whether the specter of eugenics might re-arise and result in a societal stigma against those born with known genetic mutations (Schover, 1999). Finally, at the family level, what are the implications for children who may be born with known mutations – might they be treated differently by parents who are aware of this risk (Schover, 1999)?

Class issues are also related. We must recognize that, at least currently, these procedures are only available to those who can afford to pay for them out-of-pocket,

since insurance companies currently will not cover the use of PGD or PND for the purpose of *BRCA1/2* (Schover, 1999). In the long term, this could result in the “breeding out” of genetic disease among the affluent while the less affluent remain at the mercy of chance, resulting in increasing class division and health disparities.

As previously discussed, for couples who elect or are advised to use PND rather than PGD, receiving a positive result for a prenatal BRCA test leads to a choice regarding abortion, and some argue that this is an inappropriate use of medical knowledge and technology (Simpson et al., 2005; Surbone, 2001). In addition, some have expressed concerns regarding whether or not genetic counseling can be appropriately value-neutral and non-directive in counselors’ interactions with pregnant *BRCA*-positive women so as to allow them to make their own independent decisions regarding their pregnancies (Surbone, 2001). In its most pernicious form, forcefully communicated negative opinions about carrying a *BRCA*-positive pregnancy to term can take the form of eugenics and may come about as a product of movement toward patient “autonomy” or economic pressure within the healthcare system (Sherwin, 1992; Surbone, 2001). The availability of high-quality, value-neutral counseling from both genetic counselors and physicians is essential in assuring that patients are both informed about all of their options and given the autonomy to make their decisions independently (Rhodes, 1988).

At the same time, there are several reasons that *BRCA1/2*-positive women may be particularly motivated to carry and deliver their own biological children. Both parity and breastfeeding are known protective factors for ovarian cancer risk in the general population (Modan et al., 2001; Rosenblatt & Thomas, 1996). Breastfeeding has been associated with reduction in breast cancer risk of *BRCA1* carriers, but not *BRCA2* carriers

(McLaughlin et al., 2007), and parity has been identified as protective for both breast and ovarian cancer in *BRCA1* carriers and for breast cancer in *BRCA2* carriers, but as a risk factor for ovarian cancer in *BRCA2* carriers (McLaughlin et al., 2007). Other studies have reported that parity increases the risk of both types of cancer in *BRCA2* carriers (Narod et al., 1998). Women who seek out information about how to influence their cancer risk may not always distinguish between findings that apply to the general population and those that apply to mutation carriers, and may believe that “recommendations” apply to them that actually are intended for a different group. These findings, in combination with recommendations from physicians and possible pressure from loved ones to proceed with oophorectomy before age 40, may result in women who are aware before they commence childbearing that they carry a mutation in *BRCA1/2* feeling a sense of pressure with regard to reproduction. In fact, our 2008 qualitative study yielded several quotes from young female *BRCA1/2* mutation carriers in this vein, such as “we talked about having children much sooner, just based on things you read, that it’s better for me to have children earlier rather than wait,” (unpublished data).

Appendix D: FORCE Website Study Announcement

NCI RESEARCHER SEEKING YOUNG FEMALE *BRCA1/2* CARRIERS FOR INTERVIEW STUDY

Researchers in the Clinical Genetics Branch (CGB) at the National Cancer Institute, in collaboration with the University of Maryland School of Public Health, are conducting an interview study of women who are aware of their positive *BRCA1/2* mutation status during young adulthood. Participants will be asked to take part in a 1-2 hour recorded telephone interview, during which they will be asked to discuss the following:

- Personal and family history of breast/ovarian cancer;
- Experience with genetic counseling and disclosure;
- Previous, current, and/or future couple relationships;
- Family formation; and
- Previous, current and planned strategies for risk management/reduction

In order to participate, women must meet all of the following criteria:

- Currently aged 18-35
- Tested positive for *BRCA1* or *BRCA2*
- Able to speak and understand English fluently
- Have previously, are currently, or will at some point in the future contemplate a couple relationship, family formation, and/or risk management/reduction

Interested individuals are invited to contact Lindsey M. Hoskins, MS, LGMFT via telephone (301-451-9732) or e-mail (hoskinsl@mail.nih.gov). A brief telephone screening interview (less than 10 minutes in length) will be required to ascertain whether eligibility criteria are met. If eligible, participants will be required to read, sign, and return NCI Informed Consent documents prior to participating in the interview.

Participants and non-participants alike will have an opportunity to take part in a young *BRCA1/2*-carrier focus group at the 2009 Joining FORCEs Conference, to be held May 15-16, 2009 in Orlando, FL.

Appendix E: Telephone Screening Interview

TELEPHONE INTERVIEW FOR POTENTIAL PARTICIPANTS

Hello, my name is Lindsey Hoskins and I am a pre-doctoral researcher at the National Cancer Institute. I received your contact information via [FORCE, the Breast Imaging study, the HBOC study, your e-mail/phone call] and would like to speak with you about participating in my interview study. Do you have a few minutes to talk to me and answer some brief screening questions?

Great. I'm conducting a study of women like you, who have received positive *BRCA1/2* mutation test results during their twenties and thirties. I'm interested in finding out how that event and that knowledge has shaped other parts of your life, specifically couple relationships, having children, and your current and future strategies for managing your cancer risk. If you choose to participate, you would be asked to participate in a telephone interview which will probably last between one and two hours. Does that sound like something you might be interested in and willing to do?

- Yes
- No

If YES:

Great. Well, in order to figure out if you are eligible to participate in the study, I'd like to ask you a few questions. They are just some basic pieces of information about you, what your life was like at the time you had genetic testing, and what's going on for you now. It should take less than five minutes. Do you have any questions for me before we get started?

If NO:

Participation in this study is strictly voluntary. I have no wish to pressure you into doing something in which you are not interested, but are there any questions I might answer for you to be certain you understand what it is we are requesting of you?

If YES: Answer the questions

If NO: Thanks very much for your time. [If speaking to an HBOC or BI participant] Your decision in no way affects your overall participation in the NCI hereditary breast/ovarian cancer study. Please feel free to contact us at any time if we can be of assistance to you.

1. What is your date of birth? ___/___/_____
2. When did you first learn about genetic testing for BRCA mutations? (an approximate date is fine) ___/___/_____
3. How old were you when you underwent genetic testing for BRCA? _____
4. What was the approximate date of your BRCA mutation test disclosure?
 ___/___/_____
 - a. Researcher calculate age at disclosure = ___ years
5. Did you receive a positive (you do have the mutation) or negative (you do not have the mutation) test result? Positive Negative
 - a. If positive, which mutation do you carry?
 - BRCA1
 - BRCA2
6. What was your relationship status at the time you were tested for the BRCA mutation?
 - Unmarried, not in a relationship
 - Unmarried, in a casual relationship
 - Unmarried, in a committed/serious relationship
 - Unmarried, cohabiting with a partner
 - Engaged
 - Married
 - Married, living apart
 - Married, legally separated
 - Divorced
 - Widowed
 - Other (specify): _____
7. What is your relationship status now?
 - Unmarried, not in a relationship
 - Unmarried, in a casual relationship
 - Unmarried, in a committed/serious relationship
 - Unmarried, cohabiting with a partner
 - Engaged
 - Married
 - Married, living apart
 - Married, legally separated
 - Divorced
 - Widowed
 - Other (specify): _____

8. Do you currently have children?

- Yes
- No

9. Are you considering or planning to have (more) children in the future?

- Yes
- No

10. What is your current strategy for managing/reducing your breast/ovarian cancer risk?
(check all that apply)

- Surveillance
 - Mammography
 - Breast MRI
 - Breast Self-Exams (BSEs)
 - Clinical Breast Exams
 - Transvaginal Ultrasound (TVU)
 - Pelvic Exams
 - CA-125 test
 - Other (specify): _____
- Chemoprevention
 - Oral contraceptive pills
 - Selective estrogen receptor modulator (SERM)
 - Other (specify): _____
- Risk-reducing surgery
 - Risk-reducing mastectomy
 - Risk-reducing oophorectomy/salpingo-oophorectomy
 - Other (specify)
- I am not currently doing anything to manage or reduce my cancer risk

11. What other cancer risk management/reduction strategies have you considered or might you consider in the future?

- Surveillance
 - Mammography
 - Breast MRI
 - Breast Self-Exams (BSEs)
 - Clinical Breast Exams
 - Transvaginal Ultrasound (TVU)
 - Pelvic Exams
 - CA-125 test
 - Other (specify): _____
- Chemoprevention
 - Oral contraceptive pills
 - Selective estrogen receptor modulator (SERM)
 - Other (specify): _____
- Risk-reducing surgery

- Risk-reducing mastectomy
- Risk-reducing oophorectomy/salpingo-oophorectomy
- Other (specify)
- I have not considered any risk management/reduction strategies

Great – that does it for my questions. Do you have any questions for me about the study?

Wonderful. Well, based on the answers you have provided, it seems that you [are / are not] eligible for the study.

If ELIGIBLE: The next step is for you to review, sign, and return a set of documents providing informed consent for the interview. Once I have those from you, we can schedule a time for our interview. Would you please provide the best mailing address for me to send you the documents?

Address: _____

Wonderful, thank you. Please expect that package within about a week. If you have any questions about the materials, please do not hesitate to contact me by phone (301-451-9732) or e-mail (hoskinsl@mail.nih.gov). I'm very much looking forward to our next conversation! Thanks so much for your time.

If INELIGIBLE: Although you're not eligible for this particular study, I would love to keep your contact information on hand for participation in future studies for which you might be eligible. Would that be alright with you?

- Yes
- No

Thanks so much for your time.

Appendix F: Interview Informed Consent Form

INSTITUTE: NCI

PRINCIPAL INVESTIGATOR: Mark H. Greene, MD
Chief, Clinical Genetics Branch

LEAD INVESTIGATOR: Lindsey M. Hoskins, MS, LGMFT
Pre-Doctoral Fellow, Clinical Genetics
Branch

STUDY TITLE: A Qualitative Exploration of the Impact of Positive *BRCA1/2* Mutation Status on the Lives of Women During Young Adulthood

INTRODUCTION

We invite you to take part in a research study at the National Institutes of Health (NIH).

First, we want you to know that:

Taking part in NIH research is entirely voluntary.

You may choose not to take part, or you may withdraw from the study at any time. In either case, you will not lose any benefits to which you are otherwise entitled. However, to receive care at the NIH, you must be taking part in a study or be under evaluation for study participation.

You may receive no benefit from taking part. The research may give us knowledge that may help people in the future.

Second, some people have personal, religious or ethical beliefs that may limit the kinds of medical or research treatments they would want to receive (such as blood transfusions). If you have such beliefs, please discuss them with your NIH doctors or research team before you agree to the study.

Now we will describe this research study. Before you decide to take part, please take as much time as you need to ask any questions and discuss this study with anyone at NIH, or with family, friends or your personal physician or other health professional.

DESCRIPTION OF RESEARCH STUDY

This is a qualitative investigation, a type of research aimed at increasing understanding of important questions within a group of individuals by studying a few of its members in depth. Please take the time you need to make your decision about taking part. You may discuss your decision with your friends and family. You can also discuss it with your health care team. If you have any questions at any point, you can ask the investigator for more explanation.

You are being asked to participate in this study because you have been identified as a carrier of a *BRCA1* or *BRCA2* mutation who learned of her mutation status prior to age 35.

The purpose of this study is to explore the impact of receiving a positive BRCA mutation test result on young women's lives and decisions, specifically with regard to couple relationships, family formation, and risk management/reduction. We want to find out how things like couple communication and relationship development, reproductive decision-making, and planning for risk management now and in the future might be altered when women learn at a young age that they carry a BRCA mutation. This research is being done as an extension of an earlier pilot, which focused primarily on couple relationships.

resurface. Therefore, there is a chance that you may experience emotional discomfort or anxiety related to the topics discussed in the interview.

There is no risk of physical injury associated with participating in this study.

There is no financial cost associated with taking part in this study. When long-distance phone calls need to be made in order to conduct interviews, the researcher will place the telephone call to you.

RESEARCH SUBJECT'S RIGHTS

Taking part in this study is your choice. You may choose either to take part or not to take part in the study. If you decide to take part in this study, you may leave the study at any time. No matter what decision you make, there will be no penalty to you and you will not lose any of your medical benefits. Leaving the study will not affect your medical care, your ability to continue as a participant in the Breast Imaging or HBOC studies, or your ability to join future research studies conducted by the Clinical Genetics Branch. You can still get your research-related medical care from our institution.

We will tell you about new information or changes in the study that may affect your health or your willingness to continue in the study.

All information you provide will be kept confidential. Because of the qualitative nature of this study, the findings will likely be published using quotations from participants that effectively illustrate emergent themes of the study. However, all study participants will be referred to only by pseudonyms, so true identities will not be revealed. All information collected from you will be kept in either locked drawers or in password-protected computer files. Access to this information will be restricted to only the researchers involved in this study.

ACCESS TO RESEARCH INFORMATION

You can talk to the study investigator about any questions or concerns you have about this study. Contact Lindsey Hoskins, MS, LGMFT at (301) 451-9732 or hoskinsl@mail.nih.gov.

For questions about your rights while taking part in this study, call the NIH Clinical Center Patient Representative at (301) 496-2626.

Certificate of Confidentiality

This study is covered by a Certificate of Confidentiality issued by the National Cancer Institute on behalf of the Secretary of the Department of Health and Human Services. This Certificate provides additional legal protection against involuntary release of information about you collected during the course of this study that may identify you individually. Of course, such information can be released if you or your guardian requests it in writing. A Certificate of Confidentiality should prevent researchers involved in this project, to the full extent permitted by the Courts, from being forced to disclose your identity or any information about you collected in this study in any legal proceedings at the Federal, State, or local level, regardless of whether they are criminal, administrative, or legislative proceedings. However, this Certificate does not prevent the review of your research records under some legally required circumstances (for example, under the Federal Food, Drug, and Cosmetic Act or during the course of an internal program audit or evaluation).

OTHER PERTINENT INFORMATION

1. Confidentiality. When results of an NIH research study are reported in medical journals or at scientific meetings, the people who take part are not named and identified. In most cases, the NIH will not release any information about your research involvement without your written permission. However, if you sign a release of information form, for example, for an insurance company, the NIH will give the

insurance company information from your medical record. This information might affect (either favorably or unfavorably) the willingness of the insurance company to sell you insurance.

The Federal Privacy Act protects the confidentiality of your NIH medical records. However, you should know that the Act allows release of some information from your medical record without your permission, for example, if it is required by the Food and Drug Administration (FDA), members of Congress, law enforcement officials, or other authorized people.

2. Policy Regarding Research-Related Injuries. The Clinical Center will provide short-term medical care for any injury resulting from your participation in research here. In general, no long-term medical care or financial compensation for research-related injuries will be provided by the National Institutes of Health, the Clinical Center, or the Federal Government. However, you have the right to pursue legal remedy if you believe that your injury justifies such action.

3. Payments. The amount of payment to research volunteers is guided by the National Institutes of Health policies. In general, patients are not paid for taking part in research studies at the National Institutes of Health.

4. Problems or Questions. If you have any problems or questions about this study, or about your rights as a research participant, or about any research-related injury, contact the Principal Investigator, Mark H. Greene, M.D., at 301-594-7642; Executive Plaza South, Room EPS 7032, Telephone: 301-435-8062. You may also contact the Lead Investigator, Lindsey M. Hoskins, MS, LGMFT, at 301-451-9732.

You may also call the Clinical Center Patient Representative at 301-496-2626.

5. Consent Document. Please keep a copy of this document in case you want to read it again.

COMPLETE APPROPRIATE ITEM(S) BELOW:			
<p>A. Adult Patient's Consent I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby consent to take part in this study.</p> <p>_____ Signature of Adult Patient/Legal Representative Date</p>		<p>B. Parent's Permission for Minor Patient. I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby give permission for my child to take part in this study. (Attach NIH 2514-2, Minor's Assent, if applicable.)</p> <p>_____ Signature of Parent(s)/Guardian Date</p>	
<p>C. Child's Verbal Assent (If Applicable) The information in the above consent was described to my child and my child agrees to participate in the study.</p> <p>_____ Signature of Parent(s)/Guardian Date</p>			
<p>THIS CONSENT DOCUMENT HAS BEEN APPROVED FOR USE</p> <p>FROM [DATE] THROUGH [DATE].</p>			
<p>_____ Signature of Investigator Date</p>		<p>_____ Signature of Witness Date</p>	

Appendix G: Interview Protocol

Date: _____
Location of Participant (U.S. State): _____
Interview #: _____
Recruitment Source: _____
Code Name: _____

INTRODUCTION

Lindsey to the participant: Thank you so much for your willingness to participate in this interview study. I'd like to tell you a bit more about the purpose of this study. Through this research, I'm hoping to explore the experiences of women who learn that they carry *BRCA1/2* mutations during young adulthood, both as an individual and within family and couple relationships. I'm going to ask you to think back to the earliest time that you knew about the presence of breast and ovarian cancer in your family, and about how your family communicated about that issue. When I say "family," I'm not limiting this to your parents, but including whoever you define as family (e.g. siblings, grandparents). I'm going to ask you to share your experience of genetic testing, and how that was handled in your family. Then, I'd like to ask you about how you have previously or are currently thinking about issues of couple relationships, creating your own family, and managing your personal cancer risk via surveillance, chemoprevention, and/or risk-reducing surgery.

I want to remind you that what you tell me is confidential, in that I will not use your name or your family members' names or any identifying information when reporting my findings. I also want to remind you that as a marriage and family therapist who works with families facing medical and genetic challenges, I am deeply and personally committed to understanding your unique and personal experience of being a young mutation carrier, and how that may impact other important aspects of your life.

The results I generate may help inform the body of knowledge about psychosocial issues affecting mutation carriers, as well as the manner in which challenges faced by mutation carriers are understood by physicians, genetic counselors, and therapists with whom they interact in the physical and mental healthcare settings.

Do you have any questions for me about the purpose of the study or what I hope to do with the results of the study?

Ok, let's get started. Please remember that if you don't feel comfortable answering any of the questions, just let me know and we can skip them. Does that sound ok?

SECTION A: DEMOGRAPHICS & PERSONAL HISTORY

Great. First I'd like to get some basic demographic information from you.

1. What is your date of birth?
2. With what racial and ethnic group or groups do you identify?
3. When did you undergo testing for *BRCA1/2*?
4. Have you ever been diagnosed with cancer?
 - a. *Have you had any cancer scares? For example, abnormal test results, finding a lump during a breast self-examination, etc.*²⁰
 - b. *How have you dealt with these experiences?*

SECTION B: FAMILY CANCER HISTORY

Lindsey: Okay, for the next few questions, I'd like you to think back to the time *before* you underwent genetic testing.

1. Tell me about your family during your childhood.
 - a. *Where did you grow up?*
 - b. *What was your childhood like?*
 - c. *What do you remember about your family at that time?*
 - d. *How did your family deal with health-related issues?*
 - e. *How did your family deal with cancer-related issues during your childhood?*
 - f. *To what extent did members of your family see themselves as "at-risk" for breast and ovarian cancer?*
 - g. *How did these things change over time as you grew up?*
2. Can you tell me about the history of breast/ovarian cancer in your family?
 - a. *How did you experience breast/ovarian cancer in your family? Can you share some of your earliest memories about it with me?*
 - b. *What family members were affected?*
 - c. *With which cancers?*
 - d. *What were their health outcomes?*
 - e. *Which cancer in your family was the most challenging for you to cope with personally? Tell me about that experience.*

²⁰ Italicized questions are "probes," which serve as reminders for the interviewer to gather this information; these items will not be directly requested unless the interviewee does not talk about them in her answer to the larger question.

SECTION C: GENETIC TESTING

Lindsey: Okay, now I'd like you to share with me a little bit about how you came to the decision to pursue genetic testing for *BRCA1/2*.

1. How did you first learn about genetic testing for *BRCA1/2*?
 - a. *Who told you about it?*
 - b. *How was it presented to you?*
 - c. *How did you feel about genetic testing when you first learned about it?*
2. Tell me about how you made the decision to undergo testing.
 - a. *What were your expectations about what testing would be like?*
 - b. *Did you have a feeling one way or the other about what the results would be? Tell me about that.*

Lindsey: Next I'd like to hear about what the testing process itself was like for you.

1. What was your experience with genetic counseling and/or education?
 - a. *What type of counseling and/or education was available to you prior to testing?*
 - b. *What information did you receive during the counseling and/or education process?*
 - c. *How did the genetic counselor communicate with you about your risk of being a mutation carrier? About your risk of developing cancer?*
2. Going into the test, how did you think about your breast/ovarian cancer risk?
3. Tell me about what it was like waiting for your results.
 - a. *How long did you have to wait?*
 - b. *What emotions did you experience during that time?*
 - c. *How did your ideas about your chances of being a carrier change during that time?*

Lindsey: Can you remember what it was like for you to receive your results?

1. How were the results communicated to you?
2. To what extent did you find the results surprising or unexpected?
3. What else happened during the genetic counseling session in which you received your results? What else was communicated to you?
4. Tell me about what the experience was like for you.

Lindsey: Can you tell me about how receiving your results affected your personal sense of vulnerability to cancer?

1. How do you currently understand your risk of developing breast and/or ovarian cancer at some point in your life?
2. How do you assess your cancer risk compared to people who do not carry a *BRCA1/2* mutation?

Lindsey: Great, this has been very informative so far. Do you have any questions for me at this point or anything else you'd like to add about what we've discussed up to this point?

SECTION D: COUPLE RELATIONSHIPS

Okay, next I'd like to spend some time talking with you about romantic relationships.

1. I'd like to get a sense of your relationship history. Can you tell me about the significant couple relationships you've had in your life?
2. Tell me about what's going on in your life right now in terms of relationships.
 - a. *Are you in a relationship?*
 - b. *Are you where you thought you'd be at this point in your life in terms of relationship? Tell me about how that does or does not match up for you.*

If single,

3. How has learning about your positive mutation status influenced your perception of your ability to get to where you want to be in terms of romantic relationships?

- a. *Do you think that having a mutation will make it more or less difficult for you to find a partner? Why?*
- b. *How did learning about your positive mutation status influence your sense of a "timeline" for relationship development?*

4. How do you think being a *BRCA1/2* mutation carrier has influenced the way that you approach relationships?

5. How do you think future partners will

If in a relationship,

3. Have you shared information about your mutation with your current partner? **If yes, skip to question 5. If no, continue to question 4.*

4. Tell me about how you think about possibly talking to your partner about this in the future. **Note: May borrow questions from "single" category to capture participants' thoughts about future disclosures.*

5. Tell me about how you informed your

respond to learning about your mutation?

- a. *How do you envision telling a future partner about your mutation?*
- b. *What do you think that experience will be like for you?*

- c. *How do you think that sharing your mutation status with a partner in the future will influence the way s/he views you? The relationship?*
- d. *How do you think that sharing your mutation status with a partner in the future will influence the relationship intensity? Mutuality? Intimacy? Trajectory?*

6. What do you think the negative and positive effects of sharing information about your mutation with a future partner might be?

current partner about your mutation.

- a. *How far into the relationship did you choose to disclose your mutation to your partner?*
- b. *What was the nature of your relationship at that time?*
- c. *How did you tell your partner about your mutation?*
- d. *What was that experience like for you?*
- e. *How did your partner react?*
- f. *Did the process of telling your partner about your mutation go the way that you thought it would? How was it similar or different?*
- g. *How do you think sharing your mutation status with your current partner influenced the way your partner views you? The way your partner views the relationship?*
- h. *How do you think that sharing your mutation status with your current partner influenced relationship intensity? Mutuality? Intimacy? Trajectory?*

6. Looking back, what were the negative and positive effects of sharing information about your mutation status with your current partner?

7. In what ways does your mutation impact your relationship with your partner today? How does it come up in the way that the two of you interact?

- a. *How does your partner support you through the experience of being a young mutation carrier?*
- b. *How are current couple/relationship issues related to your status as a mutation carrier?*
- c. *How do you see your status as a mutation carrier affecting couple/relationship issues in the future?*

8. Have you shared information about your relationship with other partners prior to this one? How were those experiences similar or different?

***Note: If single individuals have been in significant relationships since learning about their mutation, applicable questions from the “not single” category will be asked to learn about their experiences in communicating information about the mutation to previous partners.*

**Note: Individuals in relationship may relate stories from multiple relationships that they have had since learning about their mutation, if applicable.*

Lindsey: Is there anything else you’d like to share about how your experience as a *BRCA1/2* mutation carrier has intersected with your experiences in romantic relationships?

SECTION E: FAMILY FORMATION

Great. Next I’d like to talk to you about your thoughts and feelings about having kids.

1. Do you currently have children?
2. Are you considering having / planning to have (more) children in the future?
 - a. *If no, tell me about how you came to that decision.*
3. How have your feelings about being a parent changed as you have moved through the different relationships we talked about a few minutes ago?
4. Tell me about ways in which your experience as a *BRCA1/2* mutation carrier has intersected with your experience as a (potential) parent?
5. How did learning about your positive mutation status influence your ideas about the future with regard to having children?
6. How do you understand the risk of passing your *BRCA1/2* mutation on to your biological children?
 - a. *To what extent is this risk something that you spend time contemplating?*
 - b. *How do you think that risk influences/will influence the way you make decisions about having kids?*
7. Have you considered any other methods of having children besides natural conception? For example, adoption, Pre-implantation genetic diagnosis and IVF, donor eggs?
 - a. *Which of these have you considered?*
 - b. *What are your thoughts about these alternative methods?*

- c. *Is this something that you and your partner (current or previous) have talked about?*
 - d. *How might you talk to a future partner about these issues?*
8. How does/did your positive mutation status influence the timeline in which you (might) have/had children?
- a. *Is this something that you and your partner (current or previous) have talked about?*
 - b. *How might you talk to a future partner about these issues?*
9. How do you think about handling BRCA-related issues with your children down the road?
- a. *How do you think you will react if you find out that your child or children is/are BRCA-positive?*
 - b. *How would you approach parenting a child who is at risk?*
 - c. *How do you plan to communicate with a mutation-positive child about what the presence of a mutation means?*
 - d. *At what age would you start communicating with your child about BRCA-related issues?*
 - e. *How does your current/future partner fit into this?*

Lindsey: Is there anything else you'd like to share with me about how your experience as a BRCA1/2 mutation-carrier intersects with issues of having children?

SECTION F: CANCER RISK MANAGEMENT

Okay, we're down to the last section now. This last set of questions focuses on how you think about managing your breast and ovarian cancer risk.

1. How do you understand the implications of your mutation status on breast and ovarian cancer screening and prevention recommendations?
2. Tell me about your experience with communicating/receiving information about your options for managing and/or reducing your risk.
 - a. *With whom have you communicated or from whom have you received this type of information?*
 - b. *What sorts of messages have you received?*
 - c. *What happens when you receive conflicting messages from different people?*
 - d. *What happens when you receive messages from others (e.g., health professionals, family) that contradict your own plans/desires?*
3. What is your current method of risk management or reduction?
 - a. *What are your reasons for using that method at this time?*

- b. *Tell me about your experience with that method of risk management/reduction. How does it impact you physically? Emotionally? Financially? Other ways?*
 - c. *How confident are you that your current method of managing risk is effective? What is the impact of that belief?*
 - d. *(If applicable) How has/have your current (or past) partner(s) been involved in the decisions you've made with regard to risk management/reduction?*
 - e. *How is your current method of risk management/reduction related to your goals or plans for having children?*
4. What other methods of risk management/reduction do you think you might utilize in the future?
- a. *Tell me about how you think about a plan for risk management/reduction as your life proceeds.*
 - b. *Why have you selected that/those method(s) of risk management/reduction?*
 - c. *(If applicable) How has/have your current (or past) partner(s) been involved in the decisions you've made with regard to risk management/reduction?*
 - d. *How are your future plans for risk management/reduction related to your goal or plans for having children?*
5. Are there any other ways in which the way you manage your cancer risk has been or may be impacted by other important issues, such as relationships or having children?

Lindsey: Those are all of the interview questions that I have for you. What should I have asked you that I didn't think to ask?

Do you have any questions for me?

Thank you so much for your help. If I have more questions for you that come up for me later, is it okay to contact you? And, if you think of something else that you forgot to say or if you just think of something else you'd like to tell me regarding your experiences as an adolescent, please call or email me – I'd love to hear from you.

There are a few small things I wanted to check with you about before we end. First, as a part of this study, I am interested in talking to other women in situations like yours, and especially interested in talking to multiple members of the same family. Do you have a sister, cousin, or other female relative who is also mutation-positive and who you think might also be interested in being interviewed for this study?

[IF YES] Would you be willing to pass my contact information along to her so that she may contact me if she is interested in participating?

[IF YES, share contact information]

Name of second contact: _____

Relationship of interviewee to above person: _____

Great. Second, I would like to use a pseudonym, or a different name, when writing about your story. This name will be used in place of your actual name so as to protect you and your family's identity. Is there a specific name that you would like me to use for you?

Ok, thanks. Finally, I will be emailing my study findings – my interpretations and conclusions – to participants to get their feedback and reactions. Would you be willing to read those findings and get back to me with comments?

YES NO

Ok, and if in the future I decide to collect some follow-up data through a survey or questionnaire, would you be willing to participate by filling out some forms like that?

YES NO

Okay, well everything you've told me has been very helpful, and I truly appreciate your taking the time to talk with me today. I will be looking forward to talking with you again when the results have been analyzed, to get your feedback on my interpretation of the data.

This is the end of the interview with [PARTICIPANT'S NAME].

Appendix H: Participant Descriptions & Index

Acacia, 30, married. (BI) As a former participant in the BI study, Acacia had been involved with CGB for several years and had known she was mutation positive for eight years. Acacia's maternal grandmother had both breast and ovarian cancer; the latter resulted in her death before Acacia was born. After Acacia's mother was diagnosed with breast cancer at age 39, she and both of her sisters underwent genetic testing for *BRCA*; Acacia's mother was the only one who received a positive result. Acacia and her sister tested shortly afterward and were both positive as well. Acacia had been with the same partner since high school, and he had accompanied her through the genetic testing and disclosure process and helped her cope with her mom's struggle with cancer. Acacia and her husband had accelerated their family formation plans to allow them to complete childbearing shortly after Acacia's 30th birthday. Their first child, a girl, had been born about two years before our interview, and they were expecting their second and final baby, a boy, shortly after. Acacia was planning to undergo both surgeries after she finished breastfeeding her son.

Quoted or referenced on pages: 92, 247, 295, 319

Annie, 32, new relationship. (FORCE) Annie had inherited her *BRCA* mutation from her mother, who had passed away from ovarian cancer at the age of 51. Annie recalled her mom's cancer as something that her mom chose not to share, and Annie regretted not having been more involved. The family was not aware of any other breast or ovarian cancer until Annie's first cousin on her mom's side—to whom Annie was very close—was diagnosed with breast cancer at the age of 37. At that point, Annie, her sister, and their other cousin were all encouraged to pursue genetic testing, and Annie learned of her mutation at the age of 31. She was acutely aware of the ticking of her biological clock, and had decided to undergo RRBm in order to mitigate her breast cancer risk. This surgery was on Annie's calendar at the time of her interview. She had just entered a new but very promising relationship with a man she met through mutual friends, and was thrilled at having just shared information about her mutation with him and being met with a very positive response.

Quoted or referenced on pages: 92, 200-202, 208, 221-222, 281-282, 321-322

Audrey, 28, married. (FORCE) Audrey had one of the most unique stories of all participants. She and her husband learned that they both carried *BRCA2* mutations when their one-year-old son was diagnosed with a rare form of leukemia that results from Fanconi Anemia (FA). Although Audrey's family had been aware of a "huge" family history of cancer, genetic testing had never been recommended. Three of Audrey's great aunts had had breast cancer, as had three of their daughters; a male cousin had also been diagnosed with prostate cancer. After Audrey's son passed away from leukemia, she and her husband struggled to decide whether to have additional children, because there was a 25% chance that any child they created together would also inherit both *BRCA* mutations and would have FA. Ultimately, they chose to have another baby, but used prenatal genetic diagnosis (PND) to screen the fetus for FA, having decided that if the test was positive, they would terminate the pregnancy. Subsequently, Audrey gave birth to a healthy daughter (who inherited neither mutation) and a healthy son (who had inherited

only one mutation). They decided they were finished having children; Audrey had complete RRBM and was contemplating RRSO.

Quoted or referenced on pages: 92, 188, 218-219, 302-303

Beth, 30, engaged. (FORCE) The youngest of three children, Beth was one of the most recently tested participants, having only known about her BRCA mutation for three months when interviewed. Beth's mom had passed away as a result of ovarian cancer when Beth was 20, and her sister, three years her elder, had become a sort of surrogate mother. Additional family cancer history included breast cancer in Beth's maternal grandmother and pancreatic cancer in a maternal great-aunt. About a year after she had her second baby, Beth's sister had been diagnosed with breast cancer, and it was that diagnosis—in light of the family history—that had prompted Beth's sister's physician to recommend genetic testing for BRCA. Beth was engaged and had a young son with her fiancé; she was urgently focused on completing her RRBM before her son reached one year of age because her sister was diagnosed a year after having her baby.

Quoted or referenced on pages: 92, 178, 207-208, 248-249, 251, 273, 277-278, 297, 308-309, 320

Charlotte, 26, single. (FORCE) Charlotte's unique family history of cancer was characterized by the presence of a BRCA mutation on her dad's side of the family; however, Charlotte's mother had been diagnosed with sporadic breast cancer when Charlotte was in middle school. Three of her father's aunts had had breast cancer many years ago, but the family was not aware of the threat of cancer until Charlotte's cousin (her father's brother's daughter) was diagnosed with breast cancer at age 37. Because of her mom's breast cancer, Charlotte had been receiving "high-risk" screening, but her risk was considered more elevated after she received her positive mutation test result at age 24. Charlotte's sister had also been tested and was negative. At the time of her mutation testing, Charlotte was in a relationship that had been ongoing since college. Shortly after learning that she carried a BRCA mutation, her relationship ended, and she attributed this in part to her former boyfriend's inability to cope with the challenges of her mutation and his heightened sensitivity to medical issues because of his own family history. Charlotte's immediate family demonstrated a great deal of concern about her health, largely because she was the only individual living with a very high risk status. She planned to have RRBM at some point in the future, but not soon. Charlotte's cousin, Kate, is also a participant.

Quoted or referenced on pages: 92, 161-162, 165-166, 171, 197-198, 204-206, 208-209, 212-213, 238-239, 312-314

Chris, 33, married. (FORCE) Chris's family history of cancer consisted of her maternal aunt's diagnosis at age 30, and her mother's diagnosis at age 50. After Chris had found several lumps herself, a doctor with whom she worked suggested that she talk to her mother about genetic testing, because an affected family member would need to be tested first. Chris had a strong sense that her mother and herself would be mutation positive, which was confirmed by the test. Having been divorced from her first husband and remarried to a man 20 years her senior, Chris's positive test results forced her to consider whether or not she wanted to have additional children with her second husband.

Ultimately, they decided not to do so, and Chris underwent both risk-reducing surgeries within 18 months of learning that she carried the mutation.

Quoted or referenced on pages: 92, 179, 187-188, 215-216, 250, 252-253, 258, 260-262, 303-304

Dawn, 27, married. (FORCE) Dawn had inherited her *BRCA* mutation from her father and was aware of a paternal aunt and paternal grandmother who had both died of breast cancer. Her aunt was diagnosed at only 27 and had died at the age of 30. In addition, three great aunts had died from breast cancer as well. With a history of fibrocystic breasts, Dawn had made and canceled several genetic counseling and testing appointments during college while she worked up the courage to get tested; she had a strong sense that she was positive. At the time of her interview, Dawn had known her mutation status for 4 months and had a two-year-old and a seven-month-old months; she had already undergone RRSO, having felt sure that she did not want any additional children. She had an RRBM scheduled for one week after our interview.

Quoted or referenced on pages: 92, 152, 154, 178, 240-241, 261-262

Elaine, 34, married. (FORCE). Elaine had grown up knowing that her mother had an intense fear of breast cancer, but her mother's diagnosis of ovarian cancer at the age of 58 was a surprise; after being told that 20% of individuals with her diagnosis survive one year, Elaine's mother lived for seven. During that time, they discovered a heavy history of cancer in her mother's family, including ten diagnoses and seven deaths. Elaine grandmother, great-grandmother, great-aunt, and several cousins had all had *BRCA*-associated cancers. Elaine had only known her mutation status for two months when interviewed, and had her RRSO scheduled for the upcoming summer. She was also pursuing RRBM but was unsure about when that procedure would occur. Elaine's primary motivation for undergoing the surgeries so quickly was to preserve her health and to be around for her three young children, a daughter and two sons.

Quoted or referenced on pages: 92, 145-146, 217-218, 253-254, 276, 308

Ellie, 27, serious relationship. Refer to "Ellie's Story" in Chapter 5 for description.

Quoted or referenced on pages: 92, 115-126, 131-133, 135, 151, 197-200

Grace, 30, married. (BI). Grace's mother was diagnosed with breast cancer when Grace was 14. Three of her great aunts and four of her mother's cousins had also had breast cancer; of these, all but the three great aunts had survived. Her mother and several members of her extended family had undergone risk-reducing surgery and had avoided cancer. Grace learned of her positive mutation status at the age of 22 and had completed the four-year Breast Imaging study; during those four years, she had met Jason and gotten married. Originally having planned to have children before pursuing RRBM, Grace's plans changed abruptly and she and Jason decided instead to complete RRBM first; she was the only participant who elected not to have reconstruction. At the time of her interview, Grace was three days past her due date to deliver her first child. She has since had a healthy baby boy. Grace planned to utilize chemoprevention for a few years to manage her cancer risk after completing childbearing, and then have RRSO near age 40.

Quoted or referenced on pages: 92, 174-175, 242-243, 258-259, 316-318, 322

Isabelle, 22, single. (FORCE). Isabelle's mother was diagnosed with breast cancer twice, at ages 25 and 36; the first of these occurred before Isabelle was born. Her maternal great-uncle had also had breast cancer, and there were several other instances of different types of cancer on the maternal side of her family. Isabelle learned of her mutation two months prior to her interview and was just getting started with high-risk surveillance. Because of the very young age at which her mother was diagnosed, Isabelle was already thinking about risk-reducing surgery and when one or both procedures might be appropriate for her. Isabelle's sister, Lilly, is also a participant.

Quoted or referenced on pages: 92, 180, 199-200, 212, 237-238, 248, 300-301, 310

Jane, 30, married. (FORCE) Jane was adopted as an infant, and there was no history of breast or ovarian cancer in her adoptive family. She learned at age 26 that there was a strong history of breast cancer in her biological mother's family, including her mother, aunt, and grandmother. Jane learned of her positive mutation status two years prior to her interview, and was relying on surveillance to manage her risk because she wanted to have and breastfeed more children (she already had one daughter). She was considering doing chemoprevention and/or risk-reducing surgery at some point in the future.

Quoted or referenced on pages: 92, 216-217, 230, 234-235, 246, 249-250, 270-271, 276-278, 307, 325

Julia, 24, serious relationship. (FORCE). Julia had known that she was *BRCA*-positive for about four months at the time of her interview. She was very close to many individuals in her mother's family because her parents had divorced when she was six. When Julia was 22, a maternal aunt was diagnosed with breast cancer at the age of 46 and because of her age, she was referred for genetic testing. One year later, Julia's mother was diagnosed with breast cancer, and both women tested positive for *BRCA*. It later came to light that Julia's grandfather's mother had died of breast cancer, and an uncle had had testicular cancer. Julia was in a serious dating relationship of 2 ½ years, and her partner knew about her mutation but "wasn't a worrier." The two had talked about the possibility of making their relationship permanent, but had been unable to agree about whether or not to have children in the future. Julia was open to considering all types of risk-management in the future. Her younger sister, Nichelle, is also a participant.

Quoted or referenced on pages: 92, 163-164, 171, 284

Kate, 35, serious relationship. (FORCE). Kate was the youngest of three sisters and had inherited her *BRCA* mutation from her father. Kate first became aware of breast cancer in her family when an aunt to whom she was not biologically related developed breast cancer when Kate was 20. Her aunt's cancer was caught very early and there was never any doubt that she would make a full recovery. This led to a perception that breast cancer was easily treatable and not much of a threat. More than a decade later, Kate's sister was diagnosed with stage II breast cancer; when their cancer-free paternal grandmother learned of this, she disclosed that all three of her sisters had also had breast cancer. Kate's diagnosed sister was immediately tested and was positive for *BRCA*, and then Kate and the remaining sister also got tested; Kate was positive and the third sister was negative. Kate had two young children and was going through a nasty divorce during this time, and

underwent RRBM without the support of her soon-to-be ex-husband. Since then, she had started a new relationship with a man whom she planned to marry and have one or two additional children; she then planned to have RRSO. Kate's cousin, Charlotte, is also a participant.

Quoted or referenced on pages: 92, 179-180, 187, 195, 204, 259-260, 292-293

Kristy, 29, single. (BI) Kristy, a PhD student, inherited her *BRCA* mutation from her father. Her paternal aunt and grandmother had both had breast cancer, and at the time of our interview, Kristy's aunt was near the end of a long struggle. Kristy learned of her positive mutation status at the age of 23 and had participated in the intensive BI screening study for the full four years before deciding to undergo RRBM at the age of 28.

Throughout her mutation-positive experience, up to and including her surgery, she was in a committed partner relationship; that relationship had recently ended and she was adjusting to her newly-single status. Kristy believed she would undergo RRSO at some point in the future.

Quoted or referenced on pages: 92, 161-162, 174-175, 202, 206-207, 213, 242, 255-256, 263, 285

Leigh, 35, married. (FORCE) Leigh had three young children ranging in age from three to ten and was a busy working mom. Her own mother was diagnosed with breast cancer at age 66 when Leigh was 23, but has since gone into full remission. Her maternal grandfather and aunt also had breast cancer. Leigh had known herself to be *BRCA*-positive for two years at the time of her interview and was the only mutation-positive female in her generation of the family. After participating in surveillance for most of that time, she had decided to undergo RRBM and was at home recovering when interviewed. She also had her RRSO planned for the following summer.

Quoted or referenced on pages: 92, 147-148, 151, 155-157, 166, 172-173, 178, 183, 188, 245-246, 272-273, 276-277, 308

Libby, 32, married. (FORCE) Libby had known that she was *BRCA*-positive for 11 months at the time of her interview. The history of cancer in her family included her grandmother's two breast cancer diagnoses (the first at age 38), a great aunt who died from ovarian cancer in her 50s when Libby was a child, and her mom's breast cancer diagnosis when Libby was 28. Both her mother and grandmother had had mastectomies, and both women saw this as a very negative outcome because when her grandmother had hers, there were no reconstructive options. Libby was planning to wait until after she was done with childbearing to undergo testing, but when fertility challenges sent her to a specialist, he advised her to test earlier because the fertility drugs could be very dangerous if she were positive. After learning that she carried a mutation, Libby had decided to undergo RRBM prior to getting pregnant so that she could use the fertility drugs without having to worry about developing cancer while pregnant. She was recovering from her surgery and looking ahead to her first pregnancy at the time of her interview. She planned to pursue RRSO after completing childbearing.

Quoted or referenced on pages: 92, 172, 176, 247, 252, 273-275, 295

Lilly, 25, single. (Snowball) Lilly remembered herself as an anxious, worried child who was always concerned about her health. She shared that her family viewed her as “the worrier” and thought that she’d gotten everybody’s share of the “worrying gene.” In fact, her family sometimes kept information from her because they knew how much she would worry about it. Lilly’s mother was diagnosed with cancer at age 25 and again at 36, and Lilly and her sister had always been aware of this; they remembered their mother’s “chicken cutlets” – their name for the breast prostheses she wore when they were children. At the age of 14, Lilly had informed her family that she wanted to have her breasts removed so that she wouldn’t have to get cancer, and she remembered her mom being very upset and feeling guilty about this. As an adult who could understand what this really meant, Lilly was still considering having the surgery, especially because she had just reached the age at which her mother was first diagnosed. Lilly had received her mutation test results just two months prior to her interview. Lilly’s sister, Isabelle, is also a participant.

Quoted or referenced on pages: 92, 177-178, 247, 252, 273-275, 295

Lynn, 24, casual relationship. (FORCE) Lynn learned of her positive mutation status at the age of 19, shortly after her mother was diagnosed with breast cancer. Other cancers in her family included breast cancer in two of her mother’s cousins and her maternal great-grandmother, ovarian cancer in her maternal grandmother, and both breast and ovarian in a maternal great aunt. Lynn had had many abnormal test results during the years that she was doing surveillance, and had undergone RRBM at the age of 23; at the time of her interview, Lynn was nearing the end of her reconstruction process and was thrilled with her decision. She had been very open in sharing information about her mutation and her surgery with various dating partners since learning about her mutation; in fact, she had told her current boyfriend about her RRBM on the night they met. Lynn felt confident that she would also have RRSO after completing family formation.

Quoted or referenced on pages: 92, 202-204, 259-260, 290, 292

Maelie, 33, married. (BI) Maelie was an active participant in the BI study from 2005 through 2008, along with two of her sisters. Her youngest sister, Trixie, is also a participant. Maelie grew up in a large family and was the second of five children (four girls and one boy). She was not aware of any history of breast or ovarian cancer in her family until her mid-twenties, when her older sister was diagnosed with breast cancer at the age of 28, shortly after having a baby. Because of her sister’s age, genetic testing was recommended and *two* mutations were identified: one in *BRCA1* and one in *BRCA2* (this is extraordinarily rare). Subsequently, each of Maelie’s parents was found to carry one mutation and the family became aware of several cancer diagnoses in distant relatives. After aggressive breast cancer treatment and a short remission, Maelie’s sister was diagnosed a second time and passed away at the age of 32, leaving behind a husband and two young daughters. After witnessing the swiftness with which cancer claimed her sister’s life, Maelie took aggressive steps to mitigate her own risk: she drastically altered her plans for family formation (choosing to start her family earlier and have fewer children closer together) and proceeded with both RRBM and RRSO only months after the birth of her third child.

Quoted or referenced on pages: 92-93, 150-151, 163, 182, 241, 294

Marie, 28, married. (FORCE) Among all participants, Marie had known about her *BRCA* mutation for the shortest time – only one month – when interviewed. Marie came from a family of extremely health-conscious women: both her mother and maternal grandmother were conscientious eaters, exercisers, and users of holistic remedies who strove to avoid western medicine when possible. When Marie was a teenager, her grandmother became very sick and was eventually diagnosed with ovarian cancer; Marie shared a bedroom with her grandmother during that illness, and when her grandmother passed away Marie felt a great sense of loss. Several years later, after avoiding seeing a doctor for her severe abdominal pain until she could no longer stand it, Marie's mother was also diagnosed with ovarian cancer. Her mother was still living two years later, and attributed her survival to healthy lifestyle and homeopathic remedies. During the first year that her mother was dealing with cancer, Marie also watched her best friend die from a bone cancer that metastasized to her lungs. Marie and her three brothers were tested for *BRCA* after their mom's doctor suggested that she be tested and the mutation was identified. Although her knowledge of her mutation was still very new, Marie was already looking ahead to both risk-reducing surgeries. Already the mother of one young daughter, she was planning to accelerate her plans for a second baby so that she could move forward with both surgeries as early as possible.

Quoted or referenced on pages: 92, 216, 229, 233, 249, 266, 294, 296-297, 299, 311, 348

Marjory, 30, serious relationship. (FORCE) Marjory learned at age 24 that she had inherited her *BRCA* mutation from her father. She was aware of three cases of *BRCA* related cancer in her father's family: one breast cancer (her aunt), one primary peritoneal cancer (her grandmother), and one ovarian cancer (a great aunt). Marjory remembered having known as a young child that many of her grandmother's siblings had died of various types of cancer, and learned as an adolescent that her aunt had previously had breast cancer (Marjory was not aware of this until her aunt had been in remission for several years). After she graduated from college, Marjory's parents informed her that based on her father's family's cancer history, the family had been enrolled in a study at a large medical research institution and a *BRCA* mutation had been identified, of which Marjory's father was also a carrier. After meeting with a genetic counselor at age 22, Marjory waited two more years before deciding she was ready to be tested herself. In the years since she learned about her mutation, Marjory had been using surveillance to manage her risk; recently, she had decided to pursue RRBM in the very near future and planned to have an RRSO after she completed childbearing. Her partner, whom she planned to marry, was fully supportive of her risk management plan and open to developing a plan for family formation that would accommodate it.

Quoted or referenced on pages: 92, 165, 167-168, 176, 213, 222-223, 233, 244, 267, 282, 291, 314-315

MaryAnn, 26, engaged. (FORCE) Mary Ann's mother was diagnosed with Stage IV ovarian cancer when MaryAnn was 15, and died when MaryAnn was 17. Her maternal grandmother and great-grandmother both had breast cancer. MaryAnn learned of her mutation status just two months prior to her interview and had just completed her first round of surveillance; she and her fiancé, both PhD students, were involved in ongoing

discussions about how her mutation would impact other parts of their lives together, and when each risk-reducing surgery would be appropriate.

Quoted or referenced on pages: 92, 229, 234, 264-265, 289-290, 293-294

Melanie, 31, married. (FORCE) Refer to “Melanie’s Story” in Chapter 5 for description.

Quoted or referenced on pages: 92, 125-133, 151, 168-169, 178, 233, 271, 275, 282, 310

Monique, 26, single. (BI) Monique’s mother died from breast cancer when Monique was only five years old; her family history of cancer also included a diagnosis at age 20 and death at age 22 in Monique’s maternal aunt, and a first cousin who had survived ovarian cancer. Monique’s older half-sister had had both risk-reducing surgeries and remained cancer-free. Monique was doubly burdened because she was also at very high risk for cervical cancer. These two risks together had created a sense of doubt for Monique about whether she wanted to (or would be able to) have children. Being single exacerbated this, because she worried that she would have trouble finding a partner willing to accept her high risk status. Prior to enrolling in the BI study, Monique was interested in RRBm but was unable to find a physician who would do the procedure on someone so young (even though her family cancer history included a diagnosis at age 20). At age 26, Monique still envisioned RRBm as part of her future risk management.

Quoted or referenced on pages: 92, 148-149, 209, 222, 312, 314

Nichelle, 21, in an open relationship. (Snowball) The youngest participant, Nichelle had been tested for *BRCA* just prior to her 21st birthday and had known she was mutation-positive for only two months when interviewed. Her family cancer history included breast cancer in her mother and maternal aunt, testicular cancer in her maternal uncle, and kidney cancer in her maternal grandmother; none of these were fatal. A maternal great-grandmother had died of breast cancer, and the family was aware of several other potential instances of cancer in more distant relatives but did not know all of the details about those. Nichelle’s mother’s cancer was caught *very* early thanks to her knowledge that her sister was *BRCA*-positive. Nichelle and her sister Julia (also a participant) were referred for genetic testing shortly after their mother tested positive for *BRCA*, and Nichelle was just starting to contemplate how surveillance would be a part of her life. Regular screening via breast MRI, mammogram, etc. would likely be several years away for Nichelle because of her age. Nichelle was in an open relationship with a partner to whom she had not disclosed any information about her mutation or the presence of cancer in her family. She envisioned RRSO, but not RRBm, as part of her future.

Quoted or referenced on pages: 92, 168, 211, 220-221, 265, 270

Noelle, 26, serious relationship. (FORCE) Breast and ovarian cancer first became personally relevant to Noelle when her paternal aunt was diagnosed with ovarian cancer at age 45 when Noelle was in college. Two years later, Noelle learned that that aunt and then Noelle’s father had tested positive for a *BRCA* mutation and Noelle’s parents suggested that she get tested, too. The genetic counseling and testing process happened very quickly, and within six weeks after that conversation, Noelle received her positive results at age 24. The family suspected that the mutation had been passed down from Noelle’s paternal grandfather, but because he was an orphan, nobody had information

about any health history in that part of the family; Noelle's aunt's ovarian cancer diagnoses was very unexpected, and Noelle's aunt felt very guilty for having "brought" this issue into the family. Other members of the family felt very lucky that Noelle's aunt's doctors had thought to recommend genetic testing based solely on the age at which her aunt was diagnosed, with no other relevant family history. Noelle was in a serious relationship that she hoped might become permanent, but was somewhat frustrated with her partner's reluctance to move forward with the relationship. She believed she would have both risk-reducing surgeries at some point in the future.

Quoted or referenced on pages: 92, 177, 197, 206, 230-231, 286-287

Pauline, 30, married. (FORCE) Pauline's first knowledge of breast cancer came when her paternal grandmother was diagnosed during Pauline's early childhood. Her grandmother had a unilateral mastectomy and died many years later of unrelated causes. Interestingly, Pauline's *BRCA* mutation came from her paternal grandfather. That man (who had prostate cancer) was one of nine siblings, eight of whom had some type of cancer, including four breast cancers, two prostate cancers, two colorectal, and one each of leukemia, bladder, brain, and lung cancers (some people had more than one primary). In addition, Pauline's second cousin of a similar age had been diagnosed with breast cancer at age 30. However, Pauline was not aware of any of this cancer history except for her grandparents, until her father's cousin called him one day to report that a *BRCA* mutation had been identified in this distant branch of the family tree. She shared the exact mutation results with Pauline's father and suggested that he be tested; when he tested positive, Pauline followed suit. When interviewed, Pauline was busily gathering information to enable her to make concrete decisions about when and where to have RRBM; she completed that surgery 9 months after her interview. She and her husband planed to accelerate family formation by several years to enable Pauline to pursue RRSO in her late 30s.

Quoted or referenced on pages: 92, 145-146, 152-153, 159-160, 165, 182, 184-185, 219-220, 227, 236-237, 248, 252, 289, 293, 313-314, 325

Rachel, 33, married. (FORCE) Rachel's family was first touched by cancer when her mother was diagnosed with breast cancer during the summer that Rachel was 12. She recalled knowing her mom was sick because she spent days in bed, whereas usually she "didn't sit still for anything." After a lengthy remission, her mom was diagnosed again when Rachel was 23, and this second bout was recalled by Rachel as much more difficult personally: knowing what cancer treatment entailed and fearing that the difficult chemo and radiation treatments would not be successful, Rachel feared that her mother would not live to become a grandmother or to see Rachel's younger sister get married. After a third diagnosis five years later, Rachel's mom decided to have both breasts and her ovaries removed and did not elect to have reconstruction. The family was also aware that Rachel's mom's paternal aunt had had ovarian cancer, and based on this family history, genetic testing was recommended; Rachel learned that she was mutation-positive shortly after giving birth to her first child. Based on that information, Rachel and her husband decided to accelerate their plans for a second baby, and after their son was born Rachel underwent both RRBM and RRSO.

Quoted or referenced on pages: 92, 150, 153-154, 160-161, 169-170, 172-241, 296-298, 301-302, 311, 349

Reina, 29, married. (BI) Reina's mother was first diagnosed with cancer twice when Reina was eleven years old, and she had vivid memories of her mom's cancer treatments and the "stitches, staples, and chemo" that came along with her experience. Shortly after the family celebrated ten years in remission, Reina's mother was diagnosed again; chemo this second time was followed by a double mastectomy and RRSO, and during this time her mother also tested positive for *BRCA* (the first in the family to undergo testing). The only other case of cancer Reina knew of in her family was her maternal grandmother's breast cancer, which occurred after her mom's cancer and when her grandmother was in her 70s. Reina struggled with a strong sense that cancer was an inevitable part of her future, and this belief was confirmed when Reina (and later her younger sister) also tested positive for *BRCA*. Since then, Reina has struggled to reconcile her experiences with her mother's cancer with what she believes about her own future health, and has tried to convince herself that her fate can be different from her mom's. She met her now-husband during her years in the BI study, and she is happily married and looking forward to starting a family within the next several years; she believes that RRSO will be her risk-management strategy of choice after childbearing is complete.

Quoted or referenced on pages: 93, 154-155, 178, 183, 214, 229, 231-235, 239, 288-289, 345-349

Rose, 30, married. (FORCE) Rose's mom was diagnosed with ovarian cancer when Rose was 10, and died when Rose was 13. Her maternal grandmother also had and died from ovarian cancer before Rose was born. Rose learned of her mutation status about six months prior to her interview; at the time of the interview, she was recovering from RRBM. The primary reason she had made this decision so quickly is that she and her husband were ready to start a family, and she wanted to put that surgery behind her before she had children. She was planning to have a risk-reducing oophorectomy after she was done having children, no later than age 35.

Quoted or referenced on pages: 93, 147, 162, 167, 215-216, 229, 231, 243, 251, 256-257, 270, 282-283, 315, 351, 396

Ruby, 34, married. (BI) Ruby was raised by a mother whose own mother and two maternal aunts had all died from ovarian cancer. The family was one of the original families involved in genetic studies for *BRCA*, and Ruby's mom's life (and to a lesser extent, her own) was permeated by the presence of this threat. Members of her mother's generation were having risk-reducing surgeries even before the *BRCA1* and *BRCA2* mutations were identified. Ruby was offered testing after she was finished breastfeeding her second child, and tested positive at the age of 31. When interviewed, Ruby was still actively participating in the BI study, but she planned to have RRSO after her time in the study was complete. A diagnosis of ovarian cancer in a distant cousin close to Ruby's age had raised her anxiety about her risk in recent years.

Quoted or referenced on pages: 93, 159, 235-236

Rylan, 34, serious relationship. (BI) The story of breast cancer in Rylan's immediate family started even before she was born; her mom was diagnosed eight years before Rylan was born, and then again in the opposite breast when Rylan was seven years old. Both of these were treated with chemotherapy and radiation, but two more diagnoses, this time of metastatic breast cancer, occurred when Rylan was 11 and 15 and ultimately resulted in her mother's death. Rylan felt as though she had an unrealistic impression of what cancer was like because her mom worked so hard to hide its negative effects from her. The other family cancer history of which Rylan was aware was ovarian cancer in a great aunt and breast cancer in her maternal aunt and grandmother. Rylan had gone through what she referred to as a "reckless" period in which she did not pay much attention to her cancer risk because she felt as though, as an almost-thirty single woman, she was missing her chance to have a family before her time "ran out" when she reached the ages at which other family members had been diagnosed. After meeting her partner, Rylan's attitude shifted and she had completed RRBM shortly before her interview. She and her partner were looking ahead to getting married soon and talking about how their family formation plans would fit in with her plans to complete RRSO in the relatively near future.

Quoted or referenced on pages: 93, 108, 157-158, 169-170, 254-255, 262-264, 285, 287, 306-307, 315-316

Sadie, 33, married. (BI) Sadie grew up hearing the story of how her first time on an airplane had been at the age of two weeks when her mom flew with her to be with Sadie's maternal grandmother, who was died from breast cancer while they were in the air. That grandmother's twin sister also died of breast cancer, and a third sister had both breast and ovarian cancer and passed away when Sadie was 19, and Sadie's great-grandmother had had breast cancer as well. Based on that pervasive family history, Sadie's mother and all four of her sisters had all undergone RRBM and RRSO prior to the identification of the *BRCA* genes, because they believed that "everyone in our family gets cancer." Sadie, who was seven when her mom had the surgeries, remembers her mother telling her, "by the time you're old enough to have these surgeries, there will be a cure for cancer," implying that the surgeries would therefore be unnecessary. After learning that she was mutation positive at 28 and during her engagement, Sadie chose to speed up her family formation plans so that she could complete her family and then proceed with surgery. However, she and her husband were unsuccessful in conceiving after several years of trying, and ultimately decided that they did not want to have children. Sadie had since undergone RRSO and was contemplating when RRBM would also be completed.

Quoted or referenced on pages: 93, 150, 155-156, 181, 186-187, 244-245, 290-291, 304-305

Serena, 30, single. (BI) In Serena's family, the "magic age" for cancer is 46 – her mom had been diagnosed with ovarian cancer at that age, and her maternal aunt and grandmother had both been diagnosed with breast cancer at that age. All of these women had survived and were in full remission. Consequently, Serena viewed cancer as something that would happen to her in the future (probably at age 46), and that she would survive, and her life would go on. She was participating in surveillance, but did not go as often as she should because to her thinking, cancer was still more than a decade away.

Serena was not planning to have any risk-reducing surgeries, but acknowledged that she might have to have surgery as treatment in the future. She wanted to have a family, but also wanted that to occur in the context of marriage and was having trouble identifying desirable dating partners in her area.

Quoted or referenced on pages: 93, 169, 205-206, 214, 266, 283, 300-301

Shannon, 31, married. (FORCE) Shannon first remembered being impacted by cancer when her maternal aunt was diagnosed shortly after giving birth. Over the next five or six years, her aunt had another baby, was diagnosed a second time, and passed away when Shannon was 11. Shannon's maternal grandmother and her three sisters had also been diagnosed with breast cancer, and all but one of these passed away as a result. Shannon's mother was diagnosed with breast cancer when Shannon was 15, and Shannon remembered that this was when she first felt that cancer was a risk for herself. Her mom's cancer treatment was very aggressive, and she had been in remission ever since. Shannon decided early on that she would have RRBm, and was not interested in genetic testing – she felt as though even if she received a negative result, she would want to have the surgery because cancer was so pervasive in the family. However, she was unable to find a physician who would perform the surgery without the genetic test results, and so Shannon was tested at age 29. Shannon and her husband had two young children, and they were undecided about whether or not to have a third baby. Shannon's RRBm was scheduled for several weeks after her interview, and they planned to revisit the issue of a third child after the surgery was complete. Once that decision was solidified, they would select an appropriate time for RRSO.

Quoted or referenced on pages: 93, 151, 239, 297, 299, 302, 310-311, 320

Sophie, 31, married. (BI) Sophie inherited her *BRCA* mutation from her father. Two of her paternal aunts were diagnosed with and died from ovarian cancer when Sophie was in her late teens; in addition, her father's father's mother (her great-grandmother) died from ovarian cancer. Sophie had been doing surveillance through the NIH for the past four years; to her knowledge, nobody in Sophie's family had done any risk-reducing surgeries. She was planning on RRSO in her mid-30s and an RRBm at some point. Sophie's two sisters were also both mutation positive.

Quoted or referenced on pages: 93, 158-159, 180-181, 223-224, 288, 310

Trixie, 27, serious relationship. (BI) Trixie was an active participant in the BI study from 2005 through 2008, along with two of her sisters. Her oldest surviving sister, Maelie, is also a participant. Trixie grew up in a large family and was the youngest of five children (four girls and one boy). She was not aware of any history of breast or ovarian cancer in her family until her early-twenties, when her oldest sister was diagnosed with breast cancer at the age of 28, shortly after having a baby. Because of her sister's age, genetic testing was recommended and *two* mutations were identified: one in *BRCA1* and one in *BRCA2* (this is extraordinarily rare). Subsequently, each of Trixie's parents was found to carry one mutation and the family became aware of several cancer diagnoses in distant relatives. After aggressive breast cancer treatment and a short remission, Trixie's sister was diagnosed a second time and passed away at the age of 32, leaving behind a husband and two young daughters. Trixie's experience with watching her sister struggle with and

ultimately succumb to breast cancer was a very powerful one, and she had decided that she did not want to pass along that genetic legacy to a next generation; she had decided not to have biological children. Trixie chose to undergo RRBM shortly after finishing her four years in the BI study, and was contemplating when RRSO would also be appropriate; the only thing keeping her from doing it was her worry about the long-term effects that the hormonal shifts associated with RRSO would have on such a young women.

Quoted or referenced on pages: 92-93, 163, 170, 174, 198-199, 240, 255, 262, 305-306

Valerie, 29, married. (FORCE) Valerie's mom was diagnosed with breast cancer twice, once when Valerie was five and again when Valerie was seven; she died when Valerie was ten. Two of her maternal aunts had also had breast cancer, as did her great-grandfather, as well as several other more distant relatives on that side of her family. Valerie learned of her mutation status at age 25 and underwent RRBM at age 28. Because her family was part of a large HBOC study, Valerie had been doing high-risk screening since she was about 20. She and her husband were both in graduate school and had decided almost definitely that they did not wish to have children; Valerie thought of her ovaries as "on notice" that if anything suspicious was detected, they would be removed without hesitation.

Quoted or referenced on pages: 93, 143-144, 175-176, 179, 185-186, 220, 246-247, 254-255, 258, 261, 264

Wanda, 32, married. (BI) Wanda lost her mother to ovarian cancer when she was only ten years old; she remembered somehow knowing that this news was coming after her mom had been "more in that out" of the hospital over several months, and Wanda connected her knowledge that her mom's illness was called "cancer" with her recollection that her cousin's mom (not a biological aunt) had died from cancer several years earlier. Wanda's mother's sister had had ovarian cancer and cancer in both breasts, but was still living in remission at the time of Wanda's interview. Wanda first learned about *BRCA* mutations during graduate school—she studied in molecular biology and ended up at a University with a large medical center that was running *BRCA*-related studies during her tenure there. Wanda was able to be tested and found out at age 24 that she was positive. Wanda felt that her educational background gave her helpful perspective in thinking about her mutation status not as a death sentence, but as an important piece of information that could inform her decision-making. Wanda was the mother of a two-year-old daughter and expecting a baby boy at the time of her interview. She and her husband were contemplating having a third baby, and believed that RRSO would be their risk-management strategy of choice after they felt certain that their family was complete. Regardless of what they decided, they wanted to be finished with childbearing before Wanda reached 37, the age at which her mother was first diagnosed. She was leaning toward not having RRBM unless a problem in one of her breasts was detected, believing that breast screening techniques were good enough that she would feel protected by continued surveillance.

Quoted or referenced on pages: 93, 145, 165, 176-177, 242, 298-299, 302

Appendix I: Codebook

Level	Code Label	Code Description	Sources	References
..... TREE NODES				
..... DEMOGRAPHIC INFORMATION				
1	Interview date/time	When telephone interview occurred	40	40
2	Date of birth/age	Date of birth and age	40	40
	Race/ethnicity	Self-described race and ethnicity	40	40
	Testing date	Date on which participant learned of positive mutation status	40	40
..... FAMILY AND CANCER HISTORY				
1	Cancer Scares	Abnormal screening tests, other events that elicited fear of cancer diagnosis for self	40	42
1	Family History	General descriptions of family history and relationships (not cancer-related)	23	33
2	Family breast/ovarian cancer experience	Knowledge of breast/ovarian cancer-related incidents in relatives	40	143
3	Breast/ovarian cancer deaths	Knowledge of relatives who have died from breast or ovarian cancer	27	68
	Male breast cancer	Knowledge of male relatives diagnosed with breast cancer	4	5
2	Family other cancer history	Knowledge of incidence of other types of cancer in relatives	20	28
	Family awareness of cancer risk	Description of relatives awareness of hereditary cancer risk	9	11
	Testing of relatives	General knowledge of genetic testing experiences of relatives	32	63
3	Inheritance theories	Family or personal theories about how/why mutation was inherited	19	21
	Inheritance hopes	Hopes about their own or others' mutation statuses	9	13
2	Relatives' surgeries	Knowledge of risk-reducing surgery of family members	28	48
	Unique family experiences	(Category; descriptions below)		
3	Adoption	Participant or close other was adopted	2	7
	Alcoholism	Participant or close other addicted to alcohol	1	1
	Living overseas	Participant spent part of life living overseas	2	2
	Loss of a parent	Impact of a parent's death	12	37
	Loss of both parents	Impact of death of both parents	1	1
	Cancer expert in family	Member of family of origin in profession dealing with cancer	3	5

3	Holocaust survivors	Legacy of holocaust survivorship in family	1	1
	Parents are first generation in US	Participant's parents are first generation born in US (grandparents immigrated)	1	3
	Multiple mutations	More than one <i>BRCA</i> mutation is present in the family	3	7
4	Fanconi Anemia	Diagnosis of Fanconi Anemia in child resulting from presence of 2 <i>BRCA2</i> mutations	1	11
3	Serious accident	Impact of a serious accident in family	1	1
	Mental illness	Impact of diagnosis of mental illness in a family member	2	3
	Special needs child	Impact of participant's special needs child	1	2
2	Non-family cancer experiences	Exposure to cancer experiences of close others not in family	9	12
1 FAMILY OF ORIGIN RELATIONSHIPS			
2	Parents	Relationship with both parents	20	37
3	Mom	Relationship with mother (biological or adopted)	35	190
	Dad	Relationship with father (biological or adopted)	32	86
	Step-parents	Relationship with step-parent(s)	5	6
2	Siblings	Relationship with siblings, generally	7	8
3	Sisters	Relationship with sister(s)	27	122
	Brothers	Relationship with brother(s)	19	32
	Step-siblings	Relationship with step-sibling(s)	1	1
2	Aunts	Relationship with aunt(s)	28	83
	Uncles	Relationship with uncle(s)	16	22
	Grandparents	Relationship with grandparent(s)	35	79
	Cousins	Relationship with cousin(s)	22	44
	Great-grandparents	Relationship with great-grandparent(s)	7	9
	More distant relatives	Relationship with other third+ degree relatives	14	16
1 GENETIC TESTING			
2	Awareness of <i>BRCA</i> in family	How family members became aware of presence of mutation	23	32
	Awareness of testing	How family members became aware of opportunity to be tested	28	34
	Genetic counseling	Experience of genetic counseling	29	45
	Decision-making about testing	How participant decided to undergo genetic testing	31	51
	Expectations prior to testing	Belief about test result prior to receiving it	33	41
	Waiting for results	What it was like to wait for results of genetic testing	21	21

	Receiving results	General experience receiving genetic testing results	5	5
3	Emotional reaction	Emotional reaction to receiving results	30	47
	Procedural	Procedural dynamics in result disclosure (<i>e.g.</i> , given by phone)	29	39
	Support person	Who was with participant when she received her results?	25	32
2	Information provided	Information provided to participant by person who disclosed results	22	32
	Cancer risk perception	Perception of cancer risk immediately after learning results	20	23
	Overlap with couple relationship	Ways in which genetic testing and couple relationship issues are related	17	28
	Overlap with family formation	Ways in which genetic testing and family formation issues are related	3	4
	Change in access to care	Noted differences in ability to access care after receiving results	8	10
1 BREAST/OVARIAN CANCER RISK PERCEPTION			
2	General	Thoughts about cancer risk	11	17
	Anxiety	Experienced anxiety about risk	14	26
3	Alleviation of risk anxiety	How risk anxiety has been alleviated	6	8
	Anxiety-reducing cognitions	Cognitive processes that lower anxiety	9	17
2	Breast vs ovarian cancer risk	Differential perception of risk at these two sites	16	23
	Perceived risk of other cancers	Perception of risk at sites other than breasts or ovaries	7	10
	<i>BRCA1</i> vs <i>BRCA2</i>	Understanding of variation of risk by mutation	4	5
	Sense of inevitability	Belief that a cancer diagnosis is inevitable	15	18
	Fatalism	Fatalistic views about cancer risk	3	3
	Specific periods of risk	Belief that risk changes over time	14	18
	Inaccurate information	Perception of risk based on believing inaccurate information	12	12
	Does not feel vulnerable	Participant does not feel vulnerable to cancer	1	1
1 COUPLE RELATIONSHIPS			
2	General relationship history	Description of general history of couple relationship(s)	35	56
	Disclosure of mutation	General comments about disclosure of mutation status to partner	2	2
3	To spouse	Experience disclosing mutation to spouse	11	16

	To existing partner	Experience disclosing mutation to partner in existing non-marital relationship	19	27
	To new partner	Experience disclosing mutation to partner in relationship that began after genetic testing		
4	Actual	Lived experience of disclosure to new partner	18	28
	Hypothetical	Imagined future experience of disclosure to new partner	12	25
	Timing within relationship development	Selecting when to inform a new partner about mutation	7	9
3	Non-disclosure	Chose not to tell a partner (past or current) about mutation		
4	To past partner	Chose not to tell a past partner about mutation	4	4
	To current partner	Chose not to tell current partner about mutation	1	1
3	“Damaged goods”	Felt that mutation is a flaw that others will perceive as a reason not to be together	7	10
	Disclosure as relationship test	Perceives that partner disclosure was/will be a test of solidity of relationship	7	12
2	Urgency to find LTR	Felt a sense of urgency to identify permanent partner	13	24
	Provision of support	Partners’ provision of support to participants, generally	9	12
3	Lack of support	Felt a lack of partner support	10	14
	Support wishes	Specific types of support desired	10	13
	Ways of supporting	Specific types of support received	32	86
2	Partner’s perception of participant	Belief about how one is viewed by partner	14	18
	Partner’s emotional reaction	Belief about partner’s emotional experience of <i>BRCA</i>	21	28
	Discussions about family formation	Discussion with partner about family formation, generally	22	31
3	Partner’s concerns about children	Perception of partner’s concerns about health of future children related to <i>BRCA</i>	3	4
	Partner’s opinion about family formation	Perception of partner’s opinion about family formation	22	31
2	General impact on couple relationship	Perception of general impact of <i>BRCA</i> on couple relationship	9	11
3	Positive impact	Ways that <i>BRCA</i> has impacted couple relationship positively	19	20
	Negative impact	Ways that <i>BRCA</i> has impacted couple relationship negatively	20	29
	Pace of relationship development	Impact of <i>BRCA</i> on pace of relationship development	8	12

2	Ongoing discussion about <i>BRCA</i>	Current discourse with partner about <i>BRCA</i>	21	31
	Sexual relationship	Ways that <i>BRCA</i> has shaped sexual relationship with partner(s)	9	18
	Educating partner	Experience educating partner about <i>BRCA</i>	10	11
	Lifestyle changes	Lifestyle changes made with partner because of <i>BRCA</i>	2	2
1 FAMILY FORMATION			
2	Current children	Children currently in family (biological or adopted)	20	25
	Plans for (more) children	Current thinking about whether participant will have additional children, generally	17	19
3	Urgency to complete family	Feels a sense of urgency to complete childbearing quickly	22	32
	Family formation timeline	Preferred or chosen timeline for childbearing	33	69
	Family size	Preferred or chosen total family size	26	41
	Competing demands	Description of demands that compete with family formation priorities	17	21
	Childless by choice	Does not have children by own choice	4	5
	Childless NOT by choice	Does not have children for reason outside of control	1	2
	Fertility concerns	Worry about ability to have biological children	12	17
	Discussions with medical professionals	Discussion with providers about family formation, recommendations	18	19
2	Pregnancy	Experience of pregnancy as a mutation carrier	8	11
3	Pregnancy and cancer risk	Concern about influence of pregnancy on cancer risk	8	9
2	Risk of passing mutation to child(ren)	Thoughts about possibility of passing mutation to child(ren), generally	14	15
3	Hopes for advances in medical technology	Hopes that scientific advances will mitigate future risk to children	24	29
	Anxiety-reducing cognitions	Cognitive processes that reduce anxiety about risk to children	14	17
	Outsiders' perceptions	Opinions of outsiders about risk of passing mutation to child(ren)	3	3
	Willingness to accept risk	Reason for belief that risk to children is acceptable	16	23
1 RISK REDUCTION & MANAGEMENT			
2	Current assessment of	Belief about risk currently	40	40

	risk			
	Recommendations	Advice received from providers about risk management	33	60
	Availability of information	Access to desired information about risk management options	14	25
	Surveillance	General thoughts about surveillance as a risk-management strategy	10	13
3	Surveillance during pregnancy	Ability to be reliably screened while pregnant	7	12
	Experience of surveillance	Experience of relying on surveillance for risk-management	31	64
	“False positives”	Experience with abnormal tests that did not actually indicate cancer	10	14
	Confidence in protection with screening	Confidence that surveillance was effective in protecting from cancer	26	31
	Future surveillance	Plans to use surveillance in the future	10	14
	Why chosen	Reasons for choosing surveillance as a risk-management strategy	11	15
	Failure of surveillance to catch something	Surveillance failed to detect a <i>BRCA</i> -related problem	7	7
	Surveillance for other cancers	Undergoing surveillance for cancer other than breast or ovarian	1	1
	Hopes for improvement	Hopes that effectiveness of surveillance will improve with time	2	2
	Access to needed screening	Ability to access needed surveillance tests	9	11
2	Chemoprevention	Use of chemopreventive agents to manage cancer risk	15	19
	Risk-reducing surgery	Thoughts about surgery, generally	3	3
3	Which surgeries	Which surgeries are desired, planned or completed	34	62
	Surgery timeline	When surgeries will occur	33	82
	Interaction with providers	Surgery-related interactions with healthcare providers	16	26
4	Services not provided	Noted deficits in services provided by healthcare system	5	6
3	Reconstruction	Decisions and thoughts about reconstruction options	27	57
	Hormone replacement	Decisions and thoughts about use of hormone replacement therapy	7	10
	Decision-making about surgery	How decisions were or will be made about surgeries	33	89
	Preparing for surgery	How carrier has prepared or will prepare for risk-reducing surgery	6	11

	Grief about surgery	Carrier's feelings of grief about surgery	5	5
	Experience of surgery	General experience of surgery	2	2
4	Physical	Physical experience of surgery	10	13
	Emotional	Emotional experience of surgery	12	15
3	Confidence in protection from surgery	Confidence that surgery will effectively mitigate cancer risk	23	31
	Recovery	Process of recovery from risk-reducing surgery	11	20
	Surgical menopause	Experience of surgical menopause after RRSO	19	22
	Emotional impact	Emotional impact of risk-reducing surgery	9	17
	Additional surgeries in future	Plans for another surgery in the future	23	30
2	Overlap with relationships	Ways in which issues related to risk-management and relationships are related	20	44
	Overlap with family formation	Ways in which issues related to risk-management and family formation are related, generally	4	10
3	Breastfeeding	Ways in which issues related to risk-management and breastfeeding are related	17	24
	Fertility	Ways in which issues related to risk-management and fertility are related	18	29
	Timing	Ways in which issues related to risk-management and timing of family formation are related	25	41
	Parenting during surgery and recovery	Experience of parenting during surgery and recovery	7	9
..... FREE NODES				
	Advice for other carriers	Best advice for other carriers for successfully dealing with positive <i>BRCA</i> mutation status	4	6
	Body image	Impact of mutation on body image	16	24
	Burden	Perception that one is a burden on others because of mutation	4	6
	Change in outlook on life	Ways that knowledge of <i>BRCA</i> mutation has altered one's outlook on life	7	11
	Control	Issues related to feeling "in control"	22	29
	Culture	Ways in which issues related to mutation and culture are related	8	14
	Education and college	Ways in which issues related to mutation and education are related	16	29

Financial and insurance issues	Impact of mutation on financial and insurance issues	32	65
FORCE	Role of FORCE in <i>BRCA</i> experience	10	16
Generativity	Ways that participants have been generative with regard to their mutation status	16	32
Guilt and blame	Feelings of guilt and blame in interpersonal relationships	16	30
Humor	Use of humor in coping with positive mutation status	8	19
Interactions with medical community	Notable interactions with healthcare providers	31	74
Religion and spirituality	Ways in which mutation status and religion/spirituality are related	9	12
Representation of <i>BRCA</i> in media	Noted representation of <i>BRCA</i> -related issues and experiences in books, magazines, news, television, etc.	12	14
Sexuality	Impact of mutation on sexuality, not specific to sexual relationships	6	10
Study involvement	Participation in research studies related to positive <i>BRCA</i> mutation status	14	47
Support	Dynamics around support, generally	22	47
Tension in family of origin	Presence of tension in relationships with members of family of origin	8	9

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