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I Wish I Had Known This!: Impact of Age on Life Choices and Testing Satisfaction for BRCA1/2 Mutation Carriers who Underwent Genetic Testing By Age 25

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I Wish I Had Known This!:
Impact of Age on Life Choices and Testing Satisfaction for *BRCA1/2* Mutation Carriers who Underwent Genetic Testing By Age 25

by

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Dedication

While reading through survey responses, I came across one that just stopped me. It is just a simple thing, but it was something my mom always tells me. The participant had responded to a question, which asked them to give advice to those who will find out they are *BRCA1/2* positive in the future. She wrote: *"I would tell them to breathe. Knowledge is power and knowing is such a blessing. Many people will never know that they will get cancer. This is such a blessing to those who can know and do something about it."*

This response reminded me so much of something my mom would say to me. So—I dedicate my thesis project to my mom, the lady who made me breathe and remember the blessings I do have throughout the writing of this thesis. In addition, I would like to dedicate this to anyone who is BRCA1/2 positive. There is so much we could all learn from the strength and kindness of the amazing women I encountered through the collection of this data you are about to see.

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Abstract

Objective*:* To examine if *BRCA1/2* mutation carriers who received their positive genetic test result by age 25 were satisfied with their decision to undergo genetic testing and with the choices made regarding family planning, surveillance, and surgery. **Methods:** 72 participants recruited via social media completed a survey hosted by SurveyMonkey.com. Sixty-three met study criteria and were asked 40 quantitative and qualitative questions designed to assess family planning, surveillance, and surgery needs of young BRCA carriers, which included a six question Satisfaction with Decision Scale. **Results:** Regardless of age, participants were very satisfied with the decision to undergo genetic testing. Recommendations were made for the counseling and care of *BRCA1/2* mutation carriers under age 25, which included: (1) Right Reproductive Organs, (2) Risk Reducing Mastectomy, (3) Risk Figures, (4) Reproductive Options, and (5) Resource for Future. **Conclusions:** Participants desired more clear and unbiased care and counseling, where they felt supported. The complexity of HBOC plus the variable lives *BRCA1/2* positive emerging adults face led us to propose a core set of counseling recommendations for young BRCA mutation carriers under age 25. Incorporating the five recommendations is essential to achieving full patient autonomy and unbiased decision facilitation.

Keywords: cancer, oncology, *BRCA1*/*2*, genetic testing, testing satisfaction, emerging adults

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List of Abbreviations

ANOVA ..... Analysis of Variance Test

BRCA1 ..... Breast Cancer Gene 1

BRCA2 ..... Breast Cancer Gene 2

BSO ..... Bilateral Salpingo Oopherectomy

CBE ..... Clinical Breast Exam

HBOC ..... Hereditary Breast and Ovarian Cancer

IVF ..... In-Vitro Fertilization

NSGC ..... National Society of Genetic Counselors

PBM ..... Prophylactic Bilateral Mastectomy

PBO ..... Prophylactic Bilateral Oopherectomy

PGD ..... Pre-implantation Genetic Diagnosis

SWDS ..... Satisfaction with Decision Scale

Chapter 1: Background

1.1 *BRCA1* and *BRCA2* Basics

Cancer is a major health problem in many countries. In the United States, it causes one in four deaths. In 2012, there were 229,060 new cases of breast cancer. Of these, 226,870 were females and 2,190 were males. Breast cancer was the cause of 39,920 deaths. Of these deaths, 39,510 were females and 410 were males. Ovarian cancer was reported in 22,280 new cases and 15,500 deaths. (Siegel, Naishadham, & Jemal, 2013).

Breast and ovarian cancer are not unusual in the general population. One in eight, or 12.5% of women will develop breast cancer over their lifetime while one to two in one hundred, or 1-2% of women will develop ovarian cancer. The average risk for cancer gradually increases over one's lifetime (Petrucelli, Daly, & Feldman, 2011). While a majority of cancers are sporadic or randomly occurring, about 10% of all breast and ovarian cancer cases are hereditary, or passed down though families.

Individuals with hereditary cancer have a higher risk of developing cancer than individuals in the general population. Hereditary cancer is due to germline mutations, or genetic changes that are passed down from parent to child. Among the individuals that fall within this hereditary breast cancer susceptibility group, 35% of cases will be due to a mutation in the *BRCA1* gene while 37% of cases will be due to a mutation in the *BRCA2* gene. Among those in the hereditary ovarian cancer susceptibility group, 80% will have a *BRCA1* mutation and 15% will have a *BRCA2* mutation. The remaining cases are due to other genes or unknown causes (Clark & Domchek, 2011; Ford, et al., 1998).

BRCA1 and *BRCA2* are the two genes that cause the genetic condition known as Hereditary Breast and Ovarian Cancer Syndrome (HBOC). HBOC is a hereditary cancer predisposition syndrome that is caused by mutations or changes in one of these two genes. Each gene has two alleles, or copies. One is passed on from the father and one is passed on from the mother. Receiving a mutated copy of one of these genes from either parent can increase an individual's risk for various types of cancer. In nearly all cases, these mutations are passed down in families. Very few *de novo*, or randomly occurring mutations in these genes have been reported. The incidence of *de novo* mutations in either the *BRCA1* or the *BRCA2* gene is thus unknown.

HBOC syndrome is inherited in an autosomal dominant fashion, meaning that just one mutated copy of the *BRCA1* gene or the *BRCA2* gene will cause this predisposition that may lead to cancer. If a parent has a mutated copy, each child has a 50% risk of inheriting the mutated copy of the gene. The highest cancer risk is for breast or ovarian cancer, but other cancers such as pancreatic, prostate, or fallopian tube cancer can result. A slight yet increased risk has been found for melanoma in *BRCA2* carriers and for endometrial cancer in *BRCA1* carriers (Clark & Domchek, 2011; Segev et al., 2013).

The overall prevalence of *BRCA1* and *BRCA2* mutations in the general population is between 1/400 and 1/800 individuals. The incidence depends on ethnicity. The highest incidence is seen in the Ashkenazi Jewish population. The incidence is 1/40 primarily due to three common founder mutations, 187delAG and 5385insC for *BRCA1* and 6174delT for *BRCA2*. Dutch and Icelandic populations also have a higher incidence of *BRCA1* or *BRCA2* gene mutations (Petrucelli et al., 2011).

There are differences in lifetime cancer risk for women depending on whether they carry a mutation in the *BRCA1* or the *BRCA2* gene. On average, 60-70% of women with a *BRCA1* gene mutation will develop breast cancer by the age of 70, while an average of 40% will develop ovarian cancer, which includes fallopian tube and primary peritoneal carcinomas. For women with a *BRCA2* gene mutation, an average of 45-55% of women will develop breast cancer by age 70 while 20% will develop ovarian cancer. Women also have an increased risk for other cancers such as pancreatic cancer or melanoma (Clark & Domchek, 2011). The highest risk for these two types of cancer in women is thus associated with a *BRCA1* gene mutation (Petrucelli et al., 2011). Women have up to a 10% risk of pancreatic cancer as well (Ford, et al., 1998).

Males with a *BRCA2* gene mutation have up to an 8% risk of developing breast cancer. Those with a *BRCA1* mutation have up to a 1% risk (Petrucelli et al., 2011; Shiloh, Dagan, Friedman, Blank, & Friedman, 2013). Males have up to a 2-6% risk of gastric and pancreatic cancer, with the highest risk being for males with a *BRCA2* gene mutation. Men have a slightly increased risk for prostate cancer over that of the general population (Shiloh et al., 2013). The age of cancer onset is similar to that of sporadic breast cancers in the general population with *BRCA2* mutations (Ford et al., 1998).

1.2 Targeting Individuals who Tested BRCA Mutation Positive as Emerging Adults

Individuals who are between the ages of 18 and 24 are in a highly transitional period of their lives now known as emerging adulthood. Emerging adulthood, a phrase coined in 2000 by Arnett, describes a unique demographic period that pertains to the exploration of one's identity through the postponement of adulthood. Young, BRCA mutation positive individuals are potentially making major choices and decisions that will

impact their lives for years to come. These decisions can include career, significant other, family planning, and geographic location choices. By the late 20s, a majority of individuals have made decisions in their lives that will have lasting consequences. Further, when adults retrospectively considered the most crucial events in life, they mainly cited events in this period (Arnett, 2012).

As a result of these fast-paced changes and choices, researchers have started to examine if having a *BRCA1* or *BRCA2* gene mutation impacts this tumultuous time in life (e.g. Hoskins, Werner-Lin, etc.). Individuals who fit into this emerging adulthood group and who are from families with an identified *BRCA1/2* gene mutation may base decisions regarding normal life transitions upon a timeline of when they expect illness to occur. Anxiety, worry, and grief regarding disease have been found to play an underlying role in this transitional era in life when hereditary cancer is an issue (Werner-Lin, 2007).

There is much research that still needs to be done for this critical demographic group, because the current professional medical guidelines available to young, BRCA positive individuals suggest genetic testing be made available when they reach the age of eighteen (Trepanier et al., 2004). However, this recommendation is based on the principle that individuals of this age can now act as autonomous adults. Though they are able to make informed decisions about their own risks and genetic testing, the true autonomy is debatable. For young, high-risk individuals of this age group, autonomous decision-making is one of the milestones in development that may not yet be reached (Arnett, 2000). Many individuals in this age group may still live at home, at least part time, and still be dependent on their parents or guardians for support. This support could range from financial support to facilitating their child's decision-making processes.

The major concern is that young, BRCA positive individuals may experience greater harm and lesser benefits to their psychosocial well-being when learning about their *BRCA1/2* mutation carrier status. Compared to individuals older than 25, individuals diagnosed with a *BRCA1*/2 gene mutation as emerging adults may find that there are fewer concrete recommendations for them to utilize once they know their status. With or without the support of their family members or other health care providers, the decision-making processes that young, BRCA positive individuals undertake are distinct from individuals of the same age in the general population. This age group is also unique from all other individuals who are a part of the overall group of families with HBOC. Young mutation carriers know they are BRCA positive by age 25, but there is little published data on what actions to take regarding this life-changing information.

In two related studies, it was determined that complex decision making processes evolve over time. The ability to fully understand and act on newly discovered genetic information and to make confident, life-altering decisions is still developing in this period. Therefore, it will be critical to have support and guidance at this point in the life cycle to assure young, BRCA positive individuals make autonomous, informed choices where they fully understand both the benefits and risks of knowing one's mutation status. This study suggested that it would be critical to have concrete goals to model decision-making upon, as well as resources from providers who will deliver the genetic education and information to this age group. Support and guidance to assure young, BRCA positive individuals are making autonomous, informed choices and fully understand the benefits and risks of knowing one's mutation status is key (Werner-Lin, Hoskins, Doyle, & Greene, 2012). Specifically, these young BRCA mutation carriers report a desire for

clearer screening guidelines and continuing support regarding their medical care over time, as their life and needs evolve (Hoskins, Werner-Lin, & Greene, 2014).

In another study pertaining to women who are 18-24 years old, Patenaude et al. (2013) looked at levels of concern and knowledge about hereditary cancer in young women who have *BRCA1* or *BRCA2* positive mothers. They found that one third of the daughters reported high cancer-related distress, even though they had expressed normal levels of general distress. Knowing this genetic information about their mothers raised concerns for their futures, especially in regards to having children. The level of knowledge about HBOC was suboptimal with many misconceptions about their risk for carrying a BRCA mutation. This indicated that future studies could be vital in determining how young, BRCA positive women are coping after they undergo genetic testing themselves and also test positive for a *BRCA1* or *BRCA2* gene mutation.

This study collected data from several different, industrialized countries that may inform individuals and health providers in similarly industrialized countries. Delaying marriage and childbearing in favor of gaining an education first is increasing among industrialized countries. Individuals from industrialized countries in this age range may experience comparable dilemmas. This is because emerging adulthood is not a universal developmental period, but a stage in life seen only in cultures that postpone entry into adult responsibilities until individuals are in their early to mid twenties (Arnett, 2000).

1.3 The Risks Faced by BRCA Mutation Carriers By Age 25

Females and males under age 25 are at a low risk to developing cancer. Even though breast cancer is the leading cause of cancer death among women, breast cancer does not make the top five leading causes of cancer-related deaths in women under age

20 (Siegel et al., 2013). For young *BRCA1/2* gene mutation carriers, the relative risk at this time may seem higher. Particularly since their absolute risk or lifetime risk for cancer is significantly higher than other women in the general population.

In actuality, young *BRCA1/2* carriers have a small chance to develop an HBOC related cancer before age 30. The risk for developing breast cancer before age 30 is about 4.6% for women who carry a *BRCA1/2* mutation (Ford et al., 1998) while the risk for developing ovarian cancer is nearly zero (Stratton et al., 1999). Between the ages of 20 to 24, the risk is even lower. It is estimated that the incidence for *BRCA1* and *BRCA2* mutation carriers is .02% for breast cancer and .001% for ovarian cancer (Antoniou et al., 2003). While these numbers are low, *BRCA1* and *BRCA2* mutation carriers between the ages of 20 and 29 still have a relative risk to have breast cancer that is 5 to 20 times higher then a woman of the same age in the general population (Clark & Domchek, 2011). For males, the risk of developing cancer is even lower. Male cancer risk typically does not start until age 40 (Shiloh et al., 2013).

The looming lifetime risk for cancer may drive many young individuals to undergo genetic testing earlier then some guidelines suggest (Ormondroyd et al., 2012). This is because individuals who are 18 to 24 years in age do not yet have a fully developed frontal lobe of their brain, leading them to have impaired judgment, morality, and abilities to make long-term plans. This time in life is still a critical period in the development of the human brain (Steinberg, 2005). Young mutation carriers may feel life is now like a waiting game, or that they are a ticking cancer bomb, even though the actual risk for developing cancer is not inevitable (Kwong & Chu, 2012). Young, BRCA

positive individuals may act as though cancer development will be experienced in the near future even though this is improbable (Hoskins & Warner-Lin, 2012).

While these fears may push some young carriers to consider surgical treatment or risk-reducing drug options. Many surgeons fear that these decisions may cause regret for young, BRCA positive individuals later in life. Surgeons may be cautious to remove seemingly healthy breast and especially ovarian tissue from women in this young age group due to the risks associated with these major surgeries such as pain syndromes and body image issues, even if they do carry a genetic mutation (Kwong & Chu, 2012). Young, BRCA positive individuals may seek advice from their primary health care providers, surgeons, or gynecologists. Many health care providers are unlikely to be trained to handle the sensitive nature of topics related to BRCA for this age group. Unlike many other mutation positive individuals, young, BRCA positive individuals' surgical decisions cannot be made quickly. This group needs support to assure that they have considered all facets related to their decision and its potential ramifications, since they are less likely be mature when making independent decisions and acting autonomously (Werner-Lin, Hoskins, Doyle, & Greene, 2012).

Despite the low risks of developing cancer, young, BRCA positive individuals may continue to face anxiety and fear once they know their carrier status. Genetic counseling and care for young, BRCA positive individuals involve considering psychosocial adaptation and risk management like all other patients. This group is unique in the sense that the traditional options given to individuals, such a surgical choices and reproductive planning, may not seem to apply at the time of genetic test result disclosure. Reproductive issues and surgical options are related topics however.

Thus, they may be highly appropriate subjects for young carriers to learn of in a genetic counseling session, since they may not have begun or completed their families (Kwong & Chu, 2012; Ormondroyd et al., 2012).

While young, BRCA positive individuals may be hesitant to learn all of this information; basic knowledge will be key for future decisions. The ability to carefully think ahead and plan for the future is a rare opportunity this group has, where compared to older BRCA carriers. The sensitive and complex nature of these BRCA-related topics as well as the likelihood that they will have an impact on each individual's life requires careful discussion with trained health care providers to navigate through possible issues (Ormondroyd et al., 2012). Otherwise, there is a possibility that young, BRCA positive individuals may experience distress regarding having genetic testing or the choices that they have made based on their *BRCA1/2* positive status.

Emerging adults in this high-risk group can be made aware of recommended options for surveillance, chemoprevention, and prophylactic surgery. All surveillance guidelines state that mutation positive individuals should start formal surveillance at the age of 25 or older. Guidelines also suggest that high-risk individuals could start screening or surveillance as many as 10 years prior to the earliest age of diagnosis in their family. Although for some families, this age still may not fall before 25 years of age, continuing to leave young, BRCA positive individuals without concrete options.

When reviewing core guidelines for high-risk individuals, it is clear why this group feels worried and confused about what they should do with this knowledge regarding their genetic make-up (Werner-Lin, Hoskins, Doyle, & Green, 2012). Recommendations bypass this group. It is recommended that annual mammograms and

breast MRIs should begin at the age of 25. Clinical breast exams (CBE) should be done annually or bi-annually starting at age 25, which can include high-risk men (Berliner, Fay, Cummings, Burnett, & Tillmanns, 2013). Men are often suggested to wait until the age of 40 since they are at a lower overall risk for breast cancer (Shiloh et al., 2013). At the age of 30, women can elect to have CA-125 screening and transvaginal ultrasounds to screen for ovarian cancer, even though the utility has yet to be proven for these tools. At age 40, colorectal cancer screening via colonoscopies and PSAs for prostate cancer should be performed yearly (Berliner, Fay, Cummings, Burnett, & Tillmanns, 2013).

Chemoprevention may be considered, and there are several options for women. Oral contraceptives may be recommended, and have been known to decrease the risk of ovarian cancer up to 45-60% if used over a period of five years in high-risk women who have a history of ovarian cancer (Berliner et al., 2013). Medications such as tamoxifen or other chemopreventive drugs reduce the risk of developing breast cancer as well. In a study of women who have an increased risk for breast cancer either because of family history or because their age greater then 60, tamoxifen reduced the risk for invasive breast cancer by 49%. It also reduced the risk for women 49 and younger by 44% (Fisher, et al. 1998).

There are also two main risk-reducing surgical options for women. A risk reducing bilateral mastectomy may be considered. This surgery reduces the risk of breast cancer by at least 90%. There is no standard age for which this is recommended. In addition, this more aggressive treatment through a surgical procedure may be more strongly considered if the patient has limited access to knowledgeable physicians, chemotherapeutic drugs, and surveillance equipment, such as MRI machines. A risk-

reducing bilateral salpingo oophorectomy (BSO) can also be performed. This reduces the risk for ovarian cancer by 80-96% and reduces the risk of breast cancer by up to 50% in women who have not yet gone through menopause. This is because hormone levels in young, BRCA positive women will drop following the removal of their ovaries, resulting in a decrease in the hormones present that would fuel cancer growth. This surgery is recommended for women between the ages of 35 to 40. This surgery will cause women to enter early menopause. There are currently no guidelines and little data regarding chemoprevention or prophylactic surgeries for high-risk men (Berliner et al., 2013).

Since all of these options are recommended for those ages 25 and older, young individuals in this group may feel helpless in regard to taking control of their health. For some, the fear of developing cancer may lead them to proceed with some of these treatments or surgeries. For others, the concept of the waiting game may seem all too familiar (Hoskins & Werner-Lin, 2012). No matter what path is taken, we aim to better understand the rationale and thought processes for young, high-risk individuals though this study. The hope is that others' experiences will better aid the young, *BRCA1/2* positive individuals of tomorrow.

1.4 Need for Targeting *BRCA1/2* Positive Emerging Adults

According to guideline recommendations, this group of individuals between the age of 18 and 24 are the youngest candidates for *BRCA1* and *BRCA2* testing. While young, BRCA positive individuals have a low risk for cancer during this time in life, BRCA mutation positive individuals still feel as though they are at high relative risk for receiving a cancer diagnosis. They may find themselves in a dilemma as far as their risk management. There is a significant lack of data for individuals who underwent genetic

testing at this age. Previous work has shown that high risk women who carry a *BRCA1/2* gene mutation are satisfied with undergoing genetic testing and would recommend testing to others who are at high risk for breast cancer (Klemp, O'Dea, Chamberlain, & Fabian, 2005). The concern for these *BRCA1/2* positive emerging adults is what to do after the genetic test results are disclosed. There are essentially no guidelines for how they should manage their cancer risks, make life choices, or be counseled regarding reproductive and surgical options (Hoskins & Werner-Lin, 2012).

Our study gathered information from individuals who received their positive genetic test result by age 25, in order to provide information to service providers working with individuals of this age group who have a known positive mutation status. Information obtained is intended as a useful resource that may make it possible for health care professionals and genetic counselors to better care for and understand this unique group of individuals. This information may aid in generating better support and guidance for individuals of this often overlooked age (Hoskins & Werner-Lin, 2012).

This study built on a small but growing collection of literature regarding emerging adults with a *BRCA*1/2 mutation. There has been success with similar studies in the recent past. The population was not as geographically diverse and the sample size was smaller. This study offered participants the opportunity to participate in a fully internet-based survey method. This unique study format, when compared to previous works, enabled us to reach more young mutation carriers (Hoskins, & Werner-Lin, 2012, Hoskins et al., 2014; Werner-Lin et al., 2012).

We hypothesized that individuals who obtained their positive genetic test result by age 25 would be satisfied with their decision to undergo testing as well as with the

choices they have made regarding surveillance and surgery. Further study objectives are below.

- Comparisons will be made between participants under the age of 25 and participants age 25 or older at the time of study but had their positive genetic test result by age 25 (Ford et al., 1998; Steinberg, 2005; Werner-Lin et al., 2012)
    - We predicted participants under 25 will be less satisfied.
    - We predicted participants under 25 would feel their timeline for life is more affected.
- Life instability was analyzed (i.e., more residency changes/shorter relationships) (Ford et al., 1998; Hoskins, Roy, Peters, Loud, & Greene, 2008)
    - We predicted less satisfaction with undergoing genetic testing.
    - We predicted timeline for life and overall life plans would feel more effected.
- Different Types of Metaphors (Comparisons between otherwise unrelated things)
- Have seen and find the role of a genetic counselor important
    - More satisfied with their decision to have genetic testing
    - Timeline for life and overall life plans feels unaffected by HBOC

Chapter 2: I Wish I Had Known This! Impact of Age on Life Choices and Testing Satisfaction for *BRCA1/2* Mutation Carriers who Underwent Genetic Testing By Age 25.[1]

2.1 Abstract

Objective: To examine if *BRCA1/2* mutation carriers who received their positive genetic test result by age 25 were satisfied with their decision to undergo genetic testing and with the choices made regarding family planning, surveillance, and surgery. **Methods:** 72 participants recruited via social media completed a survey hosted by SurveyMonkey.com. Sixty-three met study criteria and were asked 40 quantitative and qualitative questions designed to assess family planning, surveillance, and surgery needs of young BRCA carriers, which included a six question Satisfaction with Decision Scale. **Results:** Regardless of age, participants were very satisfied with the decision to undergo genetic testing. Recommendations were made for the counseling and care of *BRCA1/2* mutation carriers under age 25, which included: (1) Right Reproductive Organs, (2) Risk Reducing Mastectomy, (3) Risk Figures, (4) Reproductive Options, and (5) Resource for Future. **Conclusions:** Participants desired more clear and unbiased care and counseling, where they felt supported. The complexity of HBOC plus the variable lives *BRCA1/2* positive emerging adults face led us to propose a core set of counseling recommendations for young BRCA mutation carriers under age 25. Incorporating the five recommendations is essential to achieving full patient autonomy and unbiased decision facilitation.

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2.2 Introduction

About 10% of individuals with breast and ovarian cancer will have a hereditary cancer predisposition, which can be passed down in their family. Thirty-five percent of BRCA carriers will have a mutation in the *BRCA1* gene while 37% will have a mutation in the *BRCA2* gene, resulting in a genetic condition called Hereditary Breast and Ovarian Cancer Syndrome (HBOC). The highest cancer risks are for breast or ovarian cancer.

HBOC syndrome is inherited in an autosomal dominant fashion, meaning that one mutated copy of the *BRCA1* or the *BRCA2* gene will cause a predisposition for cancer. Mutation carriers have a 50% chance to pass on their BRCA mutation to their offspring. The prevalence of these *BRCA1* and *BRCA2* mutations in the general population is between 1/400 and 1/800. It is higher in specific ethnic groups (Petrucelli et al., 2011). The incidence of *de novo* mutations in either gene is rare and still unknown (Antoniou et al., 2003; Clark & Domchek, 2011; Ford et al., 1998).

On average, 60-70% of women with a *BRCA1* gene mutation will develop breast cancer by the age of 70, while an average of 40% will develop ovarian cancer, which includes fallopian tube and primary peritoneal carcinomas. For women with a *BRCA2* gene mutation, an average of 45-55% of women will develop breast cancer by age 70 while 20% will develop ovarian cancer. A lesser yet increased risk has been found for other cancers, such as pancreatic cancer and melanoma for *BRCA2* carriers or endometrial for *BRCA1* carriers (Clark & Domchek, 2011; Segev et al., 2013).

Females who carry a BRCA mutation have a low risk of developing cancer under age 25. The risk for developing breast cancer before age 30 is about 4.6% (Ford et al., 1998) while the risk for developing ovarian cancer before age 30 is nearly zero (Stratton

et al., 1999). Between the ages of 20 to 24, the risk is lower. It is estimated that the incidence for *BRCA1/2* mutation carriers is .02% for breast cancer and .001% for ovarian cancer (Antoniou et al., 2003). Despite these numbers, *BRCA1*/*2* mutation carriers between the ages of 20 and 29 have a relative risk of breast cancer 5 to 20 times higher than that of women of the same age in the general population (Clark & Domchek, 2011).

The individuals targeted by this study who underwent genetic testing by age 25 are among the youngest genetic testing consumers for *BRCA1* and *BRCA2* single gene testing. While young, BRCA positive individuals have a low risk for cancer during this time in life, they may still feel as though they are at high risk for receiving a cancer diagnosis and may be unsure how to proceed with risk management. Few guidelines outline empirically-based risk management strategies (Trepanier et al., 2004), much less how to support life choices, and be optimally counseled regarding medical management (Hoskins & Werner-Lin, 2012; Hoskins et al., 2014).

The looming lifetime risk for cancer may drive many young individuals to undergo genetic testing earlier then some guidelines suggest (Ormondroyd et al., 2012). However, individuals who are 18 to 24 years of age do not yet have a fully developed frontal lobe of their brain, leading them to have judgment and morality that is not fully developed (Steinberg, 2005). They are also in a transitional period in life, between ages 18 and 25, known as emerging adulthood. This unique demographic period is when individuals explore their own identity through the postponement of an adult lifestyle. Young BRCA mutation-positive individuals are potentially making major choices and decisions that will impact their lives for years to come. Decisions can include career, significant other, family planning, and geographic location choices. By the late 20s, most

individuals have made decisions with lasting consequences. When adults retrospectively consider key life events, they mainly cite this period as a critical time when life-shaping decisions were made (Arnett, 2012).

Researchers have examined how having a *BRCA1/2* gene mutation impacts this tumultuous time in life (e.g. Hoskins, Werner-Lin, etc.) and shapes future life for young, BRCA positive emerging adults (Arnett, 2000). Individuals who are emerging adults and also from families with an identified *BRCA1* or *BRCA2* gene mutation may base decisions regarding normal life transitions upon a timeline of when they expect illness to occur. Anxiety, worry, and grief regarding disease were found to have an underlying role in shaping cancer risk perception at this young age (Werner-Lin, 2007).

Current professional medical guidelines available to young, BRCA positive individuals suggest genetic testing be made available when they reach the age of eighteen (Trepanier et al., 2004). Previous research has shown that high-risk women who carry a *BRCA1/2* gene mutation are satisfied with undergoing genetic testing and would recommend testing to others at high risk for breast cancer (Klemp et al., 2005). However, the concern for these *BRCA1/2* positive emerging adults is what to do after the genetic test results are disclosed. Young, BRCA positive individuals may experience greater harm and fewer benefits when learning about their *BRCA1*/*2* mutation carrier status. Compared to individuals older than 25, fewer concrete recommendations exist for known carriers under age 25. The decision-making processes young mutation carriers undertake are distinct from older BRCA carriers since there is little published data to guide what do with this powerful, personal genetic information.

Support and guidance to assure young, BRCA positive individuals are making autonomous, informed choices and fully understand the benefits and risks of knowing one's mutation status is key (Werner-Lin et al., 2012). Specifically, these young BRCA mutation carriers desire clearer screening guidelines and continuing support regarding their medical care over time, as their life and needs evolve (Hoskins et al., 2014). Patenaude et al. found that one third of the daughters of known *BRCA1/2* carriers reported high cancer-related distress, especially in regards to childbearing. The level of knowledge about HBOC was suboptimal with many misconceptions about their risk for carrying a BRCA mutation (2013).

Young, BRCA positive individuals may feel life is now like a waiting game, or that they are a ticking cancer bomb, even though the actual risk for developing cancer is low (Kwong & Chu, 2012). Young, BRCA positive individuals react to genetic testing and cope with information revealed as if cancer will be experienced in the near future (Hoskins & Warner-Lin, 2012). These fears may push some young carriers to undergo surgical treatment well before the recommended timeframe.

Young, BRCA positive individuals may seek risk management advice from their primary health care providers, surgeons, or gynecologists. Health care providers are unlikely to be trained to handle the sensitive nature of these topics for this age group. Unlike many other mutation positive individuals, emerging adults' surgical decisions cannot be made quickly. This group needs support to assure that they have considered the complexities related to their decision and its potential ramifications over the life course (Werner-Lin et al., 2012).

BRCA1/2 positive emerging adults are unique in the sense that the traditional options given to individuals, such a surgical choices and reproductive planning, may not seem to apply at the time of genetic test result disclosure. Reproductive issues and surgical options may be highly appropriate subjects for young carriers to learn of in a genetic counseling session, since they may not have not begun or completed their families (Kwong, & Chu, 2012; Ormondroyd et al., 2012). The ability to carefully think ahead and plan for the future is a unique opportunity this group has, when compared to older *BRCA1/2* carriers. The sensitive and complex nature of the topics related to BRCA as well as the likelihood that they will have an impact on each individual's life requires careful discussion with trained health care providers to navigate through possible issues (Ormondroyd et al., 2012). Fear of developing cancer may lead some to undergo surgeries or surveillance while for others the waiting game may elicit ongoing anxiety.

Guidelines suggest that high-risk individuals could start screening at age 25 or at 10 years prior to the earliest age of diagnosis in their family (Berliner et al., 2013; Trepanier et al., 2004). Recommendations typically bypass this group. It is recommended that annual mammograms and breast MRIs should begin at age 25. Clinical breast exams (CBE) should be done annually or bi-annually starting at age 25 (Berliner et al., 2013). At the age of 30, women can elect to have CA-125 screening and transvaginal ultrasounds to screen for ovarian cancer, even though the utility has yet to be proven for ovarian cancer screening. Chemoprevention may be considered. Oral contraceptives have been known to decrease the risk of ovarian cancer up to 45-60% if used over a period of five years in high-risk women who have a history of ovarian cancer (Berliner et al., 2013). Medications such as tamoxifen or other chemopreventive drugs

reduce the risk of developing breast cancer as well, reducing the risk for invasive breast cancer by about 44% for women 49 and younger (Fisher et al. 1998).

Two main risk-reducing surgical options are available for women with a *BRCA1/2* gene mutation. Risk reducing bilateral mastectomy would reduce the risk of breast cancer by at least 90%. There is no standard age for which this is recommended. A risk-reducing bilateral salpingo-oophorectomy (BSO) would reduce the risk for ovarian cancer by 80-96% and reduces the risk of breast cancer by up to 50% in women who have not yet gone through menopause. Surgery recommendations are for *BRCA1/2* positive women ages 35 to 40 (Berliner et al., 2013).

This study built on a growing collection of literature on emerging adults with *BRCA*1/2 mutations (Hoskins & Werner-Lin, 2012; Hoskins et al., 2014; Werner-Lin et al., 2012). We aimed to reach a greater number of geographically diverse participants by offering the opportunity to complete an internet-based survey. We gathered information from individuals who received their positive genetic test result by age 25 with the intension of making this information available as a resource for health care professionals. This information will enable providers to better understand the unique support and guidance needs of these very young *BRCA1/2* carriers (Hoskins et al., 2014).

We hypothesized individuals who obtained their positive genetic test result by age 25 would be satisfied with their decision to undergo testing as well as with the choices they have made regarding surveillance and surgery. Further study objectives are below.

- Comparisons were made between participants under the age of 25 and participants age 25 or older at the time of study but had their positive genetic test result by age 25:
  - We predicted participants under 25 will be less satisfied.

- We predicted participants under 25 would feel their timeline for life is more affected.

- Life instability was analyzed (i.e. more residency changes/shorter relationships):
  - We predicted less satisfaction with undergoing genetic testing.
  - We predicted timeline for life and overall life plans would feel more affected.
- We predicated finding numerous metaphors (comparing two unlike things).
- We predicted participants would recommend seeing a genetic counselor.
  - We predicted more satisfaction with the decision undergo genetic testing.
  - We predicted timeline for life and overall life plans would feel less affected.

2.3 Materials and Methods

2.3.1 Sample Selection Criteria. Participants recruited were *BRCA1* and *BRCA2* mutation positive individuals who underwent genetic testing before age 25. Criteria for selecting participants included (1) *BRCA1* or *BRCA2* mutation carriers, (2) over age 18 at the time of the study, (3) received their genetic test result before age 26, and (4) English speaking. Participants could be of any age at the time of the study. Participants not meeting criteria were excluded. Unlike previous works (Hoskins & Werner-Lin, 2012; Werner-Lin, et at., 2012), we did not exclude participants who had received a cancer diagnosis. While the study did invite male participation, the final participant pool included solely women.

2.3.2. Recruitment. Participants were recruited using convenience and purposive sampling. Participant recruitment was open from September 30, 2013 to February 6, 2014. Recruitment proceeded via different forms of social media (Appendix A). Standardized recruitment announcement templates were designed to maintain the

consistency of the invitations for the participants (Appendix B). Each message was tailored to fit the type of social media or online forum. Templates contained an embedded link to the survey and contact information for the investigators.

Recruitment proceeded via invitation posted on three FORCE message boards. Messages were posted on the Main Forum, the Young Previvors Forum, and the Research Opportunities Forum. Further recruitment proceeded through Bright Pink via two tweets from their Twitter account. Facebook groups and blogs related to BRCA aided participant recruitment by posting messages containing the survey link on their pages. Each Facebook group was contacted by direct message prior to the posting being made.

2.3.3. Survey Design. An online survey was constructed by the researcher. A survey from a related, previous work was utilized to guide question creation (Werner-Lin et al., 2012). The final survey was reviewed and approved by the thesis advisory team. The study was hosted on SurveyMonkey.com. It consisted of forty questions designed to assess both participants' satisfaction with their decision to undergo genetic testing and participants' feelings about life choices made (Appendix C). The 40-question survey included:

- Yes/no with corresponding open-ended prompt (15 questions)
- Demographics and participant characteristics (11 questions)
- Open-ended (5 questions)
- Satisfaction with Decision Scale (SWDS) (6 questions)
- Likert-scale style questions (2 questions)
- Yes/no to follow-up call with link to a support resource (1 question)

The 15 yes/no questions each had a quantitative and a qualitative component, which was designed to assess how a positive genetic test result affected decision-making and life choices made. Among these yes/no questions were three questions regarding life planning. The life planning choice questions included asking participants if they had a medical management plan in place, if they had received genetic counseling, if they had been informed about family planning or reproductive options. The Likert scale-type questions were designed to assess the degree of worry over life events and the helpfulness of specific types of healthcare providers. The life events included finding a job, finding a place to live, completing school or duties at work, finding a partner or getting married, having children or family planning, and reducing your risk for cancer via surgery, treatment, etc. Open-ended questions were designed to assess participant perception of cancer risk or knowledge gained through BRCA-related experiences. Participants were asked to share advice for future *BRCA1/2* positive people under age 25.

The demographic and participant characteristic questions were used to obtain data on current age, age at time of genetic testing, sex, race/ethnicity, personal income, and highest educational level. Individuals were asked if they have a *BRCA1* or *BRCA2* gene mutation. Current relationship length and the number of residence changes in the past eight years were asked to assess life stability, since these two factors change frequently for many emerging adults (Arnett, 2000). Participants were also asked if they had been pregnant and the number of sons or daughters that they have. In the final question of the survey, participants were asked if they would be willing to receive a follow-up phone interview if needed. Participants who responded yes were asked to leave their contact information. Participants were provided the link to the "find a genetic counselor" feature

on the National Society of Genetic Counselors (NSGC) website at the in case they felt distressed or in need of additional support. No follow-up phone calls were made.

To determine participant satisfaction with undergoing genetic testing, a previously developed survey tool was used. It was designed to gauge satisfaction with health care decisions (Holmes-Rovner et al., 1996). The Satisfaction With Decision Scale (SWDS) is positively correlated with decision-making confidence and has a proven reliability of 86%. It had a Cronbach's Alpha score of 0.86. The aim was to see if participants were satisfied with their decision to undergo *BRCA1/2* genetic testing at a young age.

The SWDS questions were reviewed and analyzed via a Cronbach's Alpha test, like Holmes-Rovner et al. (1996). It was expected to be reliable since the SWDS had been proven reliable previously in a study of *BRCA1*/*2* mutation carriers who underwent genetic testing after using a computer-based decision aid (Green et al., 2004). The SWDS also is a concise way to evaluate participant satisfaction with undergoing genetic testing. We desired to minimize the potential for loss of participants due to the length of the survey overall (Holmes-Rovner et al., 1996; Kasparian & Wakefield, 2007). This research study was approved by the Institutional Review Board, Office of Research Compliance, of the University of South Carolina, Columbia, SC.

2.3.4. Data Collection. Participants were targeted via a variety of social media venues. Participants who clicked on the embedded survey link, which was found in each of the invitation to participate templates, were brought to the first page of the study. The first page consisted of a consent document that explained the goals of the study, potential benefits and risk to participants, and the contact information for the primary researcher as well as the director of thesis. Following the consent document, the survey consisted of

eight pages of questions. Once data collection was complete, all data was downloaded as excel files and stored on a password-protected computer.

2.3.5. Data Analysis. The primary researcher met with a statistician to develop an overall plan for quantitative data analysis. SPSS predictive analysis software, version 22.0, was used to analyze of the quantitative data. Chi-square test with Fisher's exact test was used to determine correlations and infer independence among categorical data. The categorical data included participants responses to questions such as educational level reached. Spearman's Rho was employed to determine correlations and infer independence for the ordinal data. The ordinal data included participant responses to questions such as the length of time they have been with their current partner and their number of residency changes. Percentages were used to review and depict what percent of young mutation carriers were concerned about specific life events, such as finding a job or reducing their cancer risk. Cronbach's alpha was preformed to confirm the validity of the SWDS tool.

Data from the open-ended questions was analyzed using Content Analysis. Participant responses were printed onto paper, cut into strips, and color-coded based on the content of each participant's response by the researcher. Recurring themes and trends in the overall data were obtained. Frequency counts based on the number of times a particular theme was mentioned in a participant response were obtained. Tables and figures were developed to visually display key findings and participant demographics.

2.3.6. Benefits and Risks of Study. This study provided no direct benefits or risks to participants. Young, BRCA positive individuals may have benefited by having the opportunity to express the knowledge and experience gained through past experiences

(Birch & Miller, 2010). Enabling young, BRCA positive women to share and describe their experiences has potential therapeutic benefits, especially if they felt as though they were helping others (Ziebland et al., 2013). The risks to participants were minimal. When thinking about their own past experiences, participants may have felt sad or upset by their circumstances, decisions, or changes in life.

2.4 Results

2.4.1. Participants. Responses were gathered from 72 participants, but 63 met study criteria ($N = 63$) (Figure 2.1). At the time of survey, about 40% of participants were under age 25 while about 60% were 25 or older (Figure 2.2).

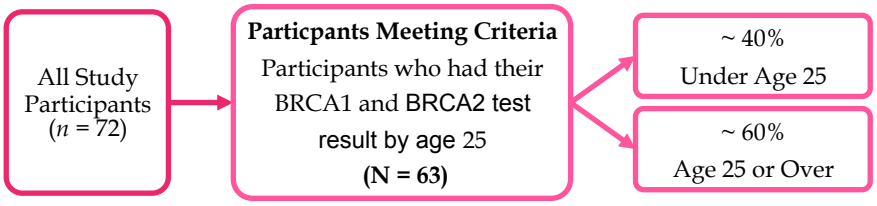

Figure 2.1. Criteria for study participation.

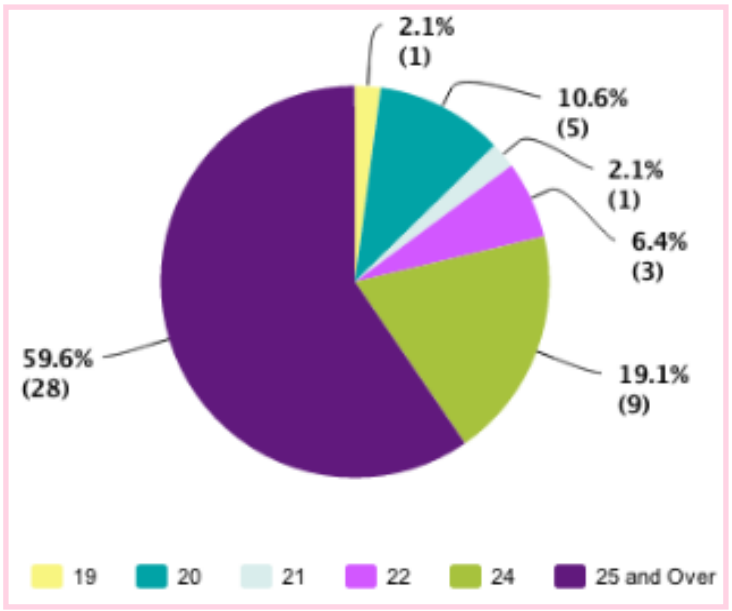

Figure 2.2. Participant ages at the time of study.

Participant ages are shown in Table 2.1. Participants' average age at the time of results disclosure was 22, while the most commonly reported age was 24. The average age at the time of survey was 26, but most participants were age 24. Most participants were White/Caucasian (87%), held a Bachelor's degree (32%), had an average personal income between $30,001 and $50,000 (37%), and had no children (67%). Most often, participants changed their place of residency three times in the past eight years and had been with their spouse or partner an average of three years.

We met our goal of exceeding 60 participants; however, our sample size lacked diversity in terms of sex, race or ethnicity, highest educational level, and income. All participants were women, a large majority were White or Caucasian (87%), over 60% had achieved a Bachelor's degree or beyond and had personal incomes of $30,0001 to $50,000 or greater. Geographically, our study succeeded in reaching out to participants beyond the United States. Facebook groups utilized in obtaining research subjects included at least three groups based out of Canada or the United Kingdom. Among participant responses, at least six participant responses acknowledged that they were located or had received care and services at countries outside of the United States.

2.4.2. Satisfaction. The reliability of the six question Satisfaction with Decision Scale (SWDS) as a single composite score for overall satisfaction with the decision to have genetic testing was determined using a Cronbach's alpha test ($\alpha = 0.835$). Alpha scores were above 0.70 indicating that the SWDS showed good internal reliability and was effective at assessing participants' satisfaction with the decision to undergo genetic testing. Table 2.2 lists the six questions that make up the SWDS.

Table 2.1. Participant characteristics and demographics.

	Average	Range	Mode
Age genetic test result received (years)	22	16-25	24
Age at time of survey (years)	26	19-40	24
Children (*n* = 35)			
Total	0.7	0-4	0
Number of daughters	0.3	0-2	0
Number of sons	0.3	0-2	0
Residency changes (last 8 years) (*n* = 47)	3	0-8	3
Years with current spouse/partner (*n* = 42)	5	0-15	3
	N	**%**	
Sex			
Female		**100%**	
Age at time of survey, by age group			
Under 25	19	40%	
25 and Over	**28**	**60%**	
Gene with mutation identified (*n* = 63)			
BRCA 1	37	59%	
BRCA2	25	40%	
Unsure	1	1%	
Race/ethnicity (*n* = 47)			
American Indian or Alaskan Native	1	2%	
Hispanic American	1	2%	
White/Caucasian	**41**	**87%**	
Other	4	9%	
Education (*n* = 47)			
Finished high school or GED	3	6%	
Some college education	9	19%	
Associate's degree	7	15%	
Bachelor's degree	**15**	**32%**	
College beyond a bachelor's degree	13	28%	
Personal Income (*n* = 43)			
Less than $12,000	8	19%	
$12,0001 to $30,000	4	9%	
$30,001 to $50,000	**16**	**37%**	
$50,001 to $100,000	14	33%	
More than $100,000	1	2%	
Pregnancy (*n* = 46)			
No	**31**	**67%**	
Yes	14	30%	
N/A	1	3%	

Table 2.2. SWDS questions and response options.

The Satisfaction with Decision Scale: **Decision to have genetic testing**
1. I am satisfied that I was adequately informed about the issues important to my decision.
2. The decision I made was the best decision possible for me personally.
3. I am satisfied that my decision was consistent with my personal values.
4. I successfully carried out the decision I made.
5. I am satisfied that this was my decision to make.
6. I am satisfied with my decision.
Response options:
1- strongly disagree, 2- disagree, 3- neither agree nor disagree, 4- agree, 5- strongly agree

Average scores for the SWDS were determined using an ANOVA test, and then compared to participant ages at the time of testing. An ANOVA test was used for the two age categories, to see if there was a difference in testing satisfaction. Results are shown in Table 2.3. Although participants under the age of 25 had slightly higher satisfaction with their decision to undergo genetic testing than the 25 or older age group, this difference was not statistically significant.

Table 2.3. SWDS and age at time of study.

	N	Average Score	Standard Deviation
Overall	**63**	**4.7**	**0.66**
Age group			
Under 25	19	4.8	0.28
25 and over	28	4.6	0.82

The SWDS was compared to the two participant age groups at the time of study, education level, and personal income to see if relationships between participant demographics and satisfaction were associated with the decision to undergo genetic testing. The SWDS was also compared to participants' life planning. Life planning choices included yes or no responses about their BRCA status affecting their timeline for life or overall life plan. It also included if they had a medical management plan in place, if they had met with a genetic counselor, or if they had been informed about family

planning or reproductive options. These comparisons were performed using ANOVA test. The results are shown in Table 2.4.

To assess participant satisfaction with the decision to undergo genetic testing, ANOVA was used to compare whether or not a participant thought their positive *BRCA1/2* mutation status had an effect their overall life plan. This was statistically significant ($p = 0.04$). Therefore, participants who report life plans were impacted by their *BRCA1/2* positive result were more likely to report an effect on their satisfaction with undergoing genetic testing. The most satisfied participants still reported that knowing that they were BRCA1/2 positive impacted their overall life plans. Therefore, it seemed that regardless of the outcome BRCA had on participants' lives, they still were still pleased with their decisions to have genetic testing.

Table 2.4. SWDS vs. participant demographics and life planning choices.

	df			
	between-groups	within-groups	*F*	*p*
Age at time of study, by age group	1	44	1.04	0.32
Education	4	38	1.24	0.31
Personal income	4	44	0.16	0.70
Timeline for life	1	51	1.5	0.23
Overall life plan	**1**	**51**	**4.55**	**0.04***
Medical management plan	1	51	0.61	0.69
Genetic counseling	1	51	0.14	0.71
Family planning or reproductive options	1	50	0.08	0.78

2.4.3. Quantitative Data Regarding BRCA-Related Experiences and Choices.

Participants answered 15 yes or no questions about experiences and choices they have made related to BRCA (see Table 2.5). Nearly half of participants reported that their overall life plans or goals they desired to achieve were affected by testing positive for the BRCA1/2 gene mutation (57%). Nearly three-quarters of participants responded that

testing positive for a *BRCA1/2* gene mutation caused them to feel as though the speed of their timeline for life events was altered (71%). A majority of participants had not received a cancer diagnosis (95%), were undergoing breast screening (77%) with normal outcomes (65%), had undergone some form of breast surgery (58%), and had not undergone a surgery related to the ovaries or female reproductive organs (85%). Only 2% of participants had utilized chemopreventive drugs. A majority had a medical management plan in place (88%) and all had considered risk-reducing surgeries (100%). Most received genetic counseling (84%). A majority of participants had not been introduced to reproductive or family planning options, such as PGD (preimplantation genetic diagnosis) with IVF (in-vitro fertilization), adoption, or surrogacy (52%).

Education level was compared to the three life planning choice questions and age at the time of the study (See Table 2.6). A significant association was found between participant education and if they had received genetic counseling, $\chi^2(4, N = 51) = 11.16$, $p = 0.03$. Participants were more likely to have had genetic counseling if they had reached a higher level in their education.

Regardless of age, most participants reported a mild level of worry about reducing their risk for cancer. More participants under the age of 25 selected the high level of worry option (16%) when compared to those age 25 or older (6%) regarding reducing their risk for cancer. The under 25 group also tended to have more worry regarding completing school or duties at work, with 26% selecting the high concern option. Only 10% of the women in the 25 and older group expressed high worry over school or work duty completion. A majority of women under age 25 expressed a low level of worry with finding a partner or getting married (32%) and having children or family planning (37%),

while most of those age 25 or older said they were not worried (71% and 45%, respectively) and think about these rarely. A majority of women who participated in this study expressed none to low or mild worry levels regarding these life event categories.

Table 2.5. Participants' experiences and choices related to BRCA.

Question	Yes	No	Other	
Q3: Have you ever been diagnosed with cancer? (n=59)	5%	95%		
Q4: Have you ever had a mammogram, MRI, breast ultrasound, or breast imaging study? (n=60)	77%	23%		
Q5: Have you ever had an ABNORMAL mammogram, MRI, breast ultrasound, or breast imaging study? (n=60)	35%	65%		
Q6: Have you ever had a normal or abnormal tissue biopsy? (n=59)	20%	80%		
Q7: Have you ever had breast surgery? (lumpectomy, mastectomy, etc.) (n=59)	58%	31%	11%	Planned but not taken place
Q8: Have you ever had a surgery related to the ovaries or other female reproductive organs? (Hysterectomy, tubal ligation, conization, bilateral salpingo oophorectomy, etc.) (n=59)	14%	85%	1%	N/A
Q10: Have you ever considered having a surgery to reduce your risk of cancer? (n=59)	95%	5%		
Q11: Have you ever used any chemopreventive drugs? (Ex: Tamoxifen, Evista, etc.) (n=57)	2%	96%	2%	N/A
Q13: Do you think testing positive for a gene mutation affected the timeline for your life? For example, do you feel like you need to speed up or slow down some of your plans for the future? (n=51)	71%	29%		
Q14: Do you think testing positive for a gene mutation affected your overall life plan? (Ex: desire to have children, marriage, etc?) (n=51)	34%	57%		
Q15: Do you currently have a plan in place for your future health care, related to your positive BRCA genetic testing result? (Ex: have a medical management plan including starting mammograms at an early age and....) (n=51)	88%	12%		
Q18: Have you ever received genetic counseling? (n=49)	84%	16%		
Q19: Have you ever considered a risk-reducing surgery? (n=49)	**100%**	**0%**		
Q20: Have you been introduced to different types of family planning or reproductive options based on the mutation in your BRCA gene? Examples may include: PGD (preimplantation genetic diagnosis) with IVF, adoption, surrogacy, etc. (n=52)	48%	52%		

Table 2.6. Participant level of concern or worry regarding life events (%).

	Percentage of Participants at Each Level of Concern or Worry (%)				
	Think about rarely: None	Think about once/twice a month: Low	Think about a few days a week: Mild	Think about daily: Moderate	Think about Constantly: High
Finding a job	**58%**	5%	11%	16%	11%
	65%	6%	19%	0%	10%
Finding a place to live	**58%**	26%	5%	11%	0%
	77%	10%	3%	10%	0%
Completing school or duties at work	**32%**	5%	16%	21%	26%
	42%	19%	19%	10%	10%
Finding a partner or getting married	26%	**32%**	16%	16%	11%
	71%	13%	6%	3%	6%
Having children or family planning	11%	**37%**	32%	11%	11%
	45%	32%	16%	6%	0%
Reducing your risk for cancer (surgeries, treatment, etc.)	5%	26%	**42%**	11%	16%
	23%	19%	**29%**	23%	6%

Key: Participant age at time of study, by group: Under 25 (n=19) 25 and over (n=31)

Participants were asked to rate how helpful or unhelpful specific types of healthcare providers were at providing support and guidance. They were asked to say if they found them very helpful, somewhat helpful, neither helpful or unhelpful, somewhat unhelpful, or very unhelpful (shown in figure 2.3).

The number of years participants were with their partners and the number of residency changes participants had in the past eight years were asked to gauge life stability. These questions were compared to whether or not participants felt that their positive *BRCA1/2* mutation status affected their timeline for life or their overall life plans.

These questions were compared using an ANOVA test. There was no significant effect between years with partner and timeline for life, $F(1, 32) = 0.89$, $p = 0.35$. There was no significant difference between the number of years with partner and overall life plan, $F(1, 32) = 0.46$, $p = 0.50$. There was no significant difference between number of residency changes an timeline for life, $F(1, 34) = 0.16$, $p = 0.70$. No significant difference between number of residency changes and overall life plan was found, $F(1, 34) = 0.01$, $p = 0.92$.

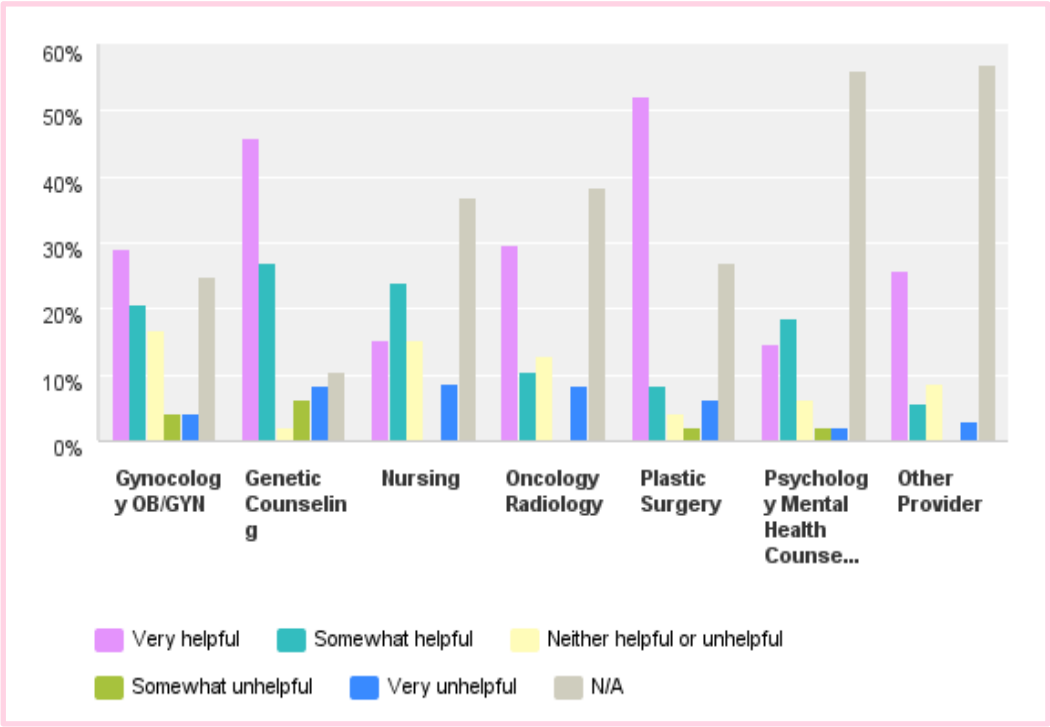

Figure 2.3. Healthcare services rated by their provision of support and guidance.

2.4.4. Themes.

Life experiences and outlook. Participants reported details about experiences they have had with family members or healthcare providers. Participants wrote responses regarding their outlook on life after testing positive for a *BRCA1/2* gene mutation (see Table 2.8), which were analyzed for common themes. Most often, participants had an

accurate perception of their cancer risk (*n* = 22) while many others either overestimated their risk (*n* = 16) or quoted the correct risk figures, yet said they thought their risk was higher based on their family history (*n* = 14). Most also noted seeing family members fight against caner (*n* = 38). A main theme was that knowledge of their *BRCA1/2* had a positive impact and they were hopeful for the future. Participants wrote that it as a chance to seize opportunity (*n* = 14) or that it would not alter plans (*n* = 9).

Positive Outlook.

- I still had children, got married, lived it up in my early 20s, went to college. BRCA positive results did not slow my life down. They gave me the tools I needed to ensure my life would go on.

BRCA was a Positive that Led to a Career Change.

- Testing positive and confronting my risk via preventative mastectomy changed my perspective on life drastically. While I always valued health, being a good person, and contributing to the world in a meaningful way, prior to my surgery I wasn't actively pursuing these priorities. My career plans have changed – rather than staying in the field of marketing I am pursuing graduate school for epidemiology. I pursue goals because I believe they are meaningful and will fulfill me, rather than for the appearance of success. As much of a burden as testing positive is, it is also a blessing in disguise if you let it.

- Made sense that I would be [positive], I can help so many with what I have access to and what I know as a nurse, and personal experience now with surgery, and passion fueled by loss of life in family members.

Table 2.8. Life experiences and outlook.

Life experiences and outlook (n = 168)	n
Family history shaping perception of risk	60
Accurate perception of cancer risk	22
Overestimating cancer risk	16
Quote accurate numbers, but believe it is higher because of family history	14
Underestimating cancer risk	3
Overestimate risk before to prophylactic surgery, now underestimate risk	2
Metaphors as a way to think about cancer risk	2
Quote accurate numbers, but believe it is higher because had cancer	1
Witnessed family members fight against cancer	38
Chance to seize opportunities	14
Positive impact on life plan / led to specific career path	4
Eat healthier and spend more time with family	4
Always felt there was no time to waste because of family history	4
Travel more	2
Conflict about what is best for young carriers	12
Tried to talk out of having prophylactic surgery	3
Pushed to have a family before they felt ready	2
Tried to talk participant into having reconstruction	1
Did not want to work around college class schedule	1
Physician disagreement over hormone replacement therapy use	1
Difficulty providing appropriate care for young carriers	9
Doctor uninformed about BRCA and not knowing how to provide care	4
Genetic counselors with recommendations not fitting for someone so young	3
BRCA will not alter plans	9
Altered self-image	6
Due to mastectomy	4
Due to screening or biopsy	2
BRCA as a death sentence	5
Is not a death sentence	4
Felt like a death sentence	1
Denial about cancer risk	5
Recent or impending loss of a family member as a reason for denial	4
Did not expect to be BRCA positive	1
Feelings of hopelessness	3
Concerns involving insurance	3
Planned for a career with good insurance because of BRCA	2
Concerns over future insurance coverage, even though protected by law now	1
Knowing your mutation status as a blessing	2
Angelina Jolie's decision as eye opening	2

Overestimation of cancer risk.

One participant wrote: I believe my risk would be approaching 100% - every woman on the *BRCA1*+ side of the family has battled cancer more than once, my aunt has had 8 diagnoses of cancer in the last 30 years. I

am the first to have engaged in risky behaviors such as smoking and heavy alcohol consumption. Statically I know that my breast/ovarian risk is quite low now, however with a 53% chance of surviving to the age of 70, I have decided to approach my carrier status as absolute, otherwise I fear gambling with statistics might cost me my livelihood or my life.

Another wrote, "95% because both my grandmothers had breast cancer and my mother had both breast and ovarian cancer and now that I know I have a *BRCA1* gene mutation I know my risk is vary high." A third explained, "I had a 98% chance by age 30, it wasn't an option to not be preventative [and undergo mastectomy]." Another participant used a metaphor to explain her risk: "Before surgery, it was 55-85% [for breast cancer] …for ovarian 10-60%, 'average risk of 31%'. The way I see it, even if my risk for breast cancer was 'only' 45 or 55%, I made the right decision for me in having prophylactic surgery. If you knew that 'only' 1 out of 2 vehicles sold by a particular manufacturer exploded spontaneously, would you still buy the car?"

Affected Self-Image.

- [I had] double-mastectomy with skin and nipple-sparing reconstruction. The months leading up to the surgery were the worst part emotionally. The anticipation and emotional preparation for losing a part of your body, particularly a part so close tied to your womanhood, is extremely difficult. I struggled with the fact that I wouldn't be able to feel my breasts anymore, that I wouldn't be able to breast-feed, and that I had no guarantee I would ever look "normal" again. Recovery was difficult, but manageable with pain medication. The hardest part for me was giving up

my mobility (not being able to use my arms, drive, exercise – even if I wasn't in pain) until my drains were removed… The expansion process brought on insecurities – did my boyfriend still find me attractive? Did people get weirded out when I hugged them and my boobs felt hard as rocks? I pushed through… The overall result has been fantastic. Cosmetically? My boobs rock. Better than my natural ones ever were. I had a great doctor!

- I had a normal MRI guided core biopsy. …This experience was considerably more trying then expected… I was surprised how negatively my self-image was affected by the procedure, the busing, the pain in my breast. It also brought forward thoughts and concerns of how I will be able to cope if I choose to go forward with surgeries in the future. I found these concerns in addition to the stress of the procedure to be more stressful than any worry about potential results.

Denial.

- Anyone who asked, I told them I was 95% sure I did not carry the gene, so I had no concern about being told [my result] over the phone. I got the call at home while my daughter was napping, and it was just me. I was devastated. I could not get off the phone fast enough because I just wanted to sit and cry by myself. After I calmed down, I called my husband at work and told him, and then cried all over again. …It was a major shock to me.

Hopelessness. Two participants wrote about hopelessness in relation to finding out their test result. One replied, "Mixed [thoughts on prophylactic surgeries]. I suspect I will do them and then die of pancreatic cancer at 60. So it feels a bit useless. Then again better to die at 60 than at 35 I guess." Anther wrote, "There was a certain amount of hopelessness that came along with my test results… it paralyzed me for a long time."

Impact of Angelina Jolie's Decision.

- I eventually came to the tentative decision that a prophylactic mastectomy was something I'd be interested in doing as soon as I moved back to the states. And then Angelina Jolie wrote that article about her own decisions and suddenly all I could focus on were the negative comments. People calling her surgery 'self mutilation' and an overreaction. I started to back-track a little. Was the decision I had come to (independent of her article) short-sided? Was I only doing it out of panic? I threw myself back into researching the same things I had before, but this time I had the added bonus of doing it AFTER everyone had read Jolie's article and suddenly there was this wealth of information and firsthand stories that I hadn't seen before. It was much easier for me to find stories and blogs of girls MY AGE who has gone through the same surgery and explicitly stated how much they didn't regret the decision.
- I received my results in a meeting with my genetic counselor. Honestly, at the time I had no idea what it meant at that time and couldn't understand why the counselor seemed so serious. I'd just lost my dad, so I guess it didn't really sink in. …I really had no idea of the gravity of the

situation until mid-2013 when Angelina Jolie announced her PBM – I thought wow, that's serious. So even though it was in the back of my mind for seven years or so, and I suffered a lot of fear from the uncertainty and my own ignorance, I only began to take it seriously after being catapulted into the BRCA world by a celebrity. Even though I have seen all the women on my dad's side suffer from HBOC syndrome and all but 2 of the have died because of it.

Insurance.

- I decided to this surgery NOW because I have found myself in a very specific position: I had just moved back to the states and was unemployed; I was under the age of 26 and could join my parents' health insurance plan; and would be living in a house with people who had already hone through the aftercare process with my mom & would know what to expect. Delaying my job search for a year (my biggest hang up) seemed a lot more logical to me under those circumstances.

- I had different priorities in my early 20s even before testing because of my family history, e.g. having a job that provided health insurance. Obamacare would have changed that for me if I was younger because I could have stayed on my parents' plan.

Screening. Participants mentioned topics related to screening 128 times (n = 128)**.** Main themes and the number of times participants reported specific details about screening are shown in Table 2.9.

Unfavorable Screening. For one, "It was really uncomfortable because I had dense breast tissues since I was young and hadn't had any kids yet." Another reported, "The mammogram was awful. The lady kept squishing them even after I said I couldn't take it anymore. The 'pads' were of no help. The ultrasound was completely okay. It felt like a massage made specifically for my boobies."

Others wrote, "I have not enjoyed any of them [surveillance]. I find it brings me down and reminds me of being ill when I'm not" and "Anxiety could be the best way to describe my experience to all the tests I went through. I was constantly worried they were going to find something and I would have to go through surgery, chemo, and radiation like my mom."

> - It was a very stressful experience to even consider that I may have cancer. The needle biopsy itself was invasive and painful. In fact, this was probably the most traumatic experience I have had in dealing with my *BRCA2* mutation (and I underwent a double mastectomy at age 22!).

Surgical Decisions. Responses were evaluated for themes using Content Analysis. The number of times participants made comments related to surgical decisions were counted and summarized in Table 2.7. Most participants mentioned surgical choices they either have made or will make in the future (n = 100). The most commonly mentioned surgery was prophylactic bilateral mastectomy (n = 40). Other trends in the data included waiting until after having a family (n = 29), having reconstruction (n = 18), having a oophrectomy in the future (n = 18), and being shaped by watching other family members experience cancer (n = 20). Fourteen people had misconceptions about surgical choices, with most noting either that they had or hoped to have a hysterectomy (n = 11).

Table 2.7. Surgical choices and decisions made.

Surgical Decisions (*n* = 234)	n
Surgery choices made or will make	100
Mentioned that they had prophylactic bilateral mastectomy (ages noted: 21, 22, 25, 28, 37)	40
Nipple sparing	*2*
Bilateral mastectomy (following unilateral cancer diagnosis)	2
Reconstruction	18
Expanders then implants	*2*
Expanders, implants, and nipple reconstruction	*2*
Direct to implants (age 28)	*2*
Tissue expanders then saline implants (age 28)	*1*
Silicone implants (age 29)	*1*
Will have prophylactic bilateral Salpingo oopherectomy and/or hysterectomy (Ages: after 30, max 35, around 35, at 38, around 38, at 38, by 40, at 40, or after 45)	18
Will have prophylactic bilateral mastectomy (by 30, at 30, around 35-40, or before 40)	12
Non-nipple or skin sparing	*2*
DEIP flap reconstruction (age 27)	*2*
Had bilateral Salpingo oopherectomy and/or hysterectomy (ages: 26, 27, 29, and 38)	9
Davinci hysterectomy	*1*
No reconstruction	1
Surgery after children	29
Already had / plan to have mastectomy, wait for oopherectomy	23
Wait for both mastectomy and oopherectomy	6
Desire to breastfeed children	*3*
Surgery is "the best option"	22
It was worth it	4
I am young so recovery will be easier and faster	3
Best option because no longer wanted to live life waiting for cancer	3
The only option for me personally	2
Family history shaping surgical choices	20
Surgery because witnessed family members fight with cancer	11
Do not want to children to have to watch me fight cancer	5
Mastectomy and later oopherectomy because family members have chosen this	4
Surgery "to prevent cancer"	16
Misconceptions about surgeries warranted because of a *BRCA1/2* mutation	14
Noted hysterectomy or removal of uterus	11
Only fallopian tube removal	1
Oopherectomy not an option until after 45	1
Noted cervical dysplasia and abnormal pap smears as motivation to undergo surgery	1
Bilateral mastectomy to alleviate worry	13
Set mind at ease / freedom from stress	8
Reduce stress caused by surveillance and the potential to find cancer	5
Unforeseen issues with surgery plans	6
Surgical course was more painful and complicated than expected	4
Skin necrosis and low blood flow following prophylactic bilateral mastectomy	*3*
Problems with infection due to the skin not healing	*2*
Complications with drains	*1*
Lumpectomy led to more serious surgery due to positive nodes	*1*
Frustration over length of time it took have surgeries completed	2
Surgery to gain control over cancer	5
Early menopause concerns	4
Concern that bilateral mastectomy will affect sex life	3
Insurance dictating timing of surgery	2

Mastectomy Prior to Childbearing.

- I originally planned to have a bilateral mastectomy after having children. I finally came to a point where I was not willing to take the risk of getting pregnant and going without effective surveillance for 9 months, while my body went through hormonal changes.

Family Experiences Impacting Surgical Choices.

- At age 28, I had a [PBM]. My mom was only 29 when she was diagnosed with breast cancer, and she passed away at age 35 when I was only 6 years old. I have two of my own young children and I vowed to do everything I could to avoid this happening to us. It has been a long road, recovery hasn't been easy, but I haven't for a minute regretted it.

- I had prophylactic bilateral mastectomy when I was 22. I watched my mother get diagnosed and fight breast cancer twice and I was determined to stop cancer before it got me. I wanted to prevent my children and family from going through what I went through watching my mom.

Healthcare and Support. Participants mentioned topics related to healthcare or support 115 times (n = 115)**.** Main themes and the number of times participants reported specific details about healthcare support are shown in Table 2.10.

Major themes among participant responses included participants feeling either supported by their health care providers (n = 61) or frustrated by them (n = 16). Most participants recommended seeing a genetic counselor (n = 14). Other women recommended finding other people who are also *BRCA1* or *BRCA2* positive (n = 10).

Table 2.9. Screening plans and thoughts on the options available.

Screening (*n* = 128)	n
Screening protocol in place	92
Yearly MRIs	12
Every six months, CA-125 and transvaginal ultrasound	11
Breast ultrasound, mammogram, and MRI (alternating)	11
Every six months mammogram or MRI (alternating)	9
Breast ultrasound and MRI (alternating)	8
Yearly Mammograms (starting at ages 21, 35 30, or after testing positive)	6
Yearly	5
Birth control as a way to reduce the risk for ovarian cancer	5
Undergoing screening for melanoma / seeing a dermatologist	4
Concerns about pancreatic cancer because of family history	4
Biannual breast MRI (post mastectomy)	4
Yearly mammograms and MRIs. (start at age 21)	3
Yearly mammograms and breast ultrasounds	3
Yearly CA-125	3
UK has no ovarian cancer screening options	2
Biannual CA-125	1
Every three months CA-125 / every six months pelvic ultrasound	1
Surveillance causing anxiety and stress	16
Toll on mental state	5
Stress over length of time an MRI takes	4
MRI is nerve-racking	2
Anxiety waiting for results after screening	2
Surveillance as frightening because of family experiences with cancer	1
Misconceptions about screening warranted because of a *BRCA1/2* mutation	7
Pap smears as screening	4
Concerns for cervical cancer screening because in family history	2
Need for colonoscopy	1
Mammogram as painful or uncomfortable	4
Dense breasts	2
Large breasts	1
Core needle biopsies or fine needle aspirations were painful	3
Rechecks are more of a hassle or inconvenience	2
Frustration obtaining insurance coverage for screening because of age	2
Surveillance causing no pain or stress	2

Supportive and Beneficial Providers. "I think all experiences have been positive thus far [with healthcare providers]. Everyone seems either very knowledgeable, or they defer to someone else who is better acquainted with BRCA." Another commented that, "Karen Brooks was amazing. She fully explained everything."

Table 2.10. Healthcare and support related to being *BRCA1/2* positive.

Healthcare and Support (*n* = 115)	n
Providers made me feel supported through decision making	61
Empowered and supported my decision	10
Explained everything so I understood	9
Listened to my questions and concerns	9
Helpful	8
Empathy and compassion	6
Explained the science	4
Continued support over time / calls to see how I am doing	4
Phone number that I can call at any time	3
Respectful of what I did/did not know and explained when necessary	2
Persistence and will not give up trying to help	2
Being open with discussions	2
Made me feel in control over my medical decisions	1
Treated me like an adult	1
Feeling frustrated by healthcare providers	16
Unsympathetic or unsupportive care providers	6
Lack of BRCA knowledge	3
Having to advocate for own care / no one taking initiative	2
Lack of clarity about when surgeries can take place	2
Getting appropriate care during recovery from surgery	1
Not trying to find answers to questions asked	1
Not remembering who you are after repeat visits	1
Recommend seeing a genetic counselor	14
Supportive	6
Helpful and accurate information	4
Recommend genetic counseling before result disclosure	4
Recommend finding other young mutation carriers	10
Become part of the sisterhood to find others like you	7
You are never alone	3
Only positive experiences related to BRCA and clinical care	8
Genetic counselors provided no new information	6
Reviewed all options with a physician prior to genetic counseling	2
Already knew a lot about BRCA from family experience	2
Would not recommend or was frustrated by genetic counseling	2

Trouble Receiving Age-Appropriate Care from Providers.

- One oncologist I saw recommended that I see a therapist, after I came to her office expressing concerns about changes in my body. I think she used 'overreacting' in the conversation, even though she knew my risk. It was incredibly frustrating to see a doctor dismiss concerns, when so much of surveillance is about doing self-breast exams and being aware of your own

body. Many oncologists deal with older women who already have breast cancer and really have no idea what it's like to live with BRCA.

- A sonographer I recently encountered made it clear that she did not agree with genetic testing or the use of surveillance techniques for BRCA+ carriers. It was a very awkward 20 minutes where I was interrogated until she felt I had justified having an ultrasound.

Trouble Receiving Age-Appropriate Care from Genetic Counselors.

- It was basically useless. …They had no clue how to talk to someone who wasn't 40. They guy spent a bunch of time explaining what kind of surgeon I needed to find to do my oopherectomy. Obviously, you need to provide information about what kind of prevention to do down the line, but they gave me really almost no sense of what I should be doing at 24. I wouldn't recommend genetic counseling to anyone.

- A second wrote: The only really bad experience I had was with a genetics counselor. He was condescending and really seemed to have no idea how to talk to me, or to formulate advice from someone in her mid-twenties.

Recommend Genetic Counseling. Fourteen participants commented favorably on genetic counseling and mentioned that they would recommend it to others. One wrote, "Just having them spending the time addressing your questions and concerns. The most important thing is to GET TO COUNSELING! This is a major decision mentally, emotionally, and physically and you need to be prepared." For another, "The genetic

counselor provided me the most helpful information and guidance in terms of risk percentages, risk reduction methods, and a preventative plan of action."

- Definitely recommend genetic counseling with someone who specializes in BRCA. There is so much misinformation on there and judgment from people or medical professionals that aren't really in the know. It's super important to talk about your specific situation and not just go based off general guidelines. There is no one size fits all when it comes to things like this.

- It was refreshing to speak to someone who was both impartial without a personal stance (that she demonstrated in her professional capacity at least) and who did not require explanations but was able to provide them. The information she provided was helpful for follow up conversations that I had with my family physician regarding my screening and medical decisions.

Genetic Counseling Provided no New Information.

- I did receive genetic counseling prior to my BRCA test. Because my mother had tested positive a few years before and I had done a ton of research. I knew most of what the CGC shared with me. None of it was truly new information but I think genetic counseling is immensely valuable for those who are not as aware of what it means to be BRCA positive.

- I had known that I may have *BRCA2* since I was ten years old and had grown up discussing the issue with my mom, who had had breast cancer

when she was 36. As a result, I didn't get any new information from genetic counseling.

- [A Genetic counselor] wanted to refer me for counselling as she felt 'I wasn't coping with the death' of my aunt (she dies from pancreatic cancer with suspected link to the *BRCA1* gene). My aunt had only died a month prior to this session and I felt (and still feel) that I was coping well and simply working my way through the grieving process.

Altered timeline for life. Participants indicated that they felt a need to speed up (n = 40) or slow down (n = 6) their timeline for life events after learning that they carried a *BRCA1/2* gene mutation (Figure 2.4). Participants reported feeling like they had to speed up having children (n = 18) or finding a husband (n = 9).

Life Timeline as Either Delayed or Rushed. One wrote, "It makes me fell like I need to get married sooner and find a partner who would not mind if I have to have my breasts and reproductive organs removed."

- Although I have been married for over four years, my husband and I have not immediate plans for children. My physicians have been questioning if this is something we would want to consider more immediately because of how it may affect future options. While I do not want this to be the reason behind our family planning, I have found it to be creating pressure to speed up our conversations and plans for the future. Simultaneously, as we do not feel that we wish to start a family at this point, I feel that the two feelings are creating tension.

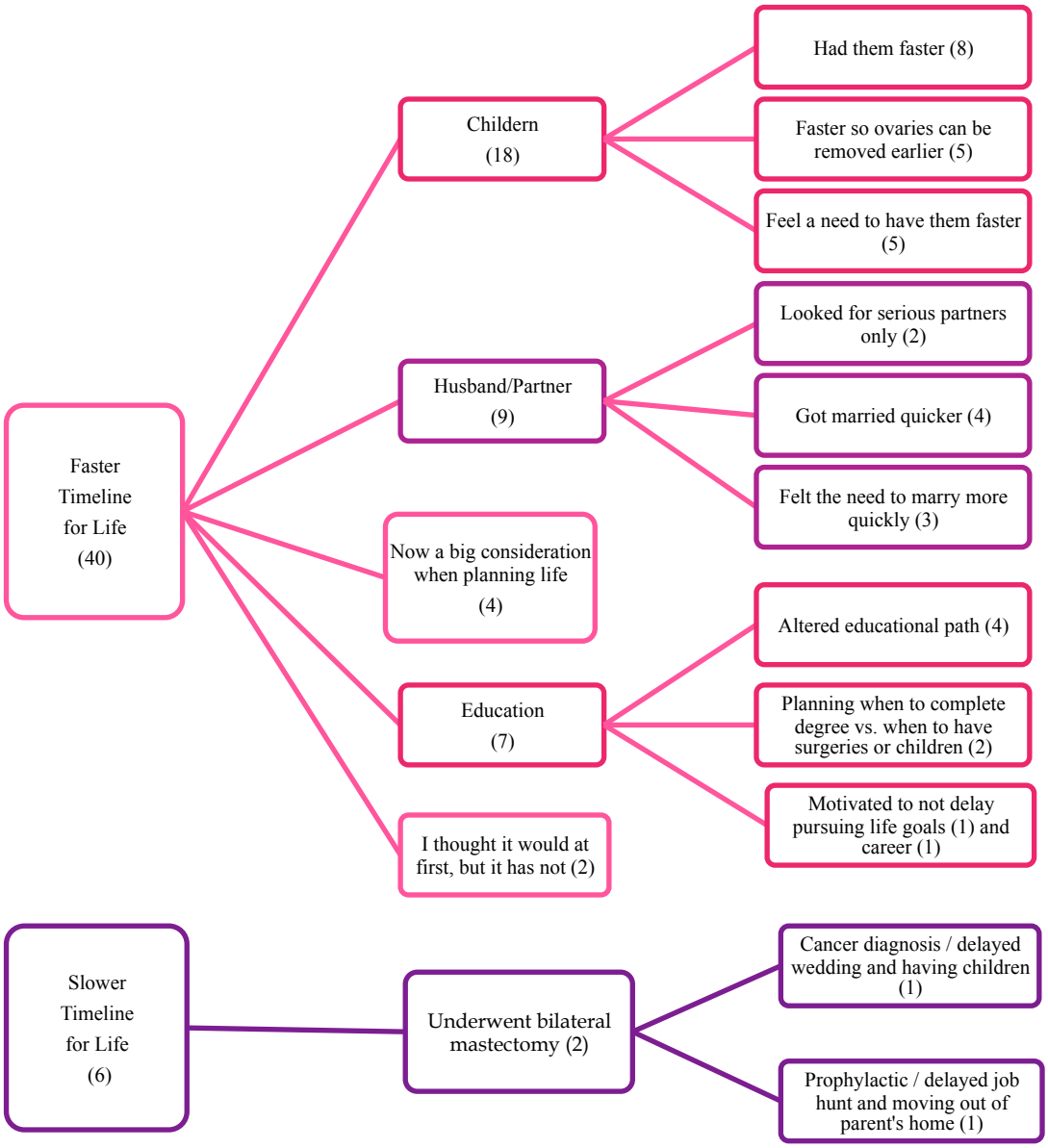

*Figure 2.4. P*articipants' *BRCA1/2* status as pressure to speed up or slow down life.

Delayed Timeline for Life.

- Testing positive didn't affect the timeline for my life, however, my cancer diagnosis did. I had to defer my education to undergo treatments …children would have to wait a year after finishing chemotherapy to ensure the drugs are out of my body.

Different Pressure than Older BRCA Carriers Feel.

- It has affected my life timeline in terms of being more aware of decisions I need to take…. as I am still very young, I don't feel under pressure by the remaining time that I have. (As opposed to women I have met who are approaching 40, single, wanting to have children, but needing to remove their ovaries soon). I hope I will not have to face these challenges.

Views on Family Planning Options. Participants mentioned or discussed family planning options seventy-three times (n = 73). Participants were asked to describe what they know or their thoughts on in-Vitro Fertilization (IVF), Preimplantation Genetic Diagnosis (PGD), adoption, or surrogacy. Participants were asked if they were pursing family planning yet or what they thought there future plans may look like (Figure 2.5).

Major themes included families being informed of PGD and IVF but not wanting to use the technology (n = 17). Others had been informed about PGD and IVF and would consider using it (n = 16). Other participants considered or planed to adopting in the future (n = 13) or elected not to have children (n = 11).

Desire to Have Children the Natural Way. One wrote, "I still plan to get married and having children. With the technology that has come out I can only hope my children have the options plus more if they were faced with the BRCA gene."

- I still want kids gene or no gene. I feel like it's not fair that I shouldn't have kids or a family just because of the gene. I am just like everyone else and there are tons of other people having kids with other genes and issues. This is just my something and it shouldn't stop me from leading a fulfilling life.

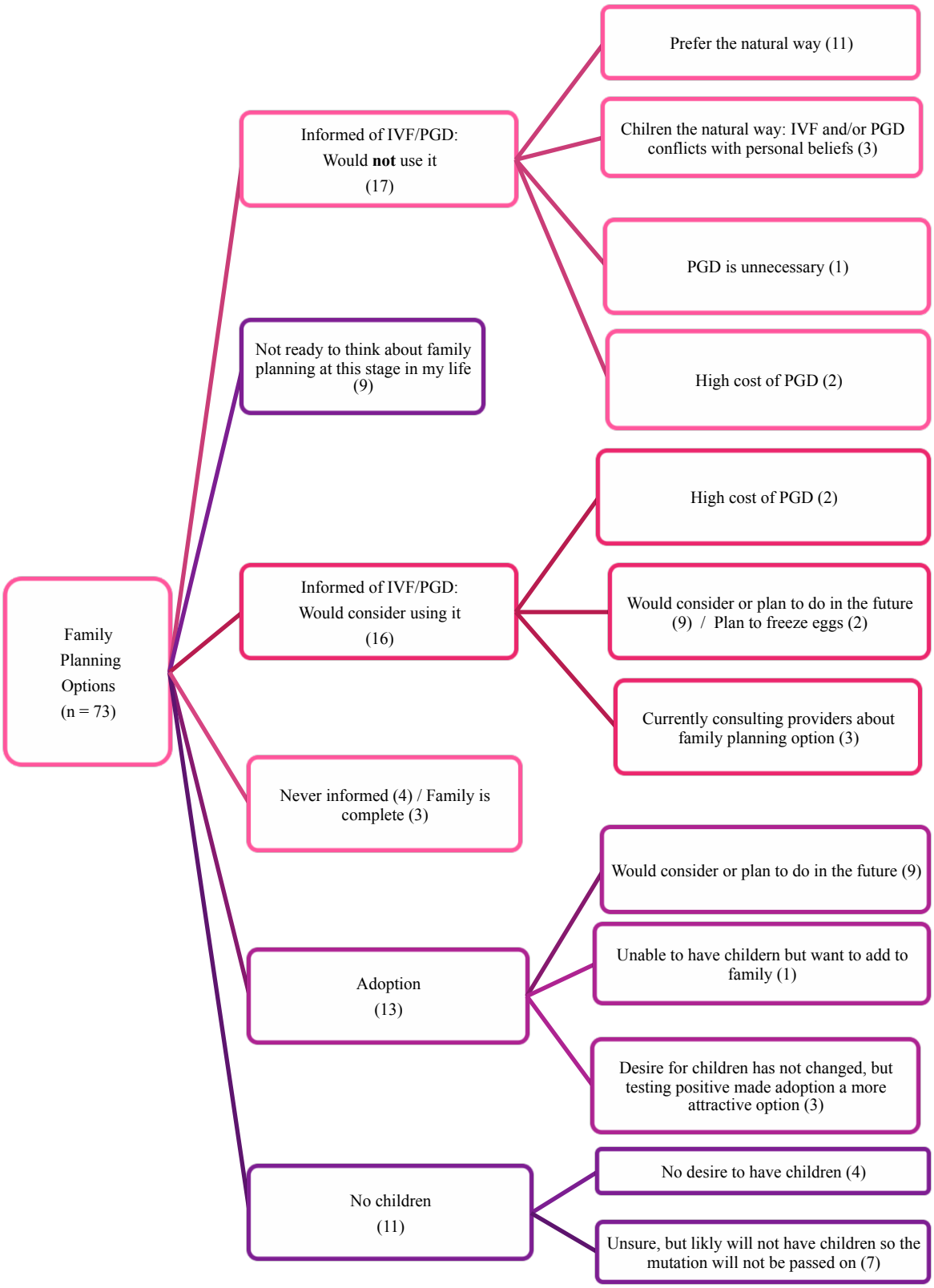

Figure 2.5. Participants' opinions on family planning options.

Would not use PGD. "It is expensive… PGD seems vaguely morally dubious." Another responded: I was advised of the option to use IVF to select embryos without the mutation. This option goes a little too close to playing with the powers of procreation for me. …I know about my mutation and can help my daughters and sons know if they have a mutation as well and lead long healthy productive lives just like I am. I don't see it as a death sentence and don't see like living a life with a BRCA mutation would be unfair to them or to me.

High Cost of IVF and/or PGD. One replied, "If I am in a fortunate financial position, I will consider PDG." Another felt, "Pre-implantation for BRCA is silly, it is not a childhood affecting disorder, it's preventable 100% if you follow guidelines and pay attention to your body. The IVF and other options are very nice, expensive but interesting." A third wrote, "I considered PGD. Then I discovered it costs 3-4 times as much as regular IVF."

Would Consider PDG. One explained that, "I have always wanted children. This changes how I am going to go about making a family."

- I've thought about adoption ONLY because I don't know if I can have kids. Seeing as how I have Endometriosis and miscarried several times, not sure if my body can handle having my own kids. Of course I'll try IVF, but if that doesn't work then I definitely want to adopt.

- A negative [of undergoing genetic testing] is that I ill consider in-vitro fertilization and *BRCA2* testing my embryos when I'm ready to have

children. I never would've thought of reproduction so clinically before, especially since I wouldn't exist if my parents had that option.

Would Consider Adoption. For felt that, for her, "Whether they are of my womb or adopted I will be there mother, there is no changing that."

- PGD and IVF were presented to my husband and I, but because of our values and beliefs we will not take part in any of those options. Adoption is an option we have considered not only because of the positive *BRCA1* results, but also because I may not be able to conceive after receiving chemotherapy.

- As far as the future of my family goes, I never had a strong desire to have children. As a carrier of this gene [mutation], I have less of a desire to bear my own children. It has made me think differently about the idea of adoption…or just having lots of awesome pets.

- I have never been very interested in having children, but having this gene mutation that I could pass down to any offspring makes me not want to have children. It almost makes me feel like it is my duty not to have children, excluding the possibility of that life experience.

- When I first got my BRCA results, I wanted to have kids before age 30, because experts say it reduces BC [breast cancer] risk. Now that I've had cancer, though, I'm not sure I want to have kids at all. I have a 50% chance of passing on the on and I'm not sure I want to do that and risk a child of mine going through cancer like I did.

2.5 Discussion

2.5.1 Participants.

Despite efforts to obtain a more diverse study sample in terms of sex, race/ethnicity, education and income, the participant demographics in this study were quite similar to the demographics achieved in related studies (Hoskins & Werner-Lin, 2012; Werner-Lin et al., 2012). Lack of variability in terms of race or ethnicity, despite efforts to reach out to race or ethnicity-specific BRCA Facebook groups, was not ideal yet not unexpected. Typically, African Americans as well as other minority groups are hesitant to take part in research (Sherman, Miller, Shaw, Cavanagh & Sheinfield-Gorin, 2014). However, the study expanded to other countries outside of the United States and reached a large number of participants. Therefore, one of the key aims to increase sample size was achieved.

2.5.2. Satisfaction. We hypothesized that individuals who obtained their positive genetic test result by age 25 would be satisfied with their decision to undergo genetic testing, but somewhat less satisfied then those who were older than age 25 at the time of the study. This was predicted since young mutation carriers are less familiar with making autonomous decisions (Arnett, 2000) and have less mental capacity to examine each facet and consequence of the decisions at hand (Ford et al., 1998; Steinberg, 2005). We found that all participants, regardless of age, were highly satisfied with their decision to undergo genetic testing. Thus, age did not have an effect on testing satisfaction as we anticipated. This was consistent with previous data collected (Klemp et al., 2005). This trend could be due to the fact that knowing in advance enabled young *BRCA1/2* mutation carriers time to plan for the future and develop an optimistic view for the future and

future research (Donnelly et al., 2013). For a number of participants, knowing their *BRCA1/2* mutation status did bring them optimism and hope for the future (n = 29) and enabled them to regain their sense of control over cancer (n =5).

2.5.3. Recommendations for care providers and genetic professionals. While every patient will be different and guide the way their care is provided, based on the data we collected, there are five recommendations we propose to guide counseling for *BRCA1/2* carriers under the age of 25. Care provision recommendations for young *BRCA1/2* carriers include the five Rs.

- *Right Reproductive Organs:* Discuss which female reproductive organs are and are not at risk (i.e., NOT cervical cancer)
- *Risk Reducing Mastectomy:* Primarily discuss prophylactic bilateral mastectomy, with less time devoted to discussing oopherectomy
- *Risk Figures:* Discuss age-specific risk figures and how lifetime empiric cancer risk figures are accurate for them, and hopefully we will have even more mutation-specific numbers in future

  (i.e., most *BRCA1/2* positive women have a significant cancer family history)
- *Reproductive Options:* Cover possible reproductive options

  (Not all pregnancies are planned and basic knowledge can be important for future or financial planning)
- *Resource for Future:* Be an educated future resource

  (Their needs and questions will change over time; be there to answer them.)

These guidelines could benefit any health care professionals who work with mutation carriers under age 25. However, both data from this study (n = 23) and previous research (Hoskins et al., 2014; Ormondroyd et al., 2012) emphasize the need for trained genetics professionals, such as genetic counselors, who fully understand BRCA. Similar to Hoskins et al., *BRCA1/2* carriers under age 25 in this study placed value providers who made them feel empowered (n = 10), had clear explanations (n = 9), listened to their questions (n = 9), were empathic (n = 6), explained the science (n = 4), and treated them like adults (n = 1) (2014). Participants expressed frustration when healthcare providers were unsupportive (n = 6) or lacked knowledge about BRCA (n = 3) and the surgery guidelines (n = 2). These findings emphasized the need for better care and more appropriate referrals to genetic professionals for mutation positive emerging adults.

While genetic professionals were reported to provide excellent guidance and care (n = 14), assuring that the care provided is relevant for this age group is critical and one of the bases for our recommendations presented. The topic of relevant care for women under age 25 is two-fold. First, our study identified a need for clear guidelines regarding screening and surgery. Conflicts regarding the best care strategies for *BRCA1/2* positive women under the age of 25 emerged from the data (n = 12). This confirmed what previous researchers found (Hoskins et al., 2014). We found participants were using over 16 different types and combinations of screening strategies. This lack of uniformity regarding a surveillance protocol again supports the idea that a set guideline is necessary in providing adequate care for those under age 25.

Second, our study identified a need for a unique counseling and care strategy for young mutation carriers. Participants wrote that providers were misinformed or

unfamiliar with *BRCA* (*n* = 4) and that genetic counselors specifically did not know what types of recommendations are fitting for such young carriers (*n* = 3). This led to participant frustration with genetics professionals who provided them no new information related to BRCA (*n* = 6). This data indicated that future steps are necessary to assure *BRCA1/2* positive emerging adults are receiving care that is relevant at their young age.

Right Reproductive Organs. The data showed that participants had many misconceptions regarding both surgeries and screening related to *BRCA1/2*. This is consistent with previous literature (Patenaude et al., 2013). Of primary concern were women discussing cervical cancer, which is a type of cancer unrelated to *BRCA1/2* gene mutations (Clark & Domchek, 2011). Participants expressed concerns about cervical cancer in their family history (*n* = 2) or personal history (*n* = 1) and that they were utilizing Pap smears as screening for ovarian cancer (*n* = 4). This was consistent with pervious works that found young daughters of known *BRCA1/2* mutation carriers also mentioned the need for increased pap smears and worry over cervical cancer risk (Patenaude et al., 2013).

One participant in particular noted that her motivation for undergoing prophylactic bilateral oopherectomy, besides being BRCA positive, was because she had cervical dysplasia and had never had a normal pap smear. An additional participant expressed the need for early colonoscopies as well, which is yet another cancer not typically associated with *BRCA1/2* gene mutations (Clark & Domchek, 2011). There needs to be increased patient education in this area. It is unacceptable that women may be making life-altering or life-risking choices based on inaccurate information surrounding topics such as cervical cancer risk.

Other participant misconceptions centered on the topic of prophylactic surgery. Eleven participants noted that they either had or planned to have hysterectomy or specifically their uterus removed. Most did not mention the need for ovary removal. This finding could have been because they simply failed to mention that their ovaries and fallopian tubes were removed or that they intended to have them removed along with their uterus. However, it is still concerning that participants are unfamiliar with the correct surgical terms given that they will likely have or already have had surgeries.

Some participants specifically mentioned that they were to have their uterus removed related to their *BRCA1/2* positive status. While there may be a slight increased risk for uterine cancer with *BRCA1* mutation carriers in particular, the key reproductive organs at highest risk for cancer are still the unmentioned ovaries and fallopian tubes (Segev et al., 2013). Another participant expressed that she hoped to have only her fallopian tubes removed and not her ovaries, despite the fact that the greatest risk for ovarian cancer (Clark & Domchek, 2011).

Another responded that having an oopherectomy was not an option until after age 45, despite the fact that she personally desired it sooner. This is an unfortunate misconception, not only since patients should be able to receive the treatments they desire but also because guidelines state that oopherectomy should be considered as early as ages 35 to 40 (Berliner et al., 2013; Clark & Domchek, 2011). This was a new finding related to young *BRCA1/2* carriers. These results clearly demonstrate a need for future education of very young *BRCA1/2* mutation carriers' pertaining to the risks specific to each female reproductive organ.

Risk Reducing Mastectomy. The data showed that all of the women who took part in this study ($N = 63$) had considered prophylactic surgery. The main topic participants discussed was mastectomy ($n = 128$), signifying that this is a critical topic for these very young mutation carriers. Surgery related to the ovaries or uterus was referenced less often ($n = 72$) with over half of the references having to do with participants wanting to wait until after they have children or were past age 30 ($n = 47$). This indicated that, while the topic of oopherectomy is important to be aware of and plan for in the future for these young carriers, it is not of main concern nor will they desire oopherectomy in the near future. Thus, young carriers only need to receive counseling and guidance regarding the salient related to oopherectomy at this age.

Healthcare providers must keep in mind that, unlike older BRCA1/2 mutation carriers, emerging adults have more time to contemplate their screening and surgery options. Carefully attention is needed when providing care for *BRCA1/2* positive emerging adults to assure their autonomy and full understanding of surgical choices (Arnett, 2000; Werner-Lin et al., 2012). Given the almost non-existent risk of ovarian cancer prior to age 30 (Stratton et al., 1999), the topic of oopherectomy is better saved for future healthcare appointments or sessions. The topic of prophylactic mastectomy, on the other hand, is critical.

Risk Figures. We found that many women either overestimated their cancer risk ($n = 16$) or quoted accurate numbers but said that they believe their actual risk for cancer is much higher because of their family history ($n = 16$). We also found that women often overestimated their risk for cancer at their current age, thinking that their current risk for cancer was more similar to the lifetime risks. This overestimation of risk was supported

findings from previous work (Patenaude et al., 2013; Werner-Lin, 2007). Thus, two recommendations were made regarding how risk figures should be discussed with BRCA positive emerging adults. First, review their current age related risk for both breast and ovarian cancer. The likelihood for cancer between the ages of 20 and 24 is very low, and patients need to know and understand this data. It is estimated the *BRCA1/2* carriers have a 0.02% for breast cancer and .001% for ovarian cancer (Antoniou, et al., 2003). However, none of the respondents quoted or mentioned that their current risk for cancer was this low. Thus, this suggests that participants are not being well informed about their risk for cancer diagnosis specific to their young age.

Second, participants need to be reminded that their lifetime risk for cancer is most accurately represented by empiric risk data for *BRAC1/2* mutation carriers. Family history does play an important role in determining who receives genetic testing and who seeks genetic testing. However, once a person is a known carrier, there are cancer risk estimates available for *BRCA1* or *BRCA2*. At this point, patients need to be reminded that, typically, family history does not elevate their risk to be higher than published data. It would be unusual to find a *BRCA1/2* mutation carrier that did not have a substantial family history of cancer due to the automsomal dominant nature of HBOC syndrome and the fact that *de novo* cases are very rare (Clark & Domchek, 2011). Most participants in our study cited extensive family histories of cancer (n = 64).

Common misconceptions included responses such as, “95% because both my grandmothers had breast cancer and my mother had both breast and ovarian cancer and now that I know I have a *BRCA1* gene mutation I know my risk is vary high” or “I had a 98% chance by age 30, it wasn’t an option to not be preventative [and undergo

mastectomy]." Based on this data, we propose that time needs to be spent focusing on known risk numbers and reminding patients that most fellow-carriers have an extensive family history of cancer experiences. An extensive cancer family history does not make the likelihood of cancer absolute (Werner-Lin, 2007).

These are recommendations, and with recommendations there always come exceptions. Thus, it is recommended providers inform patients that we may learn more about more mutation-specific risks in the future. HBOC is a cancer predisposition syndrome and not an absolute cancer-causing disease. Over time, there is hope that we may learn genotype-phenotype correlations do exist and that mutation-specific risk estimates may become available (Donnelly et al., 2013). At the current time, it is our recommendation that providers offer the most up-to-data risk estimate data both for a patient's current age and for over their lifetime, while encouraging patients to stay in touch as new data emerges in the future.

Reproductive Options. A large number of participants expressed their thoughts on reproductive issues and family planning (n = 73). Based on the volume of responses related to reproduction, we concluded that reproductive issues and family planning are highly relevant topic for young mutation carriers. Healthcare providers working with young BRCA carriers will need to assure that they are prepared to discuss these variable topics. This supported the conclusion from previous work with young *BRCA1/2* carriers (Ormondroyd et al., 2012).

A minority of written responses referenced participants not yet being ready to think about family planning at their current life stage. A majority discussed their thoughts on adoption, IVF with or without PGD, having a family the natural way, or that

they desired no children. Many noted that they had at least considered these options and that their desires related to family planning have evolved over time. This data proved that these options are of importance to most women in this age group. Even if they are not ready at the current time to utilize these options, they are apart of most participants' life plans. Based on our data, and the fact that not all pregnancies are planned, we recommend that family planning options are presented to patients. From their, patients can guide to what level of detail each option is discussed, if it at all at the current time. This way the seed is planted so that, in the future, education discussions can take place as the needs of young BRCA carriers evolve.

As anticipated, we also saw participants express that they struggled to find a balance between reducing risk and losing reproductive options (Werner-Lin, 2008). A common trend was planning to delay having surgeries so that they could have a family (n = 29). For most, oopherectomy only was postponed until childbearing was complete. However, participants also brought up the desire to breastfeed their children as a primary reason for why they desired to delay having both mastectomy and oopherectomy. Due to the variability in participant responses and desires, careful and unbiased counseling where these young carriers feel supported in their decisions will be essential to enabling full patient autonomy (Hoskins et al., 2014), as found by previous researchers as well as our study.

Resource for Future. Needs of emerging adults change over time (Arnett, 2000) and thus, so do their needs concerning cancer risk management (Hoskins et al., 2014). We propose that health care providers or genetic counselor that work with these *BRCA1/2* positive emerging adults inform these young carriers that care will extend beyond the

first appointment. Young BRCA carriers who that they are positive prior to age 25 stated that their medical management needs and questions will change over time. This could range anywhere from talking about birth control as their current mode of family planning to discussing the need to plan financially for IVF with or without PGD.

There are no one-size fits all plans. Providing young BRCA carriers the knowledge that they always will have a healthcare or genetics professional available to answer important questions that may arise is key. Young carriers felt the most pleased with their care when they are supported and guided through and beyond emerging adulthood (Arnett, 2000; Hoskins et al., 2012; Hoskins et al., 2014).

2.5.4. Limitations. Individuals were recruited via the Internet, limiting the study to individuals with Internet access that periodically visit BRCA-related websites or follow social media accounts. Participation in this study was not limited to women only. However, the websites and social media used to gather participants primarily cater to a female audience. Since HBOC syndrome affects primarily women, it is typical to obtain few to no male participants in BRCA research. In addition, we did not obtain an ethnically and socioeconomically diverse sample. Thus, findings may not generalize across ethnic groups, to people of different socioeconomic status, or across disease types.

2.5.5. Areas for Future Research. Future research opportunities identified through the course of this study, which could use additional study, are listed below.

- Targeting male participants and minority groups
    - Resources are men or minority groups using as a source of support and guidance, so they can used in future research efforts

    - What these groups' knowledge is concerning their reproductive health and personal cancer risk
- Additional studies focusing on how this group is undergoing surveillance or planning for surgeries, since we saw significant inconsistencies in their established healthcare plans
- Expanding beyond HBOC to determine the counseling needs for different caner disposition syndromes (Cowden syndrome, Lynch Syndrome, etc.)

Beyond BRCA, there is much research that needs to be done on mutation carriers under the age of 25, but with other hereditary cancer predisposition syndromes. There are more young individuals living everyday life knowing that their risk for cancers is significantly higher then that of the general population, without guidelines to inform them how they should be undergoing surveillance (Clark & Domchek, 2011).

2.6 Conclusions

Our study followed in the path of previous, related works on *BRCA1/2* positive emerging adults. We found that participants were very satisfied with their decision to undergo genetic testing, with satisfaction scores of 4.7 out of 5. Satisfaction was high regardless of age, life stability, or whether or not they had received genetic counseling. Much like these previous studies, our research found a lack of guidance, support, and consistent standard of care for mutation carriers who received genetic testing before age 25 (Hoskins & Werner-Lin, 2012; Hoskins et al, 2014). Our study was unique in the fact that we were successful in gathering information from a larger participant pool (N =63).

Based on the data obtained, five suggestions were developed to guide health care providers and genetic professionals to better care for *BRCA1/2* positive emerging adults.

Recommendations, or the five Rs included: (1) *Reproductive Organs:* Discuss which female reproductive organs are and are not at risk (i.e. NOT cervical cancer); (2) *Risk Reducing Mastectomy:* Primarily discuss prophylactic bilateral mastectomy, with less time devoted to discussing oopherectomy; (3) *Risk Figures:* Discuss age-specific risk figures and how lifetime empiric cancer risk figures are accurate for them, and hopefully we will have even more mutation-specific numbers in future (i.e., most *BRCA1/2* positive women have a significant cancer family history); (4) *Reproductive Options:* Cover possible reproductive options (not all pregnancies are planned; basic knowledge can be critical for future or financial planning); and (5) *Resource for Future:* Be an educated future resource (Their needs and questions will change over time, be there to answer them).

Participants in this emerging adult group were variable in their responses, desires, and medical management plans for the future. A clear trend was that participants desired more clear and unbiased care and counseling, where they felt supported. The complexity of HBOC plus the variable lives *BRCA1/2* positive emerging adults face led us to propose a core set of counseling recommendations for young BRCA mutation carriers under age 25. Incorporating the five recommendations is essential to achieving full patient autonomy and unbiased decision facilitation.

Chapter 3. Conclusions

Our study followed in the path of previous, related works on *BRCA1/2* positive emerging adults. Much like these previous studies, our research found a lack of guidance, support, and consistent standard of care for mutation carriers who received genetic testing before age 25 (Hoskins & Werner-Lin, 2012; Hoskins et al., 2014). Our study was unique in the fact that we were successful in gathering information from a larger participant pool (N =63) that was more geographically diverse. We found that participants were very satisfied with their decision to undergo genetic testing, with satisfaction scores of 4.7 out of 5. Satisfaction was high regardless of age, life stability, or whether or not they had received genetic counseling.

Based on the data obtained, five suggestions were developed to guide health care providers and genetic professionals to better care for *BRCA1/2* positive emerging adults. Recommendations, or the five Rs included: (1) *Right Reproductive Organs:* Discuss which female reproductive organs are and are not at risk (i.e. NOT cervical cancer), (2) *Risk Reducing Mastectomy:* Primarily discuss prophylactic bilateral mastectomy, with less time devoted to discussing oopherectomy, (3) *Risk Figures:* Discuss age-specific risk figures and how lifetime empiric cancer risk figures are accurate for them, and hopefully we will have even more mutation-specific numbers in future (i.e. most *BRCA1/2* positive women have a significant cancer family history), (4) *Reproductive Options:* Cover possible reproductive options (Not all pregnancies are planned and basic knowledge can be critical for future or financial planning) (5) *Resource for Future:* Be an educated

future resource (Their needs and questions will change over time, be there to answer them).

Participants in this emerging adult group were variable in their responses, desires, and medical management plans for the future. A clear trend was that participants desired more clear and unbiased care and counseling, where they felt supported. The complexity of HBOC plus the variable lives *BRCA1/2* positive emerging adults face led us to propose a core set of counseling recommendations for young BRCA mutation carriers under age 25. Incorporating the five recommendations is essential to achieving full patient autonomy and unbiased decision facilitation.

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Appendix A: List of Social Media Sources Contacted and/or Utilized

Facebook	
African American Breast Cancer Research Study	Just Ask! About Hereditary Breast and Ovarian Cancer (HBOC)
BRCA1	LA FORCE: Facing Our Risk Empowered
BRCA1 or *BRCA2* Genetic Ovarian & Breast Cancer Gene	Let's Free Our BRCA Data!!
BRCA1/BRCA2+ UK	Male Breast Cancer
BRCA Advanced 101 & 102	Male Breast Cancer Awareness
BRCA Brotherhood	Men Against Breast Cancer
BRCA Commons	Power of Pink! Foundation
BRCA Sisterhood	P.O.P.! Power of Pink Foundation
BRCA Sisterhood Canada	SHARE Cancer Support
BRCA Umbrella - The Breast & Ovarian Cancer Gene & You	Sisters Network Inc.
BRCA Gene Awareness Inc.	The Breast Cancer Site
Breast Cancer Campaign	Think Pink Rocks
BRCA Gene Awareness, Inc.	Ulman Cancer Fund for Young Adults
FAMILIES WHO SUPPORT BREAST CANCER SURVIVORS, Inc.	Ulman Cancer Fund's TEAM FIGHT
FORCE: Facing Our Risk of Cancer Empowered	Young Previvors
Hereditary Breast & Ovarian Cancer (HBOC) Montreal	Young Survival Coalition Bulletin Board
HBOC Society	Young Women's Breast Cancer Awareness Foundation (YWBCAF)
Imerman Angels	
Blogs	
BRCA According to Me (ponderingprevivor.blogspot.com)	
BRCA Blog Directory (brcablogdirectory.wordpress.com)	
BRCA Sisterhood Blog (brcasisterhood.wordpress.com/the-sisterhood)	
Breaking BRCA (brcaprevivor.blogspot.com)	
Fitting into my BRCA Genes (fittingintomygenes.blogspot.com)	
My BRCA Blog (brcaandme.blogspot.com)	
My Journey With *BRCA1* (brca1journey.blogspot.com)	
Previving and Thriving: My *BRCA2*+ Journey (previvingandthriving.com)	
PREvivor GENEration (previvorgeneration.com)	
Staying Positive, BRCA Positive (stayingpositivebrca.blogspot.com)	
Wearing My BRCA Genes (youngbrca1.wordpress.com)	
Young and *BRCA1* Positive and High Heals: Making Our Genes Look Good (youngbrca1pos.blogspot.com)	
Other	
Bright Pink (Twitter) (@BeBrightPink)	HBOC Society (Newsletter)
FORCE: Facing Our Risk of Cancer Empowered (Message Boards)	

Appendix B: Templates for Correspondence with Social Media Groups

Email Request for Hosting Research Survey

To Whom It May Concern:

Hello, my name is Sarah King and I am a second year genetic counseling student at the University of South Carolina. My graduate student thesis project is on unaffected BRCA1 and BRCA2 gene mutation carriers under the age of 25. The goal is to see how age affects the risk perception and life choices of these young individuals. This data will be collected via an online survey through Survey Monkey.com. I would love to potentially work with your organization to attempt to reach this population of individuals for my study.

So you all are aware, this thesis project will undergo IRB approval though the University of South Carolina and poses virtually no harm or risk to participants. In addition, I plan publish my research once the project is complete to help more individuals in this young age group in the future.

Reaching enough individuals in this specific age group will be key to obtaining statistically significant results. I think there is a great need for more information, since there is minimal literature, guidelines, or recommendations written for this age group. Therefore, I would sincerely appreciate your support.

What I am hoping is that a letter to potential participants as well as the link to my survey be posted somewhere online. This could include your website, face book page, etc. Wherever you think it would be best would be much appreciated.

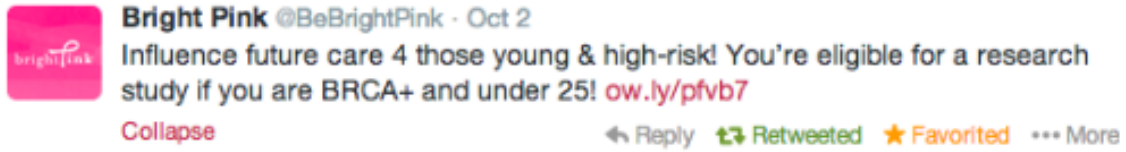
If you would like to me to call you to discuss my project, or if you need further information I would be more then happy to provide that for you. I have a tentative written thesis proposal as well as rough drafts of all survey questions if you would like to look them over.

Thank you so much for your consideration and I look forward to your reply,

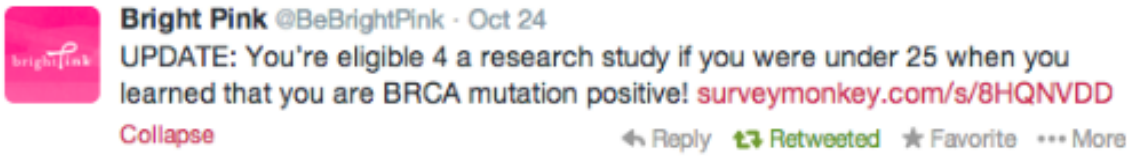
Sarah King

Social Media Templates Inviting Participants with Survey Link: Twitter

Original Post:



Updated Post:



Forum Post for FORCE Website

Hello ladies and possibly some gentleman out there as well -

I would like to invite you to take part in a graduate research study looking at BRCA1 and BRCA2 mutation carriers that are 18 to 24 years old. I am a graduate student in the genetic counseling program at the University of South Carolina School of Medicine. My study looks at the affect of age on life choices and satisfaction with the choice to have genetic testing for young people in this age group.

To take part in the study, you would need to fill-out a survey on the choices that you have made, your overall satisfaction with your choice to have genetic testing, and resources that you use for support. You are eligible for this study if you are 18 to 24 years old and have had genetic testing where you tested positive for a BRCA1 or BRCA2 gene mutation. Both men and women are welcome to take part! I encourage you all to share your stories as much as possible.

The survey looks at themes in the life choices and decisions of individuals in this young age group. If you do not wish to answer a question, please skip that question and continue filling out the rest of the survey. The survey is 40 questions in length, including the questions pertaining to demographics such as age or education level. To participate in this research please click here or follow this link:

http://www.surveymonkey.com/s/8HQNVDD

All responses from the surveys will be kept anonymous and confidential. We only ask for your name and phone number in case you would be interested in providing more information at a later date over the phone. It is not necessary that you provide this information. The results of this study might be published or presented at academic meetings; however, participants will not be identified. Your contact information will not be used for any other purposes besides a follow-up phone interview.

Your participation in this study is voluntary. By completing the study, you are stating that you have read and understand this information. At any time, you may withdraw from the study by not finishing filling out the survey.

Thank you for your time and consideration. Your responses may help health care providers such as genetic counselors better care for young, high-risk people 18 to 24 years of age in the future. If you have any questions about this study, you may contact either myself or my faculty adviser, Karen Brooks, MS, CGC, using the contact information below. If you have any questions about your rights as a study or research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803)777-7095.

Thank you all for your time and support!

Sarah

Sarah E. King, B.A.
Genetic Counselor Candidate
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Karen Brooks, MS, CGC
Faculty Adviser
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(803) 545-5746

Facebook Message for Direct Message

Hello, my name is Sarah King and I am a second year genetic counseling student at the University of South Carolina in the US. My graduate student thesis project is on BRCA1 and BRCA2 gene mutation carriers who tested positive for a BRCA1/2 gene mutation before the age of 25.

The goal is to see how age affects the risk perception and life choices of these young individuals. This data will be collected via an online survey through SurveyMonkey.com. I would love to potentially work with your organization to attempt to reach this population of individuals for my study.

So you all are aware, this thesis project will undergo IRB approval though the University of South Carolina and poses virtually no harm or risk to participants. In addition, I plan publish my research once the project is complete to help more individuals in this young age group in the future.

Reaching enough individuals in this specific age group will be key to obtaining statistically significant results. I think there is a great need for more information, since there is minimal literature, guidelines, or recommendations written for this age group. Therefore, I would sincerely appreciate your support.

What I am hoping is that a link to my survey be posted somewhere online. This could include your website, Facebook page, Twitter account, etc. Wherever you think it would be best would be much appreciated. I have included sample Facebook and Twitter posts for you.

Also, I am not sure if this was the best way to get in contact with your organization, but I believe it will be a good starting point.

Thank you so much for your consideration and I look forward to your reply,

Sarah King

Facebook Message for Facebook Wall

Hello-

My name is Sarah and I am a second year genetic counseling student from South Carolina. I have a special interest in women or men who are young survivors/previvors. I am working on my graduate thesis and am trying to gain as much information about young previvors and their hopes/plans/experiences/etc. I hope you would not mind if I posted the link for my study. Thanks for your consideration!

Also -Some of you may have already seen the link to my study-- It was tweeted by Bright Pink recently!

Study Information:

Help future young, high-risk individuals like you by participating in a research study. We would love to hear your personal story!

We are looking for women or men who discovered that they are BRCA1 or BRCA2 mutation positive before the age of 25 to take our online survey! You qualify for the study if you had genetic testing before your 25th birthday and tested positive. For more details and information or to participate in our research please click here:

http://www.surveymonkey.com/s/8HQNVDD

Social Media Templates Inviting Participants with Survey Link: Facebook

Original Post:

Help future young, high-risk individuals like you by participating in a research study. We would love to hear your personal story!

We are looking for women or men under the age of 25 who have undergone genetic testing and know that they carry a BRCA1 or BRCA2 gene mutation to take our online survey! For more details and information or to participate in our research please click here:http://www.surveymonkey.com/s/8HQNVDD

Thank you all very much for you time, support, and consideration!

Updated Post:

Help future young, high-risk individuals like you by participating in a research study. We would love to hear your personal story!

We are looking for women or men who discovered that they are BRCA1 or BRCA2 mutation positive before the age of 25 to take our online survey! You qualify for the study if you had genetic testing before your 25th birthday and tested positive. For more details and information or to participate in our research please click here: http://www.surveymonkey.com/s/8HQNVDD

Thank you all very much for you time, support, and consideration!

Study Update Post:

We would like to expand this study to anyone who found out that they are BRCA1 or BRCA2 positive before the age of 25. You qualify for the study if had genetic testing before your 25th birthday and tested positive.

Formal Survey Invitation to Participants

Dear Potential Participant:

You are invited to take part in a graduate research study looking at BRCA1 and BRCA2 mutation carriers that received a positive BRCA1/2 genetic test result when they were under the age of 25. I am a graduate student in the genetic counseling program at the University of South Carolina School of Medicine. My study looks at the affect of age on life choices and satisfaction with the choice to have genetic testing for young people in this age group.

To take part in the study, you would need to fill-out a survey on the choices that you have made, your overall satisfaction with your choice to have genetic testing, and resources that you use for support. You are eligible for this study if you were under 25 years of age when you had testing and if you tested positive for a BRCA1 or BRCA2 gene mutation. Both men and women are encouraged to take part!

The survey looks at themes in the life choices and decisions of individuals in this young age group. If you do not wish to answer a question, please skip that question and continue filling out the rest of the survey. The survey is 40 questions in length, including the questions pertaining to demographics such as age or education level. We would love to hear your story!

All responses from the surveys will be kept anonymous and confidential. We only ask for your name and phone number in case you would be interested in providing more information at a later date over the phone. It is not necessary that you provide this information. The results of this study might be published or presented at academic meetings; however, participants will not be identified. Your contact information will not be used for any other purposes besides a follow-up phone interview.

Your participation in this study is voluntary. By completing the study, you are stating that you have read and understand this information. At any time, you may withdraw from the study by not finishing filling out the survey.

Thank you for your time and consideration. Your responses may help health care providers such as genetic counselors better care for young, high-risk people 18 to 24 years of age in the future. If you have any questions about this study, you may contact either myself or my faculty adviser, Karen Brooks, MS, CGC, using the contact information below. If you have any questions about your rights as a study or research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803)777-7095.

Sincerely,

Sarah E. King, B.A.

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Appendix C: Participant Survey

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

1. Invitation to Participate!

Dear Potential Participant:

You are invited to take part in a graduate research study looking at BRCA1 and BRCA2 mutation carriers that received a positive BRCA1/2 genetic test result when they were under the age of 25. I am a graduate student in the genetic counseling program at the University of South Carolina School of Medicine. My study looks at the affect of age on life choices and satisfaction with the choice to have genetic testing for young people in this age group. To take part in the study, you would need to fill-out a survey on the choices that you have made, your overall satisfaction with your choice to have genetic testing, and resources that you use for support. You are eligible for this study if you were under 25 years of age when you had testing and if you tested positive for a BRCA1 or BRCA2 gene mutation. Both men and women are encouraged to take part!

The survey looks at themes in the life choices and decisions of individuals in this young age group. If you do not wish to answer a question, please skip that question and continue filling out the rest of the survey. The survey is 40 questions in length, including the questions pertaining to demographics such as age or education level. We would love to hear your story!

All responses from the surveys will be kept anonymous and confidential. We only ask for your name and phone number in case you would be interested in providing more information at a later date over the phone. It is not necessary that you provide this information. The results of this study might be published or presented at academic meetings; however, participants will not be identified. Your contact information will not be used for any other purposes besides a follow-up phone interview.

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Thank you for your time and consideration. Your responses may help health care providers such as genetic counselors better care for young, high-risk people 18 to 24 years of age in the future. If you have any questions about this study, you may contact either myself or my faculty adviser, Karen Brooks, MS, CGC, using the contact information below. If you have any questions about your rights as a study or research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803)777-7095.

Sincerely,

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I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

Karen.Brooks@uscmed.sc.edu
(803) 545-5746

2. Life Choices, Risk Perception, and Your Health

These questions pertain to health information that is relevant for this study. All information you provide here will be reported anonymously.

Please answer in as much detail as you can! Your help is greatly appreciated.

***1. How old were you when you received your genetic test result?**

Age:

***2. Did you test positive for a BRCA1 or a BRCA2 gene mutation?**

- BRCA1
- BRCA2
- I am unsure / Other

3. Life Choices, Risk Perception, and Your Health

3. Have you ever been diagnosed with cancer?

- Yes
- No

If YES, how old were you and what type of cancer did you have? Please explain.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

4. Have you ever had a mammogram, MRI, breast ultrasound, or breast imaging study?

- ○ Yes
- ○ No

If YES, please describe your experience.

5. Have you ever had an ABNORMAL mammogram, MRI, breast ultrasound, or breast imaging study?

- ○ Yes
- ○ No

If YES, please describe the outcome breifly.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

6. Have you ever had a normal or abnormal tissue biopsy?

- ○ Yes
- ○ No

If YES, please explain the type of tissue or reason for biopsy. Describe your expereince or the outcome breifly.

4. Life Choices, Risk Perception, and Your Health

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

7. Have you ever had breast surgery? (lumpectomy, masectomy, etc.)

- ○ Yes
- ○ No
- ○ Planned but has not yet taken place

If YES, please explain why and describe your experience.

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I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

8. Have you ever had a surgery related to the ovaries or other female reproductive organs? (Hysterectomy, tubal ligation, conization, bilateral salpingo oophorectomy, etc.)

- ○ Yes
- ○ No
- ○ N/A (Men)

If YES, please state which and describe your experience.

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I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

9. Have you ever had a surgery or treatment related to the prostate or testicles (A urogenital surgery or treatment)? For example, prostatectomy, radiation, etc.

- ○ Yes
- ○ No
- ○ N/A (Women)

If YES, please explain why and describe your experience.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

10. Have you ever considered having a surgery to reduce your risk of cancer?

- ○ Yes
- ○ No

If YES, please explain why and describe your experience.

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I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

11. Have you ever used any chemopreventive drugs? (Ex: Tamoxifen, Evista, etc.)

- ○ Yes
- ○ No
- ○ N/A

If YES, please describe your expereince and why you decided to use these chemopreventive drugs.

5. Life Choices, Risk Perception, and Your Health

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

12. What is the risk that you will develop cancer at some point in your LIFETIME? For example, you could say: "I believe I have a 5% chance of having cancer in my lifetime because...."

Please enter a percentage (%) between 0 and 100 and explain why you believe this is your risk.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

13. Do you think testing positive for a gene mutation affected the timeline for your life?

For example, do you feel like you need to speed up or slow down some of your plans for the future?

- ○ Yes
- ○ No

Please explain how your plans were changed.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

14. Do you think testing positive for a gene mutation affected your overall life plan? (Ex: desire to have children, marriage, etc?)

- ○ Yes
- ○ No

Please explain how your plans were changed.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

15. Do you currently have a plan in place for your future health care, related to your positive BRCA genetic testing result? (ex: have a medical management plan including starting mammograms at an early age and...)

- ○ Yes
- ○ No

Please explain what your PLAN is for your health care.

16. Describe how you received your positive test result? (Land line phone or cell phone? Where were you, what were you doing at this time, etc?)

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

17. Please rank each of the following based on your CURRENT level of conern or worry. Which of these do you think about rarely? Frequently?

	No Concern/Worry: Think about rarely	Low Concern/Worry: Think about once/twice a month	Mild Concern/Worry: Think about a few days a week	Moderate Concern/Worry: Think about daily	High Concern/Worry: Think About Constantly
Finding a job	○	○	○	○	○
Finding a place to live	○	○	○	○	○
Completing school or duties at work	○	○	○	○	○
Finding a partner or getting married	○	○	○	○	○
Having children or family planning	○	○	○	○	○
Reducing your risk for cancer (surgeries, treatment, etc.)	○	○	○	○	○

6. Life Choices, Risk Perception, and Your Health

18. Have you ever received genetic counseling?

○ Yes

○ No

If No, who delivered your BRCA genetic testing result? Please describe your thoughts on or experience with genetic counseling.

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I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

19. Have you ever considered a risk-reducing surgery?

- ○ Yes
- ○ No

Please describe how you feel about risk-reducing surgical options for you.

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I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

20. Have you been introduced to different types of family planning or reproductive options based on the mutation in your BRCA gene? Examples may include: PGD (preimplantation genetic diagnosis) with IVF, adoption, surrogacy, etc.

- ○ Yes
- ○ No

Please explain what you may have or have not learned about family planning options for you? Have you started thinking about family planning yet?

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21. Based on your experiences, what would you want to tell a high-risk person who has undergone genetic testing but does not yet have their result back? (Someone who may soon be in your shoes)

7. Your Decision to Undergo Genetic Testing

You chose to have genetic testing to see if you were or were not a carrier of a BRCA1 or BRCA2 gene mutation.

Please answer the following questions about YOUR DECISION TO RECEIVE TESTING.

Please indicate to what extent each statement is true for you at this time.

22. I am satisfied that I was adequately informed about the issues important to my decision.

○ strongly disagree ○ disagree ○ neither agree nor disagree ○ agree ○ strongly agree

23. The decision I made was the best decision possible for me personally.

○ strongly disagree ○ disagree ○ neither agree nor disagree ○ agree ○ strongly agree

24. I am satisfied that my decision was consistent with my personal values.

○ strongly disagree ○ disagree ○ neither agree nor disagree ○ agree ○ strongly agree

25. I successfully carried out the decision I made.

○ strongly disagree ○ disagree ○ neither agree nor disagree ○ agree ○ strongly agree

26. I am satisfied that this was my decision to make.

○ strongly disagree ○ disagree ○ neither agree nor disagree ○ agree ○ strongly agree

27. I am satisfied with my decision.

○ strongly disagree ○ disagree ○ neither agree nor disagree ○ agree ○ strongly agree

8. Supportive Resources

28. Rate how beneficial the following heath care SERVICES were for providing SUPPORT and guidance. Which were the most helpful/unhelpful?

	Very helpful	Somewhat helpful	Neither helpful or unhelpful	Somewhat unhelpful	Very unhelpful	N/A
Gynocology / OB/GYN	○	○	○	○	○	○
Genetic Counseling	○	○	○	○	○	○
Nursing	○	○	○	○	○	○
Oncology / Radiology	○	○	○	○	○	○
Plastic Surgery	○	○	○	○	○	○
Psychology / Mental Health Counseling	○	○	○	○	○	○
Other	○	○	○	○	○	○

If OTHER, please specify.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

29. Describe the most helpful/supportive experience that you have had with ANY health care provider or professional related to your positive BRCA1 or BRCA2 test result.

30. Describe the least helpful experience that you have had with ANY health care provider or professional related to your positive BRCA1 or BRCA2 test result. Please explain why you felt unsupported in this experience.

9. Demographics

Thank you so much for helping with this study! You are nearly finished.

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

***31. What is your current age?**

If OTHER, please specify

***32. What is your sex?**

- Female
- Male

***33. Which race/ethnicity best describes you?**

- American Indian or Alaskan Native
- Asian/Pacific Islander
- Black or African American
- Hispanic American
- White/Caucasian
- Other (please specify)

34. What is the highest education level that you have reached in your schooling?

35. What BEST describes your average income?

36. About how many times have you changed your place of residence/where you live in the last 8 years?

Number of residency changes:

37. How may months and/or years have you been with your current partner or spouse?

Number of Months

Number of Years

38. Have you ever been pregnant?

- Yes
- No
- N/A (Males)

I Wish I Had Known This! Choices and Testing Satisfaction for BRCA1/2

39. Do you have any children?

Number of Daughters

Number of Sons

40. Would you be willing to receive a follow-up phone call if we need to clarify any of your responses or ask you any follow-up questions?

Thank you very much for your time and participation!

Please note: If you decide to provide contact information, your first name and phone number will be included with the answers to your questions, making the data no longer anonymous. Your first name and phone number will not be included in any way in the analysis of the data, results, or published works.

Not all participants will receive a follow up phone call. If we receive responses from ~60 participants or more, no follow-up phone calls may be made.

If you receive a follow up phone call: Your first name or phone number will NOT be included in the results or published works without verbal consent from YOU through this follow-up phone call. It is up to you if you would or would not like to include your first name only.

If you now feel as though you would like a health care provider to talk with about any concerns or questions, please consult your current health care providers or find a genetic counselor near you though the NSGC website, at http://www.nsgc.org/findageneticcounselor/tabid/64/default.aspx.

- No, I would not like to recieve a follow-up phone call.
- Yes, I would be happy to recieve a follow-up phone call. Please provide your first name and the best phone number to contact you at, including area code.

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Appendix D: Advice from one *BRCA1/2* Positive Women to Another

Advice from one Young, *BRCA1/2* Positive Woman to Another
I think it is amazing that you can find out. It is stressful but a blessing to know. I would hate to not know and have to go through cancer treatments. If you know, you can plan ahead.
It's better to know. Then you can help prevent.
Befriend your oncologist, hematologist, and your obgyn oncologist. They are your guides on this journey. Research options and ask the doctors many questions. Ask them if they were in your shoes, what would they do.
You don't need to act on the results right away. You can do surveillance but it gets exhausting and the anxiety may get to you- it did for me. You can do the PBM before having children...I felt rushed to have kids and them I did my surgery but may have done it before in hindsight.
I would tell them that knowing is a gift. If you have the mutation, you're probably going to find out at some point anyway. This way it doesn't creep up on you in the prime of your life, and you have options to massively reduce your risk. It's going to be scary and frustrating, and if it's positive your life and even your identity will change. Also, good luck.
It's better to know, because it gives you the power to make healthy choices for yourself and your future. Why let yourself be blindsided down the road when you can have the comfort of knowing you are doing all the best you can for yourself?
It depends on the person.
Just take it a day at a time. Find a support group that works for you. I have a few on Facebook that have truly brought me through all of this. You don't have to be alone, and you aren't. It is hard for friends and family who are not going through it to always understand, so having a group of people who do is major. Also, research, research, research. You need to be your own advocate, and often times you will find yourself educating the health care providers. The more you know, the easier it will be!
Don't fret. It's truly not the end of the world. Think of it as a Godsend because if you DO test positive just remember it doesn't mean you will end up with cancer. And if you do end up with it, you'll have caught it early enough so there won't be much of a problem. Just try to relax, and don't freak out.
I would tell her to make sure she's really ready for what a positive result means. A lot of people test so they can hear it's negative.
Whether it is positive or negative, it is good news. The knowledge gives you amazing power.
Surround yourself with good literature, good people and supportive environment, be true to you, and find out your values, know that it's ok to be scared of the unknown. Don't take no for an answer, you know your body... Better than any dr
Just as much info as possible, so they can go in armed with questions if need be.
It’s not the end of the world and make sure that you know surgery is not the only option it was the only option i was given and I would have tried preventative drugs had i known about them,
I would recommend the FORCE website.
Think about what you will do with the information when you get it
My decisions have always boiled down to: why not now? Is there a reason I should wait until I'm older to get a procedure done? You need to take everything into account when answering those questions. It's not just about the procedure themselves, but also very much where you are in your life at this moment and where you'd like to be in the immediate future. I was in an 'in-between' moment in my life. I had just ended a big part of my life and hadn't yet started something new. Don't feel rushed. Find the right moment.
That there is pros and cons to being positive or negative. You can't listen to what you’re being told is your right option because it is your body, your right and your decision. You also can't let it control your life but now you have to live life to the fullest.
You are not defined by your genes! Find your community and start learning the stories of others who have been through this (I tend to trust FORCE more than I trust Bright Pink). There are some incredible online communities out there that can support you and answer your questions. Here is one of them: https://www.facebook.com/groups/bravebosom/

No matter what the result is, it is not a death sentence - it's a blessing. This test gives you your power back. Power over your health, the power to make decisions, the power to DECIDE how to live your best life. This test doesn't change anything about your health - if you have a mutation, it has been there since the moment you were conceived. BUT, this test DOES change what you can do about it. IF you have a mutation, you get to DECIDE how it will impact your life. You get to protect yourself and your health.
I would say that everyone is different, but in my opinion, it is better to know either way so that you can better prepare for your future instead of wondering and not taking the proper preventative actions, which would only put him/her at a higher risk of developing cancer. It is best to know so that you know what to do about it.
It's not really so big a deal. The risk of getting cancer is high for everyone, but if you get a positive result at least you will be studied regularly. Also mastectomy is not as bad as it might seem, at least I am very happy of my decision.
Don't take this as an end to your life look at it as a wake up call and do what you want to do and what you feel is right for you
Knowledge is power and finding out one way or another is a positive and brave thing to do. You are taking control of your future and you can decide what you want to do going forward. If you do test positive for a faulty gene, do not rush in to making any decisions about preventative surgery. If you do decide to undergo preventative surgery, do as much research as possible before seeing a surgeon.
Don't feel pressurized by the media or medical professionals into making massive decisions about your body. Only you, with support from your nearest and dearest, can decide what is right for you.
No matter what the result is you may still get cancer or you may never get cancer. Look at the result as a tool to leading a healthier life. If it is positive, it really is so much better to know so if you ever do get cancer it can be caught at an early stage
That person would strongly benefit from a little company & probably some healthy distractions. Jumping into education regarding their options would only overwhelm them in the meanwhile.
Not to worry, and to only see a positive test as knowing as early as possible so you can get on the track to being aware of the risks
One day at a time
I would tell them that they will be the same person after their results as they are before and their beliefs and priorities should stay pretty much the same. The timeline of your life and the things you need to do to stay healthy may change, but your life is no less beautiful after the results than before. And knowing about a mutation is actually a beautiful gift because it changes you into a proactive person who can have a dramatic impact on your own future instead of just waiting around and wondering. It helps you make the most of your life in a way that most people can't.
No matter your result, you are healthy. I have found this to be an important thought to return to as the screening process, the options, and the risks laid out can easily paint a picture and feeling of a health crisis. Having a support network is very important, as is being able to take time for yourself away from the business, the tests, and sometimes other people, as you need.
I would say to just take the first month or two and let the results sink in. You will feel a lot of feelings and be very overwhelmed for a while, but don't make any decisions right then because you won't have enough distance to be objective. Once the dust settles, then you should really start researching your options. Go to a conference (like the Facing Our Risk of Cancer Empowered conference), reach out to support groups to talk to other women, and talk to a lot of doctors. You may not feel like you have time, but you also shouldn't make any decisions without being completely informed first.
I recommend that everyone at genetic risk for HBOC meet with a genetic counselor, develop a family tree, and a list of family cancers and the ages diagnosed. I also recommend that they hear the results in person, not over the phone or via mail as I've heard some folks have.
That everything will be ok and that even though positive result sucks and will probably turn your life upside down, it's wonderful that this option is available. Being able to take preventative measures is better than having to fight cancer and possibly lose your life.
I would tell them to breathe. Knowledge is power and knowing is such a blessing. Many people will never know that they will get cancer. This is such a blessing to those who can know and do something about it.
Don't start researching until you get your result, you will just freak your self out!
I would tell them that I know how nerve-wracking it can be to wait, and that the wait will be over soon! I'd encourage them to do research if they are comfortable doing so, and consider the choices that they might make if they test positive. I'd also encourage anyone who is considering testing or waiting for results to find a community of people in the same boat - FORCE, Bright Pink's PinkPal program, Facebook groups like BRCA Sisterhood or Young Previvors are all invaluable options!
That the mutation is not a death sentence, nor should it have a negative impact on your life. Knowing about the mutation is knowledge, and knowledge is such a powerful thing. It gives us the power to prevent cancer before it occurs, or at that very least significantly lower our risks. It allows us to be in control of our health, and life.

Too not worry too much until you get the result back. Once you do have the results take your time before you do anything and weigh all your options.
It's a blessing to know. You are the lucky ones who have precursors and you have the ability to set your future and be proactive if you are positive
Relax. You tested for a reason. You must have a family history. Your surveillance will be high even if you are negative. If you are positive. Welcome to the sisterhood, it won't always be easy, it won't always be cheap, and you won't always understand it. But you are never along, you will din courage you never knew that you had. You will have a chance to survive. My positive is a positive. I am positive and I know how to live the rest of my life now.
Try to keep your routine and carry on. You can't dwell on it, it is what it is, you were born with it and it has been apart of you for X amount of years. Get educated and know your options and choices. FIND THE BEST DOCTORS!!
No matter your result you are still you. Even if you end up positive you can still have a normal life.
To take things slowly and take time to process everything. To think about what you will do one way or another. But not to freak out... Medicine is amazing and there are so many bugged things in life than BRCA.
It is normal to be concerned, worried and anxious to get results back. Bring someone to the appointment if you can to help take notes or just to have a second set of ears, sometimes you don't remember everything when you are given life altering news. Think about the different options prior to getting results back, but don't make any quick decisions once you have been given positive results.
That knowledge is empowering, whatever that knowledge is it comes with choices that you can make. It's all about what is right for you, so take it one-step and one day at a time and be grateful that you get to make a choice many women are denied.
Listen to the opinions around you, but choose what makes you most comfortable in the end.
Although it seems like a negative thing or burden, these results do not determine your life. The results do not change you, and thankfully to medical advancements there are precautionary steps to take. Do not let the result define you! If you are positive, be thankful you were given the chance to know, and can take preventative measures! If you are negative, still keep up on mammograms and self-checks, there is still family history! Live your life!
Just think positive and you will get through this.

Appendix E: Additional Results

Satisfaction and Participant Worry. Each participant's level of worry or concern regarding finding a job, finding a place to live, completing school or duties at work, finding a partner or getting married, having children or family planning, or reducing the risk for cancer via surgeries, treatment, etc., was assessed. An ANOVA test was preformed comparing each of these levels of worry or concern to the SWDS, and was not found to be statistically significant. The p values ranged from $p = 0.119$ to $p = 0.593$.

Education vs. Life Planning and Age at Time of Study. Education level was compared to the three life planning choice questions and age at the time of the study (See Table 2.6). A significant association was found between participant education and if they had received genetic counseling, $\chi^2(4, N = 51) = 11.16$, $p = 0.03$. Participants were more likely to have had genetic counseling if they had reached a higher level in their education. However, this test violated the rules of Chi-square test, since there were only four participants total who did not have genetic counseling.

Income vs. Life Planning and Age at Time of Study. Most participants had a personal income that was between $30,001 and $50,000 (37%) (see Table 2.7). No significant relationships were found between participant income and life-planning choices, or between participant ages at the time of the study.

Table 2.11. Participant educational level vs. life planning choices or age at time of study.

	Medical management plan in place		Genetic Counseling*		Family planning or reproductive options		Age at time of study	
Education	Yes	No	Yes	No	Yes	No	Under 25	25 and Over
Finished high school or GED	2	1	1	2	1	2	2	1
Some college education	9	0	8	1	2	7	5	4
Associate's degree	7	0	5	2	4	3	3	4
Bachelor's degree	**14**	**0**	**17**	**0**	**10**	**6**	**5**	**12**
College beyond a bachelor's degree	11	2	13	2	6	9	4	10
Total	43	3	44	7	23	27	19	31
Chi-square	$\chi^2(4, N = 47) = 4.72, p = 0.32$		$\chi^2(4, N = 51) = 11.16, p = 0.03$*		$\chi^2(4, N = 50) = 4.56, p = 0.34$		$\chi^2(4, N = 50) = 3.35, p = 0.50$	

Table 2.12. Personal income vs. life planning choices or age at time of study.

	Medical management plan in place		Genetic Counseling		Family planning or reproductive options		Age at time of study	
Personal income	Yes	No	Yes	No	Yes	No	Under 25	25 and Over
Less than $12,000	8	0	8	1	4	5	6	3
$12,0001 to $30,000	3	1	2	2	1	3	2	2
$30,001 to $50,000	**15**	**1**	**15**	**2**	**8**	**9**	**6**	**10**
$50,001 to $100,000	13	1	13	2	8	6	2	13
More than $100,000	1	0	2	0	0	2	0	2
Total	40	3	40	7	21	25	16	30
Chi-square	$\chi^2(4, N = 43) = 2.69, p = 6.11$		$\chi^2(4, N = 47) = 4.50, p = 0.34$		$\chi^2(4, N = 46) = 3.13, p = 0.54$		$\chi^2(4, N = 46) = 8.60, p = 0.72$	

Table 2.13. Participant ages by group vs. life planning choices.

	Timeline for Life		Overall Life Plans		Medical Management Plan in Place		Genetic Counseling		Family Planning or Reproductive Options	
Age at time of study	Yes	No	Yes	No	Yes	No	Yes	No	Yes	No
Under 25	15	4	9	10	18	1	16	3	8	11
25 and Over	20	11	14	17	26	5	27	4	14	16
Total	35	15	23	27	44	6	43	7	22	27
Chi-Squared/Fisher's Exact Test	$\chi^2(1, N = 50) = 1.17, p = 0.23$		$\chi^2(1, N = 50) = 0.02, p = 0.55$		$\chi^2(1, N = 50) = 1.32, p = 0.5$		$\chi^2(1, N = 50) = 0.08, p = 0.54$		$\chi^2(1, N = 49) = 0.10, p = 0.49$	

Table 2.14. Participants who received genetic counseling vs. life planning.

	Timeline for Life		Overall Life Plans	
Genetic Counseling	Yes	No	Yes	No
Yes	33	12	23	22
No	5	3	1	7
Total	38	15	24	29
Chi-square/Fisher's Exact Test	$X^2(1, N = 53) = 0.40, p = 0.40$		$X^2(1, N = 53) = 4.09, p = 0.47$	

Life Stability and Genetic Testing Satisfaction. The number of years participants were with their partners and the number of residency changes participants had in the past eight years were asked to gauge life stability. These questions were compared to whether or not participants felt that their positive *BRCA1/2* mutation status affected their timeline for life or their overall life plans. These questions were compared using an ANOVA test. There was no significant effect between years with partner and timeline for life, $F(1, 32) = 0.89$, $p = 0.35$. There was no significant difference between the number of years with partner and overall life plan, $F(1, 32) = 0.46$, $p = 0.50$. There was no significant difference between number of residency changes an timeline for life, $F(1, 34) = 0.16$, $p = 0.70$. No significant difference between number of residency changes and overall life plan was found, $F(1, 34) = 0.01$, $p = 0.92$.

Genetic testing results disclosure. Participants were informed of their positive genetic test result in clinic ($n = 26$) or outside of clinic ($n = 27$). The number of times participants reported specific details about their experience were counted (Table 2.15).

A main theme among responses by participants who received their result while outside of clinic was recalling what they were doing or where they were at the time of results disclosure ($n = 23$). Most participants who received their result outside of clinic were at home when they received the call ($n = 12$) and were called by a genetic counselor

(n = 8). Most of the participants who received their test result in clinic also spoke to a genetic counselor (n = 16) and brought along family members or a significant other (7).

One participant, who relived her result outside of clinic, wrote "I originally asked for my results by letter but when it came down to knowing the answer, I decided to rip it off like a Band-Aid and find out. I was at work and I got a call from my genetic counselor (on my cell)." Another wrote, "I was at work when I received the call. It was upsetting to find out but I knew I was at risk because of my family history."

Table 2.15. Genetic testing results disclosure.

Genetic Testing Results Disclosure (n = 61)	n
Outside of clinic	**27**
Cited specific details about the moment of results disclosure	23
At home	*12*
At work	*5*
In the car	*3*
At lunch	*2*
At college	*1*
Cell phone	14
Over the phone	10
Called by a genetic counselor	8
Sought support from family/significant other after the news	8
Expressed that they were upset by the result	6
Called by doctor	4
Follow up appointment with a doctor	4
Previously devised a plan for results disclosure with a provider	2
Follow up appointment with a genetic counselor	2
Expressed that they were happy to know their result	2
Land line phone	1
Posted while on nursing placement	1
Over Skype by mother - while in Japan	1
In clinic	**26**
Genetic counselor	16
Came to appointment with family or significant other	7
Doctor	5
Letter in the mail	3
Oncologist	2
Oncologist and genetic counselor	1
Geneticist	1
Follow up appointment with a genetic counselor	1
Not surprised when informed of result	8

> One of the participants who came to clinic with her family members wrote: Once my mum was confirmed BRCA [mutation carrier] the females in my family were given the option to be tested. We did this altogether and so we all went to the hospital as a family to get our results… my auntie and myself were positive and my sister was negative. It was strange: my sister was upset for me, I was upset for my auntie and my mum was just devastated and pleased at the same time…. Pleased for my sister by devastated for me and my auntie and we were all sad that they didn't know about this and we could have perhaps saved my grandma.

Another who was not surprised by her result wrote, "In my heart I knew I was positive. I was prepared. He [the geneticist] was most shocked when I thanked him. He was concerned as at the time I was the youngest person he had to give results to in the clinical [HBOC] study."