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Learning About a Twist in the Road: Perspectives of At-Risk Relatives Learning of Potential for Cancer

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The following statement illustrates the complexity of communicating newly discovered inherited breast and ovarian cancer risk in families.

We aren't really that close . . . and she basically sent out an e-mail to the family . . . and she said, "The good news is that I am done with chemo and everything is fine, . . . the bad news is I got my genetic test back and I have this mutation, and you probably have it too, so go get tested." . . . It came out of the blue.

More than 200,000 American women are diagnosed with in situ or invasive breast cancer every year. About 5%–10% of breast cancers are caused by inherited mutations in genes such as *BRCA1* or *BRCA2* (American Cancer Society, 2009). Each first-degree relative of a mutation carrier has a 50% chance of inheriting the same mutation. Women with a *BRCA* mutation have a 14%–87% chance of a breast cancer diagnosis by age 70 and a 10%–68% chance of ovarian cancer (National Cancer Institute, 2010). Individuals can benefit from knowing their *BRCA* status because the knowledge can assist them with decision making for screening and surveillance, chemoprevention, lifestyle changes, and risk-reducing surgery. In secondary prevention, the information is used for definitive treatment decisions (Schwartz, Peshkin, Tercyak, Taylor, & Valdimarsdottir, 2005) because women with *BRCA* mutations are at significantly increased risk of future contralateral breast cancer (Smith & Issacs, 2006–2007).

Genetic testing is not without limitations, including the potential for inconclusive results and a lack of certainty because of the variable penetrance and expressivity of *BRCA* mutations. Additionally, risk-reducing procedures such as prophylactic mastectomy and oophorectomy do not guarantee a future without breast or ovarian cancer. The psychosocial consequences of breast cancer genetic testing are unfolding as researchers study the impact of genetic testing on worries, depression, anxiety, emotions, and family and social relation-

Purpose/Objectives: To describe the experiences of women who accessed the Facing Our Risk of Cancer Empowered (FORCE) Web site after learning of a family *BRCA1* or *BRCA2* mutation.

Research Approach: Interpretive phenomenology based on Heideggerian hermeneutics.

Setting: Telephone interviews of women living in the United States who accessed FORCE.

Participants: A purposive sample of eight women aged 19–47 years.

Methodologic Approach: Team interpretation using Diekmann, Allen, and Tanner's seven-step process.

Main Research Variables: Experience of family communication of *BRCA* results.

Findings: Women described (a) finding out, (b) unexpected feelings, (c) mulling it over, (d) finding support, (e) seeking direction from healthcare professionals, (f) redefining future possibilities, and (g) navigating a twist in the road.

Conclusions: Many healthcare professionals are not prepared to address genetic risk. Some women who learned of potential risk experienced turmoil as potential risk for cancer unfolded. They felt isolated and unsupported by healthcare providers. They desired assistance in navigating the healthcare system to protect their future health.

Interpretation: Healthcare professionals have important roles in (a) assessing support networks of individuals seeking *BRCA* testing, (b) providing anticipatory guidance on risk communication, (c) remaining sensitive to the impact of seeing cancer as a future possibility, (d) allowing time for individuals to process such news, (e) assessing the psychosocial impact of news of a family *BRCA* mutation, and (f) providing referrals for support and health needs. Women desire decision support from healthcare providers. Future research should examine cancer risk communication in diverse groups of women.

ships. Studies exploring the psychosocial consequences of genetic testing for hereditary breast and ovarian cancer (HBOC) syndrome found that most women do not experience clinically significant levels of distress, depression, and anxiety (Crotser & Boehmke, 2009).

However, in qualitative studies, women expressed that communication of genetic test results to family members is distressing (d'Agincourt-Canning, 2006; Kenen, Ardern-Jones, & Eeles, 2006), and they desired support and resources when determining which family members to tell, as well as when and what to tell them (Forrest et al., 2003; Liede et al., 2000; Segal et al., 2004). Little is known about the needs of family members receiving news of a family *BRCA* mutation.

The individuals tested (informers) have the responsibility to communicate genetic test results to at-risk family members (receivers). Informers have described communication of results as a burden; however, they perceived it as their duty to inform relatives (Claes et al., 2003) and their own children (Forrest et al., 2003). Informers often feel torn between their duty to inform relatives of the potential health threat and the wish to protect the receivers from distress and uncertainty (Foster, Eeles, Ardern-Jones, Moynihan, & Watson, 2004; Green, Richards, Murton, Statham, & Hallowell, 1997; Hallowell et al., 2005; McGivern et al., 2004). Informers communicate genetic test results to the receivers to provide risk information (Hughes et al., 2002; McGivern et al., 2004), encourage testing, and receive emotional support (Hughes et al., 2002).

Informers have described the experience as distressing when receivers did not want the information (Blandy, Chabal, Stoppa-Lyonnet, & Julian-Reynier, 2003; d'Agincourt-Canning, 2006). Informers believed that receivers had difficulty understanding and processing the information (Daly et al., 2001; Wagner Costalas et al., 2003). The findings of research studies have emphasized the need to prepare individuals seeking genetic testing for HBOC (informers) for the potential reactions of receivers.

Informers have reported difficulty explaining results of *BRCA* tests and desire support from healthcare professionals. More than 75% of informers wanted resources, in particular an opportunity to speak with others who have lived through the *BRCA* testing and disclosure experience (Forrest et al., 2003; Segal et al., 2004). Additionally, researchers reported that informers desired support groups (Liede et al., 2000). However, little is known about recipients' perspectives and experiences of being told results—the healthy individuals, faced with potential for future cancer diagnosis. Therefore, the purpose of this study was to describe the experiences of women who received news of a family *BRCA* mutation from a biologic relative. Because this healthy population was difficult to identify, the researchers recruited participants from the organization Facing Our Risk of Cancer Empowered (FORCE) via the Internet and at an annual conference sponsored by FORCE. The organization provides peer-reviewed informational support, emotional support, and advocacy for individuals affected by *BRCA* mutations. Women at risk for HBOC and women with

known mutations establish connections through Internet message boards as well as an organizationally sponsored annual conference. The specific aims of the current study were to (a) describe the experience of learning about a family genetic mutation and understand what approaches are most helpful or not helpful in disclosing the news of a family mutation, (b) describe the meaning of genetic risk to biologic relatives of *BRCA* mutation carriers, and (c) gain an understanding of the practical knowledge used in living with this risk.

Methods

The researchers used Heideggerian hermeneutic phenomenology to guide this study. Heidegger (1962/1927) described the use of language as the source of understanding and wrote that “the intelligibility of being in the world expresses itself through discourse” (Heidegger, 1962/1927, p. 151). Thus, researchers can interpret, through analysis of interviews, how the receivers of the genetic risk information interpret their lives, make meaning of what they experience, and gain practical understanding. Genetic risk disrupts the sense of future possibilities as women “see cancer in their future” (Di Prospero et al., 2001). Through hermeneutic analysis, researchers can gain an understanding of how women think about future possibilities (Johnson, 2000).

Sample

The researchers recruited participants with an invitation and informational flyers at the FORCE conference and a posting on the FORCE Web site. Eligibility criteria included (a) 18 years or older, (b) English speaking, and (c) considering or having completed genetic testing or counseling after receiving notification of a family *BRCA* mutation. Interested people contacted the researchers by e-mail and then received information about the study and provided consent prior to the interview. The researchers recruited additional participants until no new themes emerged and saturation occurred.

Data Collection and Analysis

After institutional review board approval, the researchers obtained consent and conducted individual, in-depth interviews via telephone. The interviews lasted 45–60 minutes and were recorded on audiotape and transcribed. Transcripts were deidentified and checked for accuracy.

A team including an expert in hermeneutic phenomenology analyzed the data using the seven-step process from Diekmann, Allen, and Tanner (1989). The seven-step process involves (a) reading the interview transcripts to gain overall understanding of the text, (b) writing interpretive summaries and possible themes, (c) analyzing transcripts as a team to identify themes, (d) returning to the text for clarification of disagreements

in interpretation and writing a composite analysis of each text, (e) comparing and contrasting texts to identify shared practices and common meanings, (f) identifying constitutive pattern(s) that link the themes, and (g) eliciting responses and suggestions on a draft from the interpretive team and study participants. Additionally, the rigor of the study was enhanced by use of verbatim quotations to support the identified themes.

Findings

A total of eight women participated and ranged in age from 19–47 years (see Table 1). All of the women elected to proceed with genetic testing, and seven of the women had a deleterious *BRCA1* or *BRCA2* mutation. Genetic testing was sought from genetics counselors as well as primary care physicians. Only one participant had a personal history of breast cancer.

Table 1. Sample Characteristics

Characteristic	n
Age (years)	
19–30	1
31–40	3
41–47	4
Marital status	
Single	4
Married	3
Divorced	1
Participants with children	4
How risk was communicated	
Face to face	5
Less personal (e.g., phone, mail, e-mail)	3
Decision for genetic testing	
Immediate testing	3
Waited for next physician appointment	1
Waited several months or years	2
Cancer diagnosis	1
Need for repeat biopsies	1
Risk-management strategies of women who were <i>BRCA</i> positive (N = 7)	
Surveillance and oral contraceptive	1
Prophylactic hysterectomy and BSO	2
Risk-reducing mastectomy	3
Mastectomy for breast cancer from mutation	1
Setting of genetics test	
Genetics counseling service	5
Primary care physician	2
Other	1
Roles of FORCE	
Informational support	6
Emotional support	5
Annual conference	3
Understanding types of mastectomy procedures	1
Tangible support (e.g., referral)	1

N = 8

BSO—bilateral salpingo-oophorectomy; FORCE—Facing Our Risk of Cancer Empowered

Note. FORCE served more than one role for several participants.

Analysis and interpretation of the texts revealed six themes representing the experience of learning of a family *BRCA* mutation and the subsequent implications on their lives (see Figure 1).

Theme 1: Finding Out

Risk was uncovered for many reasons and in a variety of ways. Some women received the news after a young family member's breast cancer diagnosis and subsequent genetic testing. Others had a family history of breast or ovarian cancer at a young age, which ultimately led to testing. For some women, being told that the family harbored a genetic mutation for breast and ovarian cancer was not a surprise; for others, the news came as a complete surprise. Eventually, the women received communication about family mutations through a variety of ways, including in person and less personally by e-mail, phone calls, and mailed letters.

In-person communication: Half received the news in person from a first- or second-degree relative, mostly female relatives. In-person communication provided opportunity for interaction and observations of emotional reactions, which some women found uncomfortable. One woman described her reaction of disbelief.

My father [initially informed me]. He had heard about it. I didn't take it really seriously at the time, like I hadn't bought into it. He said a test can be performed, and I said, "Yea, right, whatever, maybe one day I will look into that." . . . I can't remember what person in my life said that it is not passed paternally, and I just believed it.

For some women, the uncovering of the risk began with interactions with healthcare providers because of the women's family histories. One healthcare provider encouraged a woman to have her mother tested to get the most informative test results. She felt as if the physician "played an emotional card."

She asked if I had thought about it, and I said, "Well, not really." . . . And she used a slightly dirty tactic. She said, "If you are interested, then your mom should get tested. It is better for your mom to get tested while she is still around, and what if you wait 10 years and your mom isn't here anymore?"

Another woman asked her mother to get tested because she believed that her mother's cancer had a genetic cause and, therefore, had concerns for her own health. Ultimately, this approach led to an unfavorable reaction when her mother communicated her test results by presenting her daughter with a paper copy of the test results, saying, "Here are the test results you wanted." The results came back positive, and her mother was upset and angry. She felt guilty to have potentially passed on the mutation and worried that her children were at risk.

Theme 1: Finding out

- In-person communication
- Less personal communication

Theme 2: Unexpected feelings

- Receiving own test results

Theme 3: Mulling it over

Theme 4: Finding support

Theme 5: Seeking direction

Theme 6: Redefining future possibilities

Constitutive Pattern: Learning to navigate a twist in the road

Figure 1. Study Themes and Constitutive Pattern

Less personal communication: Some women received the news via telephone because of geographic distance. Telephone communication provided some personal connection. One woman was aware of the impending test results.

She called me in the afternoon and said I have some good news and some bad news, and the good news was that her lymph node biopsy, it was a sentinel node, was clear . . . and the bad news was that she had just gotten back her . . . genetic test and it was positive for a *BRCA1* mutation. So I remember it really well because . . . I was panicked when she told me.

Other less personal ways of communicating were by e-mail and mailed letters that included an accompanying copy of the test results. In both situations, the surprising and impersonal news came from relatives to whom the women were not emotionally close and who lived a long distance away. Receiving the news via mail or e-mail did not allow for interaction between the family members. One woman would have preferred to know about her relative's potential genetic test before getting an unexpected e-mail announcing the family mutation. As a young, single woman recalled, "Genetic testing for breast cancer was definitely something I never heard of before my aunt [was tested]; it was a surprise I wasn't expecting." They had mixed feelings about the impersonality yet ultimately were grateful for the information. As one woman said,

I was kind of angry at my sister for telling us that way. So matter of factly rather than giving us a call. . . . Ultimately, it is an altruistic thing to tell your family that they might have this issue, [but] it wasn't very empathetic.

Theme 2: Unexpected Feelings

The women often were not prepared for their reactions to the new knowledge of a family member's mutation. As one said, "She kind of just gave us the results out of nowhere." Women experienced shock, worry, anger, numbness, and isolation. Some women were not ready to receive the news.

When I talked to my younger sister about it, she . . . [said], "I don't think I want to do anything about it. I am just too overwhelmed with everything else going on in my life, I just feel differently, and I just don't know if I will do anything about it like you."

The women expressed that even though they were expecting the news, seeing the results in writing made cancer a real possibility in the future.

When I first read [my mother's] results, I felt very kind of detached, almost like I was watching, almost like an out-of-body, like I was hovering above the room, watching everything happen. . . . I felt very, I guess numb is probably what it was. I had thought about this [genetic risk] . . . but it was never real to our family until that moment. And then all of the sudden, wow, this is a real possibility.

Receiving own test results: All of the women in the study proceeded with a genetic test. Many expected that their results would be positive, and some described taking steps to "prepare for the worst," but many of the women were surprised at their reactions to their test results. One woman related, "I didn't expect to be sad. I didn't expect to get so little support. I didn't really expect to be abandoned [by the medical community]." Another woman stated that getting a genetic test result "is not like a blood test result, 'Oh, your cholesterol is high,' and it was treated the same way." She found out while she was involved in an important work project via a phone call from the nurse.

And I started crying, which surprised myself because I had a dream that I was positive, so I was already convinced that I was, but I think it was a combination of the stress and the environment and the way in which I was finding out and all that.

For one woman recently diagnosed with breast cancer and then a *BRCA* mutation, worry about and communication of the family mutation to her teenage girls was "the scariest part of getting the news for me. I think it was mostly because of my girls, thinking about them having to go through something like this." Finding out that she carries the mutation was described as the "low part of the [breast cancer] journey, even when I expected it, it really threw me for a loop."

Only one woman tested negative for a mutation, and she also was shocked with her results and how she felt: "I don't think I can believe that. . . . And I said, 'Is there any way you could fax me a copy of the results?' I just needed to see it in print."

Theme 3: Mulling It Over

Although all of the women in this study elected to proceed with a genetic test to determine their own risk, they took time to carefully consider and mull it over. The women considered their own personal family histories

and experiences with a family member's cancer. One woman explained that her 72-year-old grandmother with a positive mutation had never been diagnosed with cancer, and she wondered whether that would happen to her.

Contrary to the other women, one participant fearfully sought testing immediately after receiving the news because a genetic mutation symbolized a cancer diagnosis and fear of death despite her current health status.

I got the results, and it was before I had to leave town, and I was just in a panic, and I just wanted to get the test drawn before I left town given how long it takes to get the results back. I felt like I needed to know right then what the results were for me.

Although she did not appear to mull it over, she had a period before receiving news of the family mutation to contemplate what she would do if her family harbored a mutation: "I remember thinking about it a lot beforehand, when she said she was going to do the test. What if she was positive? What would it mean for me? What are the implications?"

Most others had to mull it over and waited several months or even years to seek genetic testing. For two young women (younger than 30 when informed), genetic testing was not an immediate priority. One young woman waited for her next appointment with her gynecologist, whereas another waited 18 months. Decisions for genetic testing were considered as women approached the age when they needed to consider screening and risk management.

All of the women in the study consulted the FORCE Web site for support, seeking varying amounts of information and talking to others who understood the experience. Two women described visiting the site several times a day, particularly as they considered surgical risk reduction. The FORCE Web site was described as "the best source of information."

One woman made the decision because of repeated needs for biopsies.

I had actually had counseling years ago when my cousin was positive and decided at that time there was no purpose in doing the blood test because what am I going to do than I am already doing differently? I am already doing [magnetic resonance imaging] and annual mammograms and twice-a-year breast exams with the doctor. And I thought, "I am not ready to have surgery or mastectomy of any kind, so there is no point in knowing those results." . . . But then I needed yet one more biopsy, so I thought . . . "I might as well go out and find first if I carry this mutation."

After genetic testing, another period of mulling it over occurred. Several participants could not fathom risk-

reducing mastectomy and oophorectomy. They took time to mull over the best strategy for managing risk. Mulling over the options caused anxiety or worry.

People talk about the anxiety of surgery and the anxiety of surveillance, but I haven't heard anyone talk about the anxiety of making a decision to do one or the other. They act like it is just something you will figure out.

Another woman said,

I knew what I needed to do, but the idea of actually doing it . . . I just couldn't fathom the idea of losing breasts. That could potentially be healthy, but at the same time, I may never get breast cancer, so how do I know and that kind of thing?

Several women recommended taking time to make decisions for genetic testing and managing risk. Although the period of "mulling it over" was anxiety provoking, time was needed to "come to grips" with the implications and to think through decisions for surveillance versus surgical options. Surveillance comes at a price, as summarized by one young, single woman.

It feels like a never-ending parade of doctor appointments and anxiety, and if I do this for the next four years of my life, I am going to have many biopsies that turn out to be nothing, many call backs that turn out to be nothing, and [magnetic resonance imaging] is \$5,000. . . . You know, it just seems like a lot of work, and the best I can hope for is cancer is caught early, and that's kind of a long shot . . . especially when you are talking about a cancer as aggressive as a *BRCA1*, so it just is a lot of work, a lot of expense, a lot of time, and a lot of anxiety, and a lot of worry for a payoff that actually may never come, so that's what sucks about surveillance.

One woman explained that the period of mulling continued even after she decided to have surgical risk reduction as she contemplated what type of surgery and reconstruction to pursue.

I don't think six months was really a long time. . . . I have had this mutation my entire life. Just because I got results right now doesn't mean that I need to jump in and do something that the rest of my life I am upset with the physical outcome and my risk. I took enough time for me, and everybody has their own right amount of time to be comfortable with their decision.

When faced with a concurrent diagnosis of breast cancer, one participant stated that a decision for a mastectomy in light of a positive *BRCA* mutation is a big step to take: "On one hand, it doesn't seem like it should be, that it should be a no-brainer, but on the other hand, it is a pretty big piece of yourself."

Theme 4: Finding Support

When the news was communicated to the women, it often did not come with advice about seeking support. However, women in this study sought support from their friends, families, church communities, and others who had been through the experience. They needed support when receiving news of the family mutation, making decisions for genetic tests, and determining what to do with the test results. Personal knowledge of a *BRCA* mutation is not common; therefore, finding others with personal experience to lend support is difficult. For that reason, the women in this study went to the FORCE Web site to seek the support needed. The kinds of support the women described needing was information on what it means, understanding how to live with the risk, and emotional support. For a young participant, her mother was a strong source of emotional and practical support: “[Mother] was really reassuring. . . . It all happened so quickly. . . . We just saw my aunt had breast cancer . . . so finding out we had this gene in our family was a big surprise.”

Sometimes the dynamics between informants and receivers were not supportive because the informants were experiencing their own emotional responses to the news of a family mutation or a cancer diagnosis and treatment. In such situations, the women sought support from other individuals in the family and others outside the family network. One participant related, “I knew that [mother] wasn’t going to be the kind of person who was going to take care of me emotionally in that moment. That’s the way it’s always been.” This woman described herself as “desperate to talk to someone . . . through the whole thing as I was deciding to get tested and when I was waiting for results and then when I got the results.” Finding support from others who had “the gene” reduced the women’s sense of isolation. One woman depicted her need for support while trying to find a group in a similar situation yet related that traditional breast cancer support groups did not meet her needs.

Most of the women in the group were [young] breast cancer survivors . . . so it was weird . . . even though the discussion was meant to be about the *BRCA* stuff, you get a group of women together who have all had breast cancer, you can’t stop them from talking about chemo.

She later found a group of women affected by a *BRCA* mutation without cancer; however, many were 10–20 years older and dealing with different issues in their lives, making it difficult to relate.

Other women described supportive friends. Support was expressed in several ways.

Two of my friends in particular are older than me, and they are very wise women, and they spent a lot of time listening, and neither one of them was very

familiar with the gene or what it meant or the statistics, so they would just listen as I explained it over and over again . . . and they would offer advice, or ice cream, or a glass of wine, or whatever was called for at the time, but mostly it was about listening and then offering to pray for me.

When making surgical decisions, one woman expressed family support for some decisions but not for risk-reducing mastectomy: “Both my mom and my sister have been very supportive of the hysterectomy . . . but when I start talking about the possibility of mastectomy, they both get a lot more quiet.”

FORCE was a very powerful source of support for the participants in this study. One statement illustrates the importance of the availability of information and support for healthy women when discovering their potential risk for future breast cancer.

I remember when I found [the FORCE Web site] at work. And I came home and told my husband, “I feel like I am not alone anymore.” Because . . . there are very few people in this town who know what to do with someone like me. So when I found this Web site, I felt like, oh, there is all this information, there is all these women who have been there and done that. The executive director actually e-mailed and called me and helped me. . . . So through the Web site, I got the care I am going to need.

In contrast, three women in this study were not ready for the depth of information on the FORCE Web site when they first learned of the mutation. Some women described needing to “take a break” from the online environment to process the information of a family *BRCA* mutation. “Learning about mastectomy and oophorectomy was devastating. Knowing what I needed to do and actually doing it are two different things.” However, in the following year, the FORCE Web site and conference were tremendous social supports for that woman. As the women processed the implications of a *BRCA* mutation, the support of individuals who had been through the decision-making process was invaluable. Three individuals used books in addition to the Web site to find support.

Theme 5: Seeking Direction

Participants in this study sought direction and decision support from healthcare professionals and others with a similar experience of a genetic mutation. They desired additional direction from healthcare providers, even though most participants were highly educated and several were healthcare providers themselves. However, most were unhappy with the initial support from healthcare providers. Direction was especially needed when genetic test results were received.

[The genetic counselor] knows I am in the medical field, and she says, “Do you know what to do from

here?" . . . That was the last time I ever had any contact from her. I needed a lot more than that, I needed a referral to FORCE. . . . I knew all the statistics . . . but I really didn't know where to go from there. It wasn't just sign up for surgery. It is so much more complicated.

Women became frustrated with healthcare professionals' lack of knowledge about management of individuals with *BRCA* mutations, which was another reason they used the FORCE Web site for information, as one woman related.

The first surgeon I went to didn't believe in [magnetic resonance imaging], he didn't believe in mastectomy prophylactically. As much as that is what I wanted to hear, deep down, I knew he wasn't right. . . . [He] told me, by the way, that only people with truly high risk are those who have already had breast cancer. . . . Am I supposed to sit around and wait til breast cancer comes and hope I survive it?

Other women felt that they had to "be their own physicians" and become self-advocates. They felt a sense of abandonment from the medical community.

My genetic counselor gave me a list of 12 doctors, and I was supposed to figure out which one was the best one to go to. It was very isolating. . . . It is almost like diagnosing someone with cancer and saying, "Okay, now go and find a doctor to treat you." That doesn't happen.

When receiving results of their own genetic tests, women wanted direction from healthcare providers. One told her story of receiving the results over the phone on a Friday night.

You need someone sitting across from you who can answer your questions right then or who can say, "I don't know the answers; we will find them." They didn't know who to refer me to. They could have waited to give my results until they did some research about where they were going to send me next.

Another woman received her test results by phone late Friday afternoon and said, "I think calling me at 4 on Friday afternoon was irresponsible." Because of the time of the call, she was not able to receive the support she needed after receiving positive results.

Several women said that healthcare providers had a lack of knowledge and gave little direction. Women said they were the "first" patients with a *BRCA* mutation that their physicians had experienced. In one situation, a woman felt she was educating her healthcare provider.

I have to educate my friends, and I was educating my parents, I was educating my sister, and I was educating my husband, shoot, I was educating

my OB-GYN, and it was like, okay, well, when do people start taking care of me instead of the other way around?

FORCE was able to provide emotional, informational, and tangible support for the woman. It provided assistance by finding specialists knowledgeable in management of women with *BRCA* mutations.

Theme 6: Redefining Future Possibilities

Many of the participants' stories showed that after the initial shock and time of mulling it over, they came to some sort of decision as to how they would manage their risk. One woman with breast cancer described gaining new genetic knowledge to make an informed decision.

I have had over a month now to get my head around all this and kind of be at peace with this for the most part. . . . I am putting one foot in front of the other. It has taken a few weeks to get to this place. I think I am in the place I am because I very consciously focus on the positive and not the negative; it has made all the difference.

Perspectives about risk changed over time for several of the women. For some, a shift took place as risk became a real possibility for them as individuals and for their family members. For others, shifts in perspectives on genetic testing, surveillance, and risk-reducing surgery changed over time.

Finding out that a family member has a mutation in the breast cancer gene can provide an explanation for prevalence of cancer in a family. For a young, single woman who lost her mother to breast cancer when she was a teenager, receiving news of the family member was an epiphany.

Eureka! . . . No wonder there is so much cancer in our family, because there is this gene, and it just seemed kind of obvious after I found out [my aunt] was positive. . . . I guess I felt like—not relieved—but it was nice to know there was something concrete that was causing it.

These women found out over time the true implications of the mutation on their whole lives and how it would change their views and practices. One woman reflected,

With the amount of surveillance that has been recommended, I just don't want to spend my life going to doctors . . . for someone who is healthy, it is a lot of doctors, and I do not want this to change their life (husband and children). . . . I will do what I can so they do not have to watch their mom go through cancer. That is my bottom line. It took me a while to get there, but I had to come up with a way by which I make my decisions. And that is my standard.

For some women, redefining their futures included self-transcendence. They experienced a shift in meaning

and purpose in their lives after they learned of genetic risk for breast and ovarian cancer. Self-transcendence was expressed in various ways. As one woman said, "I said to my husband, 'Maybe this is my cause . . . and maybe everything, everything was leading toward this path,' and I can be very content with that. . . . I am really content to be where I am right now."

One woman discussed her change in attitude regarding risk-reducing mastectomy and oophorectomy: "I have completely changed my mind, and it is not a never, it is just a when." The younger women also had different concerns regarding childbearing and relationships; as one said, "A lot of people look at it as a huge burden, I have got to have kids early, have my ovaries out, have my breasts removed, but for me it is kind of a blessing, you know, that I can do something to prevent it." Another described her shift in attitude after she had a serious relationship and considered having a family. This young woman decided to seek genetic testing because "when you learn that you have more to lose or you potentially have more to lose than you do before, that is when I realized I need to take action in order to save myself."

Constitutive Pattern: Learning to Navigate a Twist in the Road

The women vividly described the communication of the genetic risk and possibility for cancer diagnosis as an event that caused them to consider alternative possibilities for their future dreams, in essence a twist in the road. Whether the information was conveyed in person, by phone, through the mail, or via e-mail, in most cases the newness and existential nature of the news caused the women to experience unexpected feelings and a need to seek information and support on a topic that is not well known in society. Healthcare providers often were ill informed. Families had their own guilt and emotional reactions, which confounded the need for information and support. The FORCE Web site provided current information and access to people experiencing the same situation with its resultant emotional turmoil, decision making, and existential shift. The women advised taking time to mull it over and seeking support in the form of information and emotional support from people who understood the experiential moment when they heard their own results. Advice from others in a similar situation and the practical knowledge of living and managing their risk were found through discovery and trial and error. As one woman, a young mother of two who as a young college student watched her mother live through breast cancer, aptly summarized: "This is really the case of a double-edged sword. It has been very, very difficult, but I can also see the complete blessing in this. Totally, I am in charge right now."

Discussion

The themes emerging from this analysis offer healthcare professionals a deeper understanding of the uncertainty and turmoil women experience as a family *BRCA* mutation is revealed. Women seek understanding, support, and direction as they make decisions for genetic testing, surveillance, and surgical risk reduction (see Figure 2). The purpose of this study was to elucidate the experience of women receiving news of hereditary cancer risk and the approaches to risk communication from family that were most helpful (aim 1), to describe the significance of genetic risk on their lives (aim 2), and to gain an understanding of the practical knowledge women use in living with this risk (aim 3).

Aim 1

Experiences of phenomenon cannot be understood in isolation from the context of the world in which we live. As humans, we are inseparable from an already existing world (Draucker, 1999). Therefore, the experience of learning about a family *BRCA* mutation was influenced by the context of the individual's relationships and family communication styles (Blandy et al., 2003; Kenen, Ardern-Jones, & Eeles, 2004).

Women in this study reported a variety of ways that they learned about their risk, similar to previous studies (Blandy et al., 2003; Finlay et al., 2008). Most study participants desired personal communication and interaction when receiving news of a family *BRCA* mutation.

- Before communicating genetic test results to family members, "interview them" to find out whether they are interested in the results or whether they are overwhelmed with other things.
- Communicate test results to family members in person or over the phone if possible.
- Consider visiting a genetic counselor with the relative when the relative receives results to learn more about risk.
- If you don't have cancer, take your time making decisions for genetic testing. Think about what it will mean in your life.
- If you decide to get a genetic test, make an appointment with your healthcare provider to get your results—even if you think you are going to be positive.
- Find a healthcare professional with specialized knowledge in managing women with *BRCA* mutations.
- Talk to others who have been through the experience.
- Read about others' experiences.
- Other sources of support used by participants
 - Facing Our Risk of Cancer Empowered (FORCE) Web site
 - FORCE annual conference
 - Be Bright Pink Web site
 - *Pretty Is What Changes* by Jessica Queller
 - *Blood Matters* by Masha Gessen
 - "Ovarian Cancer Risk-Reducing Surgery: A Decision-Making Resource" by Fox Chase Cancer Center
 - DVD: "In the Family," a documentary by Joanna Rudnick

Figure 2. Practical Knowledge and Advice for Communicating and Managing Risk

Several appreciated knowing in advance that a relative was getting genetic testing to prepare themselves for results. Additionally, at the time of disclosure, they desired informational and emotional support. Women did not find it helpful to be told by relatives to “go get tested.” They needed time to understand the personal meaning of genetic testing, the options for testing, and the influence of intensive risk-management strategies on their lives. Women needed several months or more to process the information and understand the potential impact of knowing their own *BRCA* status.

Women in this study had varying levels of knowledge about hereditary cancer risk when they received the news. One woman did not understand that *BRCA* mutations could be inherited from the paternal side of a family, as found in previous research (Green et al., 1997). Women with no prior knowledge of their risk had an eye-opening experience, whereas those with significant and known family histories still experienced personalization of risk.

Study participants described social support as important and helpful. Support came from family members, friends, coworkers, others who have HBOC syndrome, and healthcare professionals. Emotional support may need to come from outside the family network because *BRCA* test results have family implications which may cause blame and guilt within the family network (McInerney-Leo et al., 2005). Seeking support from sisters was similar to a recent study of 65 sisters from 31 families with *BRCA* mutations who shared worry and stress and adapted within their natural networks (Koehly et al., 2008). This study supports the notion that support can influence coping in positive and negative ways.

As the women in this study learned of their potential risk, they found information and support from the Internet, an apomediary and very powerful source of support. Apomediation is an “agent that stands by to guide a consumer to high-quality information and services without being a prerequisite to obtain information or service in the first place” (Eysebach, 2008, paragraph 18). The FORCE Web site and other Internet resources provide access to apomediaries (women who have experienced and live with genetic risk), as well as information about *BRCA* mutations, risk management, and forums to discuss concerns.

Aim 2

Receiving news of a family mutation was a life-changing experience for women in this study. Their breasts no longer were perceived as healthy but as a threat of illness and, for some women, a threat of death. The threat became real the moment a family member received a positive *BRCA* result, and action was needed to prevent a future with cancer.

Heidegger viewed the body as more than a machine. Human bodies “provide the possibility for the concrete action of the self in the world” (Leonard, 1989, p. 52). When our bodies break down, it is more than an objective problem that needs to be fixed but changes the way we experience our world. Whether or not participants were familiar with their risk, the news of a family mutation caused serious reflection on their way of being in the world. As communication of risk occurred in the families, women realized that cancer is a real possibility in their futures and is more than a matter of cutting out tissue that harbors potential for disease and death. In this study, women described the need to “mull it over.” Taking time is an important aspect that is contrary to a culture that focuses on treating illness quickly.

Women in this study used genetic knowledge to redefine their futures—without cancer. Discovering potential for HBOC is a life-altering experience. Heidegger suggested that technology is a means to an end but that it is not neutral; in fact, technology can organize us. Technology has the potential to transform the way we know and think (Heidegger, 1977). Technology enables women to determine whether they have inherited risk for breast and ovarian cancer and increases the ability to detect breast cancer early. On the surface, genetic knowledge may be good to have, a way of gaining control over potential interruptions in life caused by cancer and cancer treatment. However, women in this study found that it was not as simple as having the information for screening or risk-reducing surgery. Surveillance and risk-reducing surgery come with their own prices. Women faced uncertainty about risk reduction because a deleterious mutation does not always mean future cancer, as one young woman reported that her 72-year-old grandmother with a deleterious mutation has not developed cancer. However, similar to findings of Hamilton and Bowers (2007), women in this study wanted to take action to alter or avoid the experience of other family members with hereditary illnesses, yet they described the need to carefully reflect and consider what knowledge of cancer risk would mean in their lives. They needed support to navigate the journey, and healthcare providers were not prepared to fulfill that role.

Women in this study found that many members of the medical community lacked knowledge of recommended risk management for individuals with *BRCA* mutations and lacked understanding of the meaning and significance of genetic knowledge on their lives. This left them with a sense of isolation and the need to advocate for themselves. As a result, women with HBOC turned to each other for informational, emotional, and sometimes instrumental support. The women in this study wanted more concrete direction from healthcare providers, possibly in the form of decision aids, to understand their risk and problem solve. Information support is helpful

in several ways. It can help individuals understand how to reduce illness threat and give them hope for a future without disease (Helgeson & Cohen, 1996).

Similar to individuals with cancer, some women experienced self-transcendence (Coward, 2003) after learning about potential risk for HBOC. Discovering a family mutation caused them to reach out to others. For several, it was a turning point in their lives and led to changed priorities; others discovered a new purpose in life, such as helping others navigate the journey.

Aim 3

The women in this study did not struggle with the decision for genetic testing; however, they cautioned others to take their time to think about how the decision would affect their lives. Once they had genetic testing and received positive results, they were confronted with another twist in the road and sensed a lack of direction. They experienced anxiety while making decisions about surveillance versus surgery. They carefully researched mastectomy procedures so that they could achieve the best outcomes in terms of cancer risk and cosmetic results to maintain a sense of womanliness. Most learning and support for this group of women occurred through interactions with other *BRCA* mutation carriers on the FORCE Web site.

Implications for Nursing

Clinical Practice

Healthcare professionals will be called upon to recommend and provide credible resources, such as FORCE, for emotional and informational support for individuals with *BRCA* mutations facing decisions for genetic testing, surveillance, and risk-reducing surgery. Additionally, nurses and other healthcare professionals can evaluate the accuracy and influence of Web-based resources for individuals seeking and reacting to results of cancer genetic testing.

Healthcare professionals have an important role in assessing the support network of individuals who have potential for HBOC to evaluate needs for referral for additional support. Education and counseling for individuals on their role in communicating genetic risk to at-risk family need to include the possible range of reactions of family members. Some family members might not want to know their risk for a variety of reasons; it may be too much information for them to process because of other life priorities and developmental tasks they face. Assessing family relationships and communication patterns is important in preparation for communication of results. The Colored Eco-Genetic Relationship Map is a tool that delineates family health history and support systems and can assist health professionals in social assessments of individuals seeking genetic counseling (Peters et al., 2004).

Women in this study expressed a preference for face-to-face meetings with their healthcare professionals when receiving their own genetic test results. Several desired specific referrals to providers who specialize in the care of women with *BRCA* mutations and information on available support networks. At the time they received their genetic test results, they desired specific information on next steps and help with decision making for surveillance and risk-reducing surgery. They wanted balanced information on the benefits and limitations of all options.

A model of care in which a tentative appointment is scheduled to discuss genetic test results at the time the results are expected would have met the needs of the women in this study. Furthermore, women with negative results desired a face-to-face meeting to discuss implications and the potential emotional and social impact of negative results. Moreover, receiving results can be overwhelming, so women might benefit from future contact with a genetic counselor or nurse to assess need for additional support.

Research

Additional research into communication of *BRCA* test results to at-risk family members is important to further understanding of ways healthcare professionals can provide anticipatory guidance and prepare individuals for possible reactions and responses to receipt of news of a family mutation.

This study was limited to women who accessed FORCE. The organization promotes aggressive surgical intervention, which might not be desired by all women with HBOC syndrome. Further study is needed to gain understanding of (a) the experience of receiving news of a family mutation from a more diverse sample, including women who do not use the Internet for support; (b) strategies to deliver support for individuals who do not have their needs met in an online environment; and (c) how to provide online support for young women who are not ready for the detail and promotion of surgical risk reduction. Understanding the needs of those early in the journey of HBOC syndrome is important for nurses who provide health education, support, referrals, and advocacy.

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