Conditions and Consequences of a BRCA Mutation in Young, Single Women of Childbearing Age

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Young women (aged 18–39 years) who are at risk for hereditary breast and ovarian cancer (HBOC) because of a BRCA1 or BRCA2 mutation face a high risk of developing cancer before age 50 and a 50% chance of transmitting that risk to each of their children (Surveillance, Epidemiology, and End Results Program [SEER], 2008). By contrast, onset most frequently occurs from 62–79 years of age in women at average risk for breast cancer who do not have a family history of the disease (SEER, 2008). After discovering a personal high risk for HBOC, women are faced with a cascade of decisions for which they may have little preparation; the most wrenching can be whether to pursue prophylactic bilateral mastectomy and oophorectomy to manage their risk. For young women, the decisions pose special challenges related to sexuality, family and social relationships, reproductive choices, and achieving important vocational or other life goals. Young women with HBOC risk who also are single may face even greater difficulty in adjusting to their risk status in their efforts to establish intimate partnerships.

Background

Young adulthood is defined by Erikson (1963) to be from age 18–39. Challenges across young adulthood are seeking independence from parents, establishing gender identity, internalizing moral values, and examining career choices. Young adults typically are exploring intimate relationships while making childbearing, work, and lifestyle decisions (Newman & Newman, 2006).

Several sociologic, anthropologic, and psychological studies (Arnett, 1994, 1997, 1998, 2000; Perry, 1999; Schlegel & Barry, 1991) indicate that role transitions such as marriage, finishing education, beginning full-time employment, and becoming a parent are the important transition points into adulthood (Heckhausen, 1997; Schulz & Heckhausen, 1996). Successful navigation of adult development involves active pursuit of achievable goals and adaptive relinquishing of, or compensation for, unachievable goals in the context of the shifting physical constraints of aging (Schulz & Heckhausen, 1996). For young women, the primary defining biologic constraint is reproductive capacity (Heckhausen, Wrosch, & Fleeson, 2001). Implicit in the decision about whether or when to manage risk by prophylactic surgery is whether to speed up volitional efforts to pursue childbearing (while managing anxiety about failure) or to relinquish childbearing and to gain acceptance of alternatives (e.g., adoption, childless lifestyle).

Young Americans have reported consistently that the transition to adulthood is characterized by an emphasis on individualism that includes accepting responsibility for oneself, making independent decisions, financial independence, and establishing a relationship with parents as an equal adult (Arnett, 1997, 1998, 2000, 2001). In

Purpose/Objectives: To explore the experiences of young, single women who are at increased risk for hereditary breast and ovarian cancer (HBOC) because of a BRCA mutation.

Research Approach: Qualitative.

Setting: Seven states and Canada.

Participants: 11 single women aged 18–35 years who tested positive for a BRCA mutation.

Methodologic Approach: Grounded theory with in-depth individual interviews conducted via e-mail or telephone.

Findings: Analysis resulted in three conditions and three consequences. Conditions were dating or not dating, time in a relationship, and physical impact of surgery or breast cancer treatment. Consequences were explaining their choices, experiencing a sense of urgency, and experiencing a sense of loss.

Conclusions: Young women who are at risk for HBOC face a complex array of decisions after finding out that they carry a BRCA mutation. Being single and childless adds to this complexity.

Interpretation: Nurses can listen to young women with HBOC risk, help them clarify their fears and understanding of their risk, and provide nonthreatening support that goes beyond simply providing more information and includes a nonjudgmental understanding of the young women’s experience.
young women at high risk for HBOC, interruption of the transitions (e.g., reliance on parents during cancer treatment or recovery from prophylactic surgery) can undermine their sense of independence and self-sufficiency.

Breast and Ovarian Cancer in Young Women

Although breast and ovarian cancer generally are considered diseases of postmenopausal women, they can develop in younger women. In 2008, more than 250,000 American women younger than age 40 were living with breast cancer, and more than 11,000 young women were diagnosed that year (SEER, 2008). Breast cancer incidence rates per 100,000 women are 1.3 for women aged 20–24 years, 7.7 for women aged 25–29, 25.6 for women aged 30–34, and 58.9 for women aged 35–39 (SEER, 2008). Ovarian cancer incidence rates in young women are lower, ranging from 1.84 (age 20–24) to 5.54 (age 35–39) (SEER, 2008). Diagnosis of breast cancer at a young age (younger than age 40) is an indicator of potential hereditary risk and can trigger referral for genetic counseling and testing (Daly et al., 2010).

BRCA Mutations

Of the expected 207,090 new cases of breast cancer in the United States in 2010, about 5%–10% will be associated with a germline mutation in a cancer susceptibility gene (Jemal, Siegel, Xu, & Ward, 2010). A mutation in the highly penetrant susceptibility genes BRCA1 and BRCA2 accounts for more than 50% of hereditary breast cancers (Miki et al., 1994; Tavtigian et al., 1996; Wooster et al., 1995).

Cancer risks associated with mutations in BRCA1 and BRCA2 include a lifetime risk of female breast cancer approaching 50%–85% by age 80, with much of that risk occurring before age 50, when traditional screening modalities such as mammograms are the least sensitive (Barcenas et al., 2006; Easton, Ford, & Bishop, 1995; Ford et al., 1998; Struwing et al., 1997). Women who carry the mutations also face higher risks for ovarian cancer, a disease in which screening and early detection remain elusive. Lifetime ovarian cancer risks vary by gene; women with the BRCA1 mutation have about 20%–40% risk, and women with the BRCA2 mutation have a 10%–20% risk (Barcenas et al., 2006; Easton et al., 1995; Ford et al., 1998).

Cancer risk management for women with a BRCA1 or BRCA2 mutation is complex and includes options for risk-reducing surgery, intensified cancer screening and surveillance, chemoprevention, and risk avoidance (Berliner & Fay, 2007; Burke et al., 1997; Eisen et al., 2005; Madalinska et al., 2007; Rebbeck et al., 2004). Despite accumulating evidence regarding the efficacy of each strategy, the management of HBOC risk is complicated by life planning issues typically faced by young, single women. For example, uncertainty about whether or when cancer might strike turns the timing of risk management into a gamble against the odds that putting off surgery until childbearing is complete will be safe.

Challenges for Healthcare Providers

HBOC risk in young women poses special challenges for healthcare providers. Knowledge and practices based on average risk are not fully applicable to young women with HBOC risk. Providers are required to keep up with current information and recommendations, know how to recognize high-risk family histories, and have a different index of suspicion for symptoms (Khoury-Collado & Bombard, 2004). Unfortunately, studies have shown that provider knowledge can fall short, such as failing to understand that BRCA mutations can be paternalistically transmitted (Wideroff et al., 2005). Providers also should be sensitive to how risk management recommendations will be received by young women. Healthcare providers must be prepared to offer support and referrals as the young women process intellectually and emotionally how they will incorporate risk information with their life goal.

Methods

Grounded Theory Method

This study used a grounded theory design method to describe the decisional processes of young women who are at increased risk for HBOC. Grounded theory, based on Symbolic Interaction (Glaser & Strauss, 1967), is designed to explore how people understand their situation, how they experience reality, and how those understandings are related to action. In this study, grounded theory examined young women’s understanding of HBOC risk and how being single affected the meaning of that risk, thus having significant consequences for their decisions and, ultimately, their lives. Analytic procedures in grounded theory are designed to elicit research participants’ constructions (logics, understandings, and perspectives) of the phenomenon under study and to reveal how changes in constructions shift with the social circumstances within which they are embedded (Clarke, 2005; Strauss, 1987). Using grounded theory allows exploration of the complexities of genetic testing within the context of being young and single.

Recruitment

Participants in this analysis were part of a larger study of 59 young women who had undergone BRCA testing. Participants were recruited from two support and education Web sites specific to young women with BRCA mutations or breast cancer: Facing Our Risk of Cancer Empowered (FORCE) and the Young Survival Coalition (YSC). FORCE is for individuals at risk for carrying a BRCA mutation, and YSC is for young women with breast cancer. Figure 1 lists other Internet resources
for patients with a BRCA mutation and their clinicians. Demographics such as age, race, and income of the population that uses the Internet increasingly reflects the general population (Fox, 2006; Hamilton & Bowers, 2006; Pew Research, 2007). Inclusion criteria for the larger study were women aged 18–39 years, history of genetic testing for a BRCA mutation, and having received test results. A total of 59 individuals were interviewed initially. This analysis consisted of the 11 women who tested positive for a BRCA mutation and were single and without children at the time of the interview. Four of 11 had a breast cancer diagnosis. Participants lived in seven different states in the United States and one lived in Canada. This study was approved by the institutional review board at the University of Pittsburgh. Participants were compensated $20.

Procedures

The geographic diversity required flexibility in the format used for interviews. To maximize participation, participants were given the choice of a telephone or e-mail interview (Hamilton & Bowers, 2006). Eight participants were interviewed via e-mail and three via telephone. All interviews were conducted by the primary investigator.

Consistent with the grounded theory method, interviews started with questions regarding participants’ general perceptions about their experience of BRCA mutation testing and their breast cancer risk. Subsequent questions focused on how being young influenced how they thought about their risk, intimate relationships, and reproductive choices.

Data Analysis

Data analysis, consistent with grounded theory method, included (a) question development around emerging concepts, (b) theoretical sampling, (c) constant comparison along properties and dimensions of categories, and (d) auditing of the research process through substantive and methodologic memos (Strauss & Corbin, 1998). The concept of interest in the current study was being single. Theoretical sampling was used to explore how various conditions influenced the experience of being single and BRCA positive. For example, depending on whether women had a breast cancer diagnosis, the direction of the interview diverged to capture their specific experiences. Memos were kept to record the substantive and methodologic thinking of the research process (Hutchinson & Wilson, 2001).

Data analysis in grounded theory is a recursive process of generating and comparing codes and memoing, moving from iterative to conceptual interpretation (Piantada, Tananis, & Grubs, 2004). Open coding (Strauss, 1987) was used in the early analysis to identify the dimensions of the phenomenon and how those dimensions related to each other as outlined by the participants, which yielded a complex and detailed matrix of the participants’ logic. Theoretical sampling also assists in the coding process by directing detailed coding along a particular condition (dating) or strategy (risk-reducing mastectomy) used by women to respond to increased awareness of risk and a new reality.

The transition from open coding to axial coding occurred when areas of focus were identified, such as role of age in perception of risk, whether the participant was in a relationship, and experiences with peers in relation to their genetic risk. Interview questions then were re-crafted to facilitate in-depth exploration of focus areas. Axial coding techniques (in-depth focused coding) were used at this point to link the dimensions to each other and to the context of situations (Clarke, 2005; Schatzman, 1991; Strauss, 1987). The analysis presented in this article examines the perceived effect of the participants knowing their risk for HBOC while they were single women without children.

Findings

The young women faced receiving difficult information about their BRCA-mutation status at a vulnerable and fluid time in their lives. The 11 single participants were aged 18–35 years. The range of time from having genetic testing was one month to three years, with most participants knowing their BRCA status for about 1.5–2 years. Three specific categories of experience were identified: breast cancer negative and prophylactic mastectomy positive (n = 1), breast cancer negative and prophylactic mastectomy negative (n = 6), and breast cancer positive and receiving treatment (n = 4). Three conditions and three consequences were developed from the analysis (see Table 1). The conditions captured the events that create a situation, whereas the consequences captured the actions or interactions in response to a situation (Strauss & Corbin, 1998). Participants tended to describe the consequences of being young and at risk for HBOC in terms of the action they felt required to take in relation to another person or by a changed perception in how they thought about their life and future.
### Table 1. Conditions and Consequences of a BRCA Mutation in Young, Single Women

<table>
<thead>
<tr>
<th>Condition</th>
<th>Consequence</th>
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<td>Dating or not dating</td>
<td>Explaining their choices</td>
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<tr>
<td>Time in relationship</td>
<td>Experiencing a sense of urgency</td>
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<tr>
<td>Physical impact of surgery or breast cancer treatment</td>
<td>Experiencing a sense of loss</td>
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### Dating or Not Dating

BRCA-mutation status created challenging situations related to dating for affected and unaffected participants, regardless of whether they had undergone mastectomy. One challenge that emerged was explaining their choices. Participants who were dating at the time of the interview spoke of difficulties in determining what and when to tell their date partner about their genetic risk. One participant who had prophylactic surgery said the following.

Up until the time of my mastectomy surgery, I was never really in a serious relationship before. I had a few boyfriends here and there, but I wasn’t in love with any of them. I very much do want to get back out there and meet somebody who I can be intimate with, but yes, it will be more difficult because now I have a huge situation to explain to whoever men I meet.

For participants who had a breast cancer diagnosis followed by treatment, dating was even more complicated.

As far as a new relationship and the breast cancer and BRCA result, it is very daunting, . . . How in the world do you tell someone I am 35 and this is what has happened in the last two years and this is what I am facing in the future? So the idea of dating again is so daunting, I have several girlfriends trying to set me up, but I am still dealing with nipple reconstruction, tattoos, etc. . . . I need to feel like my whole self again before I can have the confidence to put myself back out there. Obviously, still a major struggle for me.

Participants who had not chosen to have a prophylactic mastectomy but knew their mutation status were concerned about how to explain their situation.

Yeah, this is definitely something I think about a lot. I am not dating anyone, but I constantly ask myself, at what point of the dating (should anyone ever want to date me!!?) do I bring up something like this? I mean, do I tell someone I “might” get breast cancer, or I “might” have a prophylactic mastectomy when I’m 35? . . . Like there are loads of guys out there who would willingly get into a relationship with someone they know is very likely to get cancer? Who does that?

Participants expressed the desire to be close to someone but were troubled and very unsure about how to navigate a relationship, particularly in its early stages.

Participants who experienced the condition of dating or not dating also reflected the consequence of experiencing a sense of urgency. Most women described their reproductive plans because they had been told they should consider having their ovaries removed from age 35–40 years, or when they were done with childbearing. None of the participants had children; they felt their plans for starting a family were affected by their mutation status as well as their relationships.

I also told my boyfriend about my fears and concerns about needing to start a family much sooner than I had originally planned. That I wanted to be pregnant with my first child before the age of 30, so that I could have a second by 32/33 and then have my ovaries out before I was 35. . . . We are not really emotionally ready to be parents yet. If when we get to 30, we still feel unprepared, we will wait. . . . However, I don’t see us waiting too long, because I feel that I must have my ovaries removed before I am 36 to prevent ovarian cancer. I know that my risk does not really begin until I am approximately 40, but right now I don’t feel comfortable enough with the screening methods to wait that long.

Women who were not in a serious relationship talked about how they would plan things if they could.

My test results have also affected my life decisions in that I hope to have my children early in my marriage, if possible. That way if I later feel that preventative surgery is necessary, or if I am to get cancer at an early age, I will have already started my family.

Not being in a relationship put pressure on the young women about reproductive plans as well as on how they imagined their marriage would accommodate such a need.

But now I don’t even have a boyfriend. I am going to be 25 in July so I am assuming unless I meet someone in the next year I am probably not going to be married until I am in my late 20s which means it really will affect it . . . but I think for me personally my goal, like I hate to set a timeline of when I want to have a kid but I honestly want to have a kid around 28, 29, so then I can have them by the time I am in my early 30s and not have to worry about, “Okay, now it’s time to get everything out.” I want to make sure I have my kids first before that pressure comes. But it also makes another pressure, like what if I get married, do I have to conceive a
baby the next day? So it is a little bit more of a time pressure I definitely think.

Whether or not a participant was dating someone, she perceived herself as having to explain the choices she had already made as well as experiencing a sense of urgency, particularly over reproductive plans and the potential impact such plans might have on any future relationship. The perceived urgency influenced the pace of the relationship as well as the timing of disclosure about matters related to mutation status.

**Time in a Relationship**

Only three participants had been in a relationship for any period of time; the other eight were dating casually or not dating at all. Rejection by partners or potential partners can be a feared consequence of HBOC risk status, and two participants had boyfriends break up with them shortly after a breast cancer diagnosis and receiving their genetic test result, although only one of the women stated that the break up was a result of the risk experience. Two of the three that had an ongoing relationship described the consequence of experiencing a sense of urgency again mainly around the issue of having children.

We both have always wanted children. I am currently getting my undergraduate degree and then I will be going to law school. I still have about six years of school left, but we have always wanted to start a family young. We talk about the timeline we have, and I will definitely think of getting both my breasts removed when I am older, but my ovaries are a different story. I will no doubt still have children, but of course I can’t get my ovaries removed until I am sure I am done having kids. I get frustrated that we have to think and plan these things so young.

The participant who had been with her boyfriend for five years did not describe herself as feeling the sense of urgency, but that may be, in part, because she self-described both herself and him as “slow movers” and also stated that they essentially do not talk about her HBOC risk.

I have a problem communicating this kind of thing. It’s not something we sit around and talk about very often; in fact we don’t ever talk about it.

**Impact of Surgery and Breast Cancer Treatment**

The final condition identified was for the categories of breast cancer negative and prophylactic mastectomy (one participant) and breast cancer positive and receiving treatment (four participants). The individuals had undergone mastectomies, at minimum, and all participants who had a breast cancer diagnosis had undergone several rounds of chemotherapy and radiation as well as the surgery. The main consequence as a result of the condition was experiencing a sense of loss. One participant with breast cancer had been told by her physician that she most likely would not regain her fertility secondary to chemotherapy.

And that at this point, since I haven’t had any periods, [the doctor] really doesn’t think they’re going to come back at all. The percentage or chance of it coming back is very, very low.

For this 31-year-old single woman, the loss was difficult. Participants who had a breast cancer diagnosis and a positive BRCA-mutation test result received considerable bad news that left them with much to handle.

I am still dealing with the psychological effects of it all and have not dated since my diagnosis and my surgeries. My friends are pushing me to get back out there . . . but talk about baggage . . . and I think doctors and genetic counselors should be aware of that. It is life altering at an age where you never thought you would have this to deal with. I have also had doctors that were not sensitive to the fact that I have not had children yet and would like to hang on to my ovaries a little while longer until I know that may or may not be a possibility. You deal with so much as a young survivor and a BRCA1 patient . . . it can be overwhelming.

Participants in the breast cancer and receiving treatment category experienced many potential losses and did so at a younger age than most women who have breast cancer.

The participant who had not had a breast cancer diagnosis but decided to have a prophylactic mastectomy experienced a loss of worry over breast cancer.

Finally, in May 2005, I elected to get a prophylactic bilateral mastectomy and reconstruction. My surgery for the mastectomy was July 18, 2005. I recently had the surgery for reconstruction (implants) last week, January 19, 2006. I am very happy about my decision to get surgery before the possibility of ever being diagnosed with breast cancer. It has been hard, but in the long run it is worth it, to go on living and know that cancer will never affect my life. To me, it’s all about quality of life.

Although her facts are not quite accurate (e.g., no chance for cancer to affect her), her perception is that she is now free of worry. However, she also found herself explaining her choices to men with whom she was intimate.

Except for one guy, I had to explain what I went through, just because my breasts did not feel “normal” and I did not have nipples. Most of the guys were supportive and tried to understand the surgery I was explaining to them. Of course it wasn’t easy for them to grasp, being that I’m only 24 years old and I had to explain multiple times that I did NOT have cancer, I just got the surgery as a preventative measure.
Although she had lost her sense of worry, she also had lost the simplicity of a “normal” relationship.

The young women faced considerable challenges related to their risk for HBOC at a time when they may have had few previous life experiences that could help prepare them for the choices they now faced. The situation of being single and without children further complicated their decision making.

Discussion

The conditions identified and the resulting consequences provide a reflection of what young single women with a BRCA mutation experience. Their young lives are complicated by what they know and how they are counseled to manage their risk. In particular, their risk status led to concerns about how they would attain important markers of adult identity such as intimate partnership or childbearing, as well as reflection on ways in which they were “out of step” in relation to their peers. Participants who also had a breast cancer diagnosis were affected even more by the effects of treatment on fertility and potential future relationships.

Other factors also may influence the experience. For example, coping and communication styles appear to factor into the trade off between reducing anxiety related to urgency versus activation toward goals, as with the participant who described how she and her long-term (but as yet unmarried) partner avoid talking about her risk. Such an avoidant strategy may reduce short-term anxiety but inhibits activation toward goal fulfillment.

Prior research has shown that younger women tend to be more distressed after genetic testing for the BRCA mutation than older women (Lodder et al., 2002; Watson et al., 2004). Anxiety and depression also have been found to be associated with age at the time of genetic testing (Bennett et al., 2008). The current study’s results highlight some of the specific aspects of positive mutation status that could make younger women more vulnerable to distress. Because younger women also have been shown to express more interest in genetic testing than older women (Bottorff et al., 2002), a greater understanding of how dating, relationship, and fertility issues contribute to distress in young women at high risk for HBOC is important.

Contradictory information about risk reduction or increase resulting from reproductive choices also confounds the issues for young women. Some reports state that women with a BRCA1 or BRCA2 mutation who become pregnant before age 40 increase their risk of developing breast cancer (Hamilton, Williams, Bowers, & Calzone, 2008; Jernstrom et al., 1999; Narod, 2006). In addition, oral contraceptives may increase the risk of early-onset breast cancer in BRCA1 carriers but also might protect them from ovarian cancer (Narod et al., 2002). Such contradictory and changing reports further complicate decision making.

The current study of young, single women without children focused on the conditions and consequences of the experience of knowing one has a BRCA mutation in the early adulthood stage of life. The findings indicate that young women take into account their own personal circumstances and their perceived risk for HBOC when making treatment decisions. Longitudinal studies are needed to assess how young women live with the decisions they make related to their risk over time (Hamilton, Williams, Skirton, & Bowers, 2009). Learning how to support the decision-making process and the outcomes of their decisions is important for healthcare providers who interact with this patient population.

Limitations

Conclusions from the current study should be considered preliminary because of the small sample size. The developmental issues identified should be elaborated in a larger sample, with directed questions informed by the conditions and consequences discussed previously. The results may not generalize to ethnic minority women or to young women who receive inconclusive results from genetic testing and who need to make decisions without relatively definitive information about their risk status. Finally, the sample was drawn from women participating in national online support networks. Additional themes may emerge among young women who cannot or choose not to participate in this type of activity.

Recommendations and Future Directions

The current study’s results indicated that healthcare providers should consider the developmental issues of young, single women at risk for HBOC, particularly life goals related to childbearing and intimate relationships. Providers should avoid applying pressure on young women, particularly single women, to “hurry up and finish” childbearing because they may feel unable to enact the advice when the means are out of their control (e.g., lack of a partner) and fear exposure to rejection. Such advice can inadvertently promote distressing feelings. Rather, providers can assess a patient’s relationship status and her childbearing plans (if any) and offer support over time as the woman weighs the feasibility and timing of childbearing and other important goals in light of her risk management preferences and develops coping strategies to implement her decisions with minimal distress.

Providers also should be sensitive that altering life goals (and the timetable for achieving them) can put women at risk for HBOC out of step with their peers, possibly increasing isolation and decreasing available social support. Women who choose prophylactic oophorectomy or who are infertile as a result of cancer treatment may need support in learning to disengage...
from a desired outcome while maintaining ties with siblings and friends who are in active pursuit of goals they have had to abandon (Heckhausen et al., 2001).

Future research can be directed at creating quantitive instruments to address the conditions and consequences identified in the current study that can be used to determine the prevalence of such concerns among young women at risk for HBOC and allow for comparison with other at-risk subgroups. A long-term goal of the research program is to inform the creation of interventions with an adult developmental focus that facilitates articulation of life goals, supplants anxious urgency with an adaptive sense of time, offers communication skills training for interacting with intimate partners (and potential partners), and provides support for altering life plans as needed to promote healthy adult identity when facing HBOC risk.

### Implications for Nursing Practice

Oncology nurses will increasingly interact with individuals who have had or are considering presymptomatic genetic testing (Jenkins & Masny, 2003). Although nursing typically is focused on the whole person, a holistic orientation is critical when dealing with young, single women who carry a BRCA mutation. This population at risk for developing HBOC faces decisions that are out of sync with their peers and sometimes at odds with family and healthcare providers. Nurses should take the time and effort to understand the complexity of the issues and appreciate that a young woman is trying to make decisions with not only her genetic risk in mind but also her relational, familial, and professional future. Because of their young age, these women often have limited decision-making skills and only rarely have faced any decisions that require balancing so many factors at a given time. In addition, the choices for prophylactic surgeries are irrevocable; therefore, the decision-making process of the individual should be supported so the young woman feels that she has made the correct choice for her. Nurses should listen to young women with HBOC risk, help them clarify their fears and understanding of their risk, and provide a nonthreatening experience in which nurses’ support goes beyond simply providing more information and includes a nonjudgmental understanding. As more presymptomatic genetic testing becomes available, the knowledge gained in interacting with young women with BRCA mutations may be applied to other cancers as well.

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