A qualitative study finds that young women with a BRCA gene mutation have concerns about marriage and childbearing.

As strides have been made in determining the genetic risks of developing adult-onset diseases, nurses have begun to address a variety of novel issues with patients. Among these are the potential pathophysiologic implications of a gene mutation and the psychosocial impact of knowing about one’s own genetic risk. While being told you have a particular disease is generally distressing, being told you carry a gene mutation that increases your risk of a disease as well as that of your siblings and children raises other, more complex, issues. In previous studies I found that factors important to the experience of BRCA genetic testing included a woman’s age, marital status, breast cancer diagnosis, presence or absence of children, family history of breast cancer, family’s response to genetic testing, choices of surveillance or prophylactic surgery, and access to knowledgeable health care providers.1, 2

In 2006–2007, I conducted a grounded theory study of young women, ages 18 to 39, who had been tested for a BRCA mutation, the presence of which increases the risk of developing breast or ovarian cancer, or both, at an early age.1, 2 In this article I explore three characteristics of those women who tested positive: whether or not they were married, had children, or had breast cancer. I chose these characteristics because they are important aspects of young women’s lives that can be affected by knowledge of genetic risk. Also, I sought to help nurses as they interact with young women considering genetic testing for a BRCA mutation; young women who have already undergone testing and are engaged in enhanced screening; those considering prophylactic mastectomy or oophorectomy; and those who have already undergone such surgery.

GENE MUTATIONS AND BREAST CANCER

Everyone has BRCA genes: BRCA1 on chromosome 17 and BRCA2 on chromosome 13 function normally as tumor-suppressor genes. But mutations in these genes inhibit DNA repair and are inherited in an autosomal-dominant fashion, meaning that if one parent has the mutation, each offspring has a 50% chance of inheriting it.

All women who inherit a BRCA1 or BRCA2 mutation have a significantly increased risk of developing breast or ovarian cancer or both, and a mutation in BRCA2 increases risk of melanoma and cancers of the pancreas, stomach, and gallbladder. Breast cancer risk ranges from 50% to 85% by age 50 in women with the mutation and is 12% in women without it.4 Management of risk for BRCA mutation carriers includes discussing options for risk-reducing surgery, intensified cancer screening or surveillance, chemoprevention, and “risk avoidance”—behavior modification such as weight control.5, 9 Although such strategies may decrease breast cancer risk, they may also complicate the lives of women, especially those ages 18 to 39, affecting their decisions regarding relationships, childbearing, and a career. While these women’s risk of developing breast cancer is significant, their actual numbers in the population are not
OBJECTIVE: Women who carry a BRCA1 or BRCA2 gene mutation face a risk of developing breast or ovarian cancer at an earlier age than women without such a mutation. Relatively little is known about the psychosocial consequences—especially regarding marriage and childbearing—in young women who test positive for one of these mutations.

METHODS: In 2006, participants were recruited from Web sites for women with breast cancer or BRCA gene mutations. Forty-four women ages 18 to 39 from 22 states and Canada who had had genetic testing and were found to carry a BRCA mutation were interviewed by phone or e-mail. A qualitative, grounded theory analysis was performed on the data, focusing on the participants’ being young and having had genetic testing for the BRCA mutation. The findings reported here focus on three characteristics of the participants—whether or not they were married, had children, or had a breast cancer diagnosis—and how those characteristics were affected by the women’s knowledge of their genetic risk.

RESULTS: Among the 13 unmarried participants, issues of when to disclose information about their genetic risk in intimate relationships were discussed. Many of the 24 participants who had children reported “staying alive” for their children as a primary goal; the childless women reported an urgency to have children. Of the 21 who had a breast cancer diagnosis, the youngest was 24 years old, and several said knowledge of their genetic risk influenced their decision to have the unaffected breast removed prophylactically.

CONCLUSIONS: A sense of being different and not understood was expressed in these interviews. These findings suggest that nurses should be aware of psychosocial issues, especially those surrounding marriage and childbearing, in their interactions with young women who carry a BRCA1 or BRCA2 gene mutation.

Psychological consequences of being young and BRCA positive have been variously reported. A recent study indicates that younger age is associated with both higher perceptions of stigma and cancer-specific anxiety. Younger age has also been associated with an increased sense of urgency in both life partnering and childbearing. Research also indicates that the typical social supports of family, friends, and coworkers may not be adequate when a young woman is told she carries a mutation in a BRCA gene.

METHODS AND DATA ANALYSIS

I used grounded theory to explore how young women live with a BRCA mutation. In 2006, I recruited women from two Web sites offering support to women with breast cancer and/or BRCA gene mutation. My inclusion criteria were women ages 18 to 39 who had had the genetic test for the BRCA mutation and had received their results. I interviewed 59 women. My methods of recruiting and interviewing the sample are reported elsewhere. I have previously published articles examining the data collected in these interviews: one on the life trajectories of the young women who tested positive and another on the lives of those who were single at the time of testing.

Of the 59 women interviewed, 44 had received a positive test result. In this report, I explore three characteristics of the 44 who tested positive: whether or not they were married, had children, or had breast cancer. The women were from 22 states in the United States.
Marital status. Thirty-one women were married and the rest were single. (None of the participants described having a same-sex partner or living with a partner.) For young, single women, being BRCA positive is complex in ways not generally experienced by women in this age group.3 Questions about disclosure included when to tell a new partner about genetic risk or how early in a relationship to discuss having children or plans for prophylactic mastectomy or oophorectomy. For example, one 31-year-old said,

Do I tell him all this on the first date? Do I wait? Do I hide this from him now or not? I don’t know; it’s kind of a tricky issue. . . . How much information do you divulge?

Being married had certain consequences as well. One of the most poignant expressions came from a woman who visited her in-laws after being told she carried a BRCA mutation:

I didn’t tell my in-laws. I remember avoiding them when we came into the house and waiting upstairs while my husband told them the bad news. I felt like damaged goods, somehow, as if they would think my husband had made a mistake marrying me—that I was such a liability to him or to them.

In participants’ relationships overall, conversations about living with risk, having a cancer diagnosis, and having had or planning to have a prophylactic surgery were complex. It was a challenge to them to help others comprehend the degree of risk they felt.

Presence or absence of children. Of the 20 participants who had no children at the time of the interviews, 12 were single. Many women who had children spoke of “staying alive” as their primary goal related to their children:

What triggers me the most is my children, they are four and six now . . . and I don’t want them to have to go through what I did when I lost my mother. . . . I want to watch my children grow and flourish, I want to watch them get married, I want to be a grandmother, I want to be old and gray with my husband. I want life!

One consequence of this goal was making the choice to have prophylactic surgery sooner rather than later:

For my sake, my husband’s sake, and my kids’ sakes, I feel that I have to do these preventive procedures. If I didn’t, I would feel negligent. How could I tell my kids someday that I got breast cancer even knowing that I had the BRCA gene mutation and hadn’t done all I could do to prevent it?

Participants who had children also described the impact of knowing they may have passed on the mutation, which caused feelings of guilt and in some cases led to a decision to limit the number of children:

I now feel huge amounts of guilt knowing that I may have passed this on to my little girl, so how could I now knowingly do it to another child? So that is my logical answer and reason we are done [having children], but emotionally I still deal with it every day.

Although participants with children worried about the possibility of passing on the mutation, none regretted the choice to have children.

**Table 1.** Participant Characteristics (N = 44)

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age</strong></td>
<td></td>
</tr>
<tr>
<td>18–28 years</td>
<td>14 (32)</td>
</tr>
<tr>
<td>29–39 years</td>
<td>30 (68)</td>
</tr>
<tr>
<td><strong>BRCA mutation</strong></td>
<td></td>
</tr>
<tr>
<td>BRCA1</td>
<td>30 (68)</td>
</tr>
<tr>
<td>BRCA2</td>
<td>15 (34)</td>
</tr>
<tr>
<td><strong>Breast cancer</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>21 (48)</td>
</tr>
<tr>
<td>No</td>
<td>23 (52)</td>
</tr>
<tr>
<td><strong>Ovarian cancer</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>0 (0)</td>
</tr>
<tr>
<td>No</td>
<td>44 (100)</td>
</tr>
<tr>
<td><strong>Marital status</strong></td>
<td></td>
</tr>
<tr>
<td>Married</td>
<td>31 (70)</td>
</tr>
<tr>
<td>Unmarried</td>
<td>13 (30)</td>
</tr>
<tr>
<td><strong>Children</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>24 (55)</td>
</tr>
<tr>
<td>No</td>
<td>20 (45)</td>
</tr>
<tr>
<td><strong>Race</strong></td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>43 (98)</td>
</tr>
<tr>
<td>African American</td>
<td>1 (2)</td>
</tr>
<tr>
<td><strong>Risk-reducing mastectomy</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>23 (52)</td>
</tr>
<tr>
<td>No</td>
<td>21 (48)</td>
</tr>
<tr>
<td><strong>Risk-reducing oophorectomy</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>16 (36)</td>
</tr>
<tr>
<td>No</td>
<td>28 (64)</td>
</tr>
</tbody>
</table>

*One participant had both mutations.*

States (n = 41) and from Canada (n = 3). Other sample characteristics are summarized in Table 1.

**RESULTS**

**Marital status.** Thirty-one women were married and the rest were single. (None of the participants described having a same-sex partner or living with a partner.) For young, single women, being BRCA positive is complex in ways not generally experienced by women in this age group.3 Questions about disclosure included when to tell a new partner about genetic risk or how early in a relationship to discuss having children or plans for prophylactic mastectomy or oophorectomy. For example, one 31-year-old said,
Few participants decided against having children because they carried the BRCA mutation, but some considered stopping the mutation with their generation:

Testing positive for the BRCA2 gene [mutation] has made me consider the possibility of not wanting a child because of passing the gene along, but then I realized that there are so many options and in 10 years there will hopefully be more.

Having children gave these young women not just a desire but also a responsibility to stay alive—to take whatever clinical steps would increase the likelihood that they would escape cancer.

Others who had not yet had children described a “schedule” they felt they needed to keep to but also expressed ambivalence about how to manage that. One woman who’d had a prophylactic mastectomy described a “small window” of time, between ages 28 and 35, in which to have the two children she and her husband had planned for:

Right now, we cannot afford to have children but are hoping to save up some money and be ready by around 30. Right now, my husband and I would also like some time to ourselves postsurgery and prechildren to just enjoy being married to one another. We are not really emotionally ready to be parents yet.

Having a breast cancer diagnosis. Twenty-one of the participants had received a breast cancer diagnosis. Among them, the youngest was 24 years old, five were single, and 14 had children. Participants reported dramatic consequences:

Being young, I had different concerns than those who had already had their children and were older. I was still coming off a miscarriage when I was diagnosed with breast cancer.

Not only was the diagnosis difficult to take in but decisions had to be made quickly regarding the timing of surgery, chemotherapy, and radiation:

Yeah, we moved very, very quickly and so . . . by the time I was diagnosed and had the mammogram and the biopsy results on Thursday, the following Monday I went in and had a mastectomy, so it was like whoa, whoa, whoa!

Many of the participants with breast cancer did not have their genetic test prior to diagnosis and initial surgery (lumpectomy or single mastectomy), so that when they received the news that they carried a BRCA mutation they then opted to have the unaffected breast removed prophylactically:

I originally had a lumpectomy. Discovered that I was BRCA1 [positive] about a month later. My oncologist strongly recommended that I have a bilateral mastectomy with reconstruction after I finished chemo and I did. He has also recommended that I have my ovaries removed by the time I am 40. Which I will do as well.

While having breast cancer and being BRCA positive is difficult, having the mutation without a cancer diagnosis also had consequences for these young women. When asked what being BRCA positive meant, one participant summed it up well:

To me, it means daily thinking of the possibility of getting cancer. It consumes every medical thought. Even major life decisions. You don’t HAVE cancer; there aren’t walks for you, there aren’t many support groups. . . . It isn’t cancer, it isn’t not cancer. It is something in between that is about constant worry, checking, medical exams. A life filled with research and doctors.

A sense of being different and largely misunderstood pervaded these interviews. Having experiences outside those of one’s peer group can alter how a young woman thinks about herself. The overall experience was perhaps best summed up by this 35-year-old:

Even though I try not to worry, I still do. I worry that I will get cancer. I worry that I could have the recommended surgeries and have a difficult time living with the emotional and physical aftermath. I worry that I’ll make the wrong decision and hurt myself and my family. To me it’s like knowing you’re going to have a car crash and you can see it coming, but the option is to crash into something else . . . so what do you do?

DISCUSSION
Knowing that one carries a BRCA mutation when young can have many consequences. On one hand, it can empower a young woman to ameliorate her risk of developing cancer and work to detect cancer in its early stages. On the other hand, it can cause her to worry for herself, her children, and other family members. And knowing about genetic risk tends to lead to making fairly stark choices. For example, some participants said that their need to live for their children influenced their decision to move quickly in getting a prophylactic mastectomy. Other studies have reported similar findings.
Being diagnosed with breast cancer before being tested for a BRCA mutation can also dictate decision making.  
Initially, participants said, the breast cancer diagnosis was more consequential than the news of the BRCA mutation. But after breast cancer treatment ended, the BRCA mutation became more of a focus, and with it came a greater sense of vulnerability and risk.

It was clear that being young, single, and BRCA positive presented relationship challenges, but no pattern was evident as to when or how to disclose this information in a relationship. Also, other studies have found what some of the married participants in this study identified: a sense of being less than “perfect” to family members. Everyone carries gene mutations, most of them harmless, but knowing that one carries a BRCA mutation, associated with such a high-profile disease, can be particularly difficult.

Nurses should appreciate how, in young women, knowledge of genetic risk affects intimate relationships, reproductive concerns, and health insurance options.

For BRCA-positive women wanting to avoid pregnancy, oral contraceptives may be considered. The most recent meta-analysis of BRCA-positive women with breast cancer (n = 2,853) and without it (n = 2,954) found no associated risk of breast cancer development with oral contraceptives formulated after 1975. Recent data show that women with a BRCA1 or BRCA2 mutation who use oral contraceptives have about a 50% reduced risk of ovarian cancer. For young women who have opted for prophylactic mastectomy but are not ready for prophylactic oophorectomy, oral contraceptives may be an option.

Limitations. Recruitment bias may have affected these findings. I recruited all participants online from sites offering support and information to patients; the sample may have been exceptionally motivated to take action or seek help and therefore not representative of all young women who carry a BRCA gene mutation. Also, I collected and analyzed the data alone; using investigator triangulation would have ensured the trustworthiness of the data. These factors may limit the validity of the findings, although many are supported by other studies.

Implications
By exploring the experiences of young women who carry a BRCA mutation, nurses can become sensitive to the complex issues these women face. The American Association of Colleges of Nursing in the 2008 Essentials of Baccalaureate Education for Professional Nursing Practice included knowledge of genetics and genomics as critical for practicing nurses. A consensus panel of experts in education, clinical nursing, and research has developed the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics, which states, the competencies are based on the state of the evidence available at the time they were developed and reflect the MINIMAL amount of genetic and genomic competency expected by every nurse. These competencies reflect a consensus and are NOT from any federal agency or single nursing organization, and they are applicable to the practice of all registered nurses regardless of academic preparation, practice setting, role, or specialty.

While it’s critical that nurses grasp the basic science of genetics, the findings presented here suggest that nurses must look beyond the science to the patient and family. A young woman testing positive for a BRCA mutation never presents alone; even if she’s the only person in the room, the nurse is assessing an extended family because breast cancer can cross many generations. Nurses should appreciate how, in young women, knowledge of genetic risk affects intimate relationships, reproductive concerns, and health insurance options. Not discounting a young woman’s anxiety about screening, for example, can be an important aspect of caring for her.

In a recent survey of attendees at the Council for the Advancement of Nursing Science State of the Science meeting held in 2008, 79% of baccalaureate programs and 85% of graduate programs did not meet the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics. Multiple resources are available for nursing faculty to use when incorporating this information into their courses. One is the Genetics/Genomics Competency Center for Education (www.g-2-c-2.org), funded by the National Human Genome Research Institute of the National Institutes of Health, which hosts an online repository of training and educational materials for health care professionals, including nurses.

The findings presented here suggest that good nursing practice might include the following, as supported by the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics:  
- Understand the types of mutations and inheritance patterns for specific diseases.  
- Help patients understand the recommended timelines for preventative options after detection of a BRCA mutation.  
- Listen to the young woman’s concerns.  
- Acknowledge that she faces complex and unusual choices for her age group.
• Recommend support groups specific to young women.
• Appreciate that young women with a BRCA mutation may seem hypervigilant and need frequent contact with clinicians.
• Recognize that this population may be “too young to screen” (not in the typical age group for mammography, for instance) but not too young to get cancer.
• Help the patient in getting insurance coverage for screening.
• Assess the need for additional psychosocial support.
• Suggest ways to discuss the issue of a “family mutation” in the patient’s family, for example, role-play a “how would you tell your 16-year-old sister” scenario.
• Find answers to questions you can’t answer.
• Make referrals to genetics professionals who can discuss the implications, screening recommendations, follow-up protocols, and other issues specific to living with genetic risk.

In my interviews I have found this population to be resilient, courageous, and thoughtful. Nurses can and should help these young women as they navigate relatively uncharted waters. ▼

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