Breast cancer genetic testing, as a medical management tool, can detect specific harmful hereditary mutations in BRCA1 and BRCA2 and provide women with a more precise estimate of their projected lifetime risk for the development of breast and ovarian cancer. BRCA1 and BRCA2 genetic testing is not offered to everyone because of the rarity of these mutations in the general population (National Cancer Institute, 2015). Geneticists and genetics counselors offer it to individuals who meet specific hereditary breast and ovarian cancer syndrome testing criteria. A comprehensive listing of 2015 hereditary breast and ovarian cancer testing criteria can be found on the National Comprehensive Cancer Network (2015) website (http://bit.ly/1Nubll0).

A meta-analysis by Chen and Parmigiani (2007) estimated the average cumulative risk for women with a BRCA1 mutation at 70 years of age to be 57% (95% CI [0.47, 0.66]) for the development of breast cancer and 40% (95% CI [0.35, 0.46]) for ovarian cancer. The average cumulative lifetime risk for the development of breast and ovarian cancer in those who carry a harmful BRCA2 gene mutation is 49% (95% CI [0.4, 0.57]) and 18% (95% CI [0.13, 0.23]), respectively. Women who test positive for a harmful BRCA1 or BRCA2 gene mutation may find that this information enables them to make more informed decisions about risk-reducing and intensive screening options (i.e., mammography, magnetic resonance imaging, and clinical breast examinations), as well as chemoprevention drugs (National Cancer Institute, 2015). However, the knowledge gained through breast cancer genetic testing is not simply a means to make informed decisions about risk-reducing and intensive
surveillance practices to prevent or detect breast cancer; knowledge from this technology has the power to transform the self, change the way health and illness are defined, and, at the same time, raise several ethical issues for healthcare professionals. Nurses, particularly those working in oncology, palliative care, and cancer genetics, are well situated to listen, advocate for, and explore the questions and concerns of women who are considering BRCA1 and BRCA2 genetic testing. This article will explore how the power of genetic knowledge may alter meanings attached to the breast and how health is defined, some of the ethical concerns and social forces raised in the literature, and implications for nursing practice.

**BRCA Genetic Technology**

Burch (1986) urged healthcare professionals to deeply contemplate the outcome of predictive genetic knowledge. “Regardless of its demonstrable benefits, each one comes with a cost and is not just a means to an end, but is implicated in a transformation of ourselves and our world” (p. 13). Reflecting on the words of Burch, knowledge gained through BRCA genetic testing has the power to change the meanings we attach to the breast and how we define health.

For centuries, the breast has embodied an “existential tension between Eros and Thanatos—life and death—in a visible and palpable form” (Yalom, 1998, p. 8). As life-giving, the paintings of the nursing Virgin Mary show the baby Jesus suckling at an exposed breast, representing the breast as a sacred vessel that gives milk to sustain life. On the other hand, the highly visible and palpable nature of breast cancer has long represented the breast as a location harboring the potential to end one’s life (Yalom, 1998). Within this existential tension, the breast also has been symbolized as an erotic symbol of female sexuality—presented as orbs of pleasure for the other to gaze upon and caress—as well as a visible symbol of a woman’s womanliness and an integral part of a woman’s sense of self (Yalom, 1998; Young, 2005).

For some women, the threat of hereditary breast cancer changes the meanings they attach to their breasts. For example, Crotser and Dickerson (2010) found that several women in their study expressed how their perception of their breasts changed from a healthy part of their body to an area that harbored a “threat of illness and, for some women, a threat of death” (p. 731) after learning of a harmful genetic mutation in the family.

In several studies in which the experience of living with a high hereditary risk for breast cancer has been explored within populations of unaffected women (those with no personal history of breast and/or ovarian cancer) identified as carriers of a deleterious BRCA1 or BRCA2 mutation, the presence of hereditary breast cancer risk brought with it feelings of mistrust and anxiouslyness toward the breast for some women. In addition, the threat of a future diagnosis of breast cancer has been shown to alter a woman’s sense of self, temporality, and relations with others (Crump, Fitzgerald, & Legge, 2010; Hamilton & Hurley, 2010; Hamilton, Williams, Skirton, & Bowers, 2009; Werner-Lin, 2008). Therefore, the representing of the breast as a part of the body that can no longer be trusted because of a hereditary threat of future breast cancer reaffirms Heidegger’s (1977) belief that technology is not neutral in that it has the potential to reorganize and change the way people think and know the world.

**Definitions of Health**

Knowledge about one’s potential risk for disease may disturb customary definitions of health and illness. Gibbon (2006) presented a narrative of one woman to illustrate how attention to risk through vigilance and awareness may change how health is defined. In her narrative, the woman disagreed with the notion that “health is when you don’t have symptoms” (Gibbon, 2006, p. 160). The importance of being vigilant and aware of the potential for a hereditary illness was raised in the public domain when actress Angelina Jolie wrote about her decision to undergo a bilateral prophylactic mastectomy after learning that she carries a harmful BRCA1 gene mutation. Although Jolie highlighted that a woman’s decision to undergo genetic testing or pursue risk management options is a very personal one, questions have been raised as to how her celebrity status may affect a woman’s decision to undergo BRCA1 and BRCA2 genetic testing (Grady, Parker-Pope, & Belluck, 2013; Jolie, 2013; Lerner, 2003).

Thinking of oneself as unhealthy because of the presence of a genetic predisposition to cancer may result from a vagueness in the perception of genetic disease that lies in beliefs about what constitutes a disease. One of the questions faced is how to categorize a person with a predisposition for a potentially fatal disease. Di Pietro, Giuliani, and Spagnolo (2004) pondered whether this person would be perceived by the medical community and society as “ill, healthy, asymptomatic but ill, or as an ‘unpatient’—since he/she may develop symptomatic disease in the future” (p. 67). Labeling women as ill or asymptomatic but ill may unintentionally feed into a prevailing myth that the knowledge gained through predictive genetic testing is deterministic. Therefore, the question of how asymptomatic women with a genetic predisposition for breast cancer are labeled in the medical community and society is one that requires thoughtful consideration as predictive medicine advances.

The preceding discussion suggests several ways in which knowledge gained through breast cancer genetic testing and social forces may influence not only a transformation of a woman’s sense of self, but also one’s definition of health. To help women decide whether to pursue genetic testing, nurses must be attuned to the ethical dilemmas involved in the decision.

**Principles of Biomedical Ethics**

Beauchamp and Childress (2008) introduced a framework to guide moral reasoning among healthcare practitioners. Their framework builds on traditional ethical principles, such as beneficence and nonmaleficence (referring to the benefits and harms), and presents two additional principles—respect for autonomy and justice. *Respect for autonomy* supports an individual’s right to “govern herself, to formulate and pursue her own life plans, goals, and values” (Fisher, 2009, p. 12), while *justice* refers to the “duty to give each individual equal consideration based on the contextual details of the situation” (Fisher, 2009, p. 16).
Weighing the Benefits and Harms of BRCA Genetic Testing

Several researchers have identified psychological benefits to women who are undergoing testing or living with a harmful mutation. Possible positive outcomes of knowing one’s genetic status have been voiced by women as personal growth, feeling more in control over their health, taking action to attain better health, involvement in philanthropic activities, and feeling more appreciated, as well as an altruistic sense that the benefit of genetic knowledge reaches beyond the individual and family because it may contribute to the advancement of medical science (Dagan & Goldblatt, 2009; d’Agincourt-Canning, 2006; Proulx et al., 2009; Radner, 2011).

Other psychological benefits to women who undergo testing and find out they are carriers of a harmful mutation may be derived through a deeper connection with other family members. When Hamilton, Williams, et al. (2009) examined how unaffected women living with a personal BRCA gene mutation for at least four years adapted to their changed reality, some women reported an enriched and stronger connection between themselves and their family members. However, the reverse may occur. For example, one woman in a study by Hamilton, Williams, et al. (2009) described how she felt isolated and at a distance in her relations with her siblings. She said, “My sisters and brother really didn’t know how to support me . . . in the beginning they didn’t know how, or emotionally could not support me in the way that I needed. That hurt me deeply (and if I let myself, still does)” (Hamilton, Williams, et al., 2009, p. 279).

Similarly, Douglas, Hamilton, and Grubs (2009) found that several women in their study voiced a sense of being more or less connected to relatives after disclosing their BRCA mutation status. For some, negative reactions by family members to the presence of a harmful mutation were experienced through avoidance of the topic and a lack of support (Douglas et al., 2009). Although some women in the preceding studies voiced a stronger connection to family members once they disclosed their positive BRCA mutation status, the possibility of being stigmatized and distanced from relatives once a harmful mutation is disclosed is a potential reaction to consider.

The potential for psychological consequences has been studied extensively within the field of medical genetics. A meta-analysis of 20 short-term and long-term studies that followed women who underwent BRCA genetic testing for up to one year after disclosure of results found that increases in anxiety and cancer-specific distress returned to pretest baseline levels over time (Hamilton, Lobel, & Moyer, 2009). Although this evidence is reassuring, other researchers have found that some women experience varying degrees of ongoing distress related to their genetic status (Crump et al., 2010; Radner, 2011; van Oostrom et al., 2003). For example, Hamilton, Williams, et al. (2009) identified conflicted emotions in a group of unaffected BRCA-positive women as they spoke of moving between feelings of being “in control” through surveillance and risk-reducing surgeries and feelings of mistrust and anxiousness about their bodies. Similarly, in a small group of women from New Zealand, in which half of the women tested positive for a harmful mutation, the participants reported heightened feelings of mistrust, anxiety, stress, paranoia, and mindfulness that this information would always lurk at the back of their minds (Crump et al., 2010). The complex emotional and psychological reactions that women may experience after learning they are carriers of a harmful mutation necessitates thoughtfulness and reflection on a wide array of possible outcomes.

Potential for Discrimination

For some women, the decision to pursue or decline BRCA genetic testing may be influenced by concerns about possible discriminatory practices within the workplace, when applying for life or health insurance, and when considering the cost of genetic testing (Armstrong et al., 2000; Benkendorf et al., 1997; Peterson, Milliron, Lewis, Goold, & Merajver, 2002). For instance, Armstrong et al. (2000) identified fear of health insurance discrimination as a very important factor in the decision to decline an offer for testing in 47% of eligible women who declined BRCA genetic testing. Other factors considered salient in the decision to decline an offer for genetic testing in these women included fear of discrimination in employment (28%) and of life insurance implications (42%) (Armstrong et al., 2000).

The potential for health and life insurers to use BRCA genetic information to increase premiums or deny applications for coverage among female carriers of a BRCA gene mutation prompted the U.S. government to legislate the Genetic Information Nondiscrimination Act of 2008 (H.R. 493). In Canada, Bill S-201—a bill that seeks to prohibit genetic discrimination—is before the Standing Senate Committee on Human Rights (Parliament of Canada, 2015).

In addition, the potential exists for discriminatory practices in the workplace because women who are carriers of a BRCA gene mutation request more time away from work to undergo intensive biannual surveillance testing or recover from risk-reducing prophylactic and reconstructive surgeries (Surbone, 2004). Access to testing is another issue to consider. In the United States, the cost for BRCA genetic testing can reach several thousands of dollars—making the cost of this test prohibitive for women who cannot afford it or do not have coverage through their health insurance provider. In Canada, however,
BRCA genetic testing is publicly funded for individuals who meet the eligibility criteria (National Cancer Institute, 2015).

Respect for Autonomy

Genetics counselors and geneticists provide individuals with sufficient information in a nondirective manner so that a decision can be made autonomously by the individual considering testing. Given that genes are shared within families, the potential for the presence of an inheritable BRCA gene mutation may “foster a novel appreciation of morality based on relational rather than on individual values” (Surbone, 2004, p. 162). In other words, the view that morality may be a product of one’s relations with others raises questions as to whose values are being served within the decision to undergo genetic testing. This conundrum requires thinking about how family dynamics may influence, in a positive or detrimental fashion, an individual’s right to choose.

For example, familial pressures that are placed on other family members to pursue genetic testing have the potential to create negative changes in family dynamics. In a study by Freedman (1998), familial concerns arose in one situation in which an unaffected young adult woman attending a consultation with her mother (who had been diagnosed and treated for breast cancer) was interested in knowing what her mother’s genetic status was but was not interested in knowing her own genetic status. In this situation, the desire of the mother to know the genetic status of herself and her daughter created conflict in their relationship.

Dancyger, Smith, Jacobs, Wallace, and Michie (2010) found that some of the relatives in their study were torn between their sense of a family duty to get tested and the pressure they felt when they were not sure they wanted to be tested. Although genetic testing is not a legal obligation, the sense that it is a family duty was voiced by some of the participants as a force that was so strong that they felt that their rash decision to be tested did not allow them time to fully consider the consequences of an unexpected positive result, culminating in a negative emotional reaction. On the other hand, for many women, the decision to undergo BRCA genetic testing is a decision that interconnects personal interest with responsibility toward their offspring and other family members in a positive way (d’Agincourt-Canning, 2006; Foster, Watson, Moynihan, Arder-Jones, & Ecles, 2002).

Conclusion

This article explored how knowledge of one’s genetic status may change the way a person sees and knows the world and described some of the psychosocial issues that may influence a woman’s decision to pursue genetic testing. An awareness of these issues may enhance the nurse-patient relationship as nurses help these women explore potential benefits and repercussions of knowing one’s genetic status, deal with social and familial forces that may influence their decision, and work through ethical dilemmas that may arise because of the sharing of genes. For more information about breast cancer genetic testing and the psychosocial issues related to it, see Figure 1 for a listing of educational resources. The aim of this article was to enliven an ongoing ethical dialogue to understand the overall impact of knowing about one’s BRCA1 and BRCA2 genetic status on the self and to arrive at decisions that are, in the end, based on what is ethically best for each woman involved.

Implications for Practice

- Find out what women think and feel are the benefits and repercussions of knowing their genetic status.
- Use insights from a review of the social and familial forces that may influence a woman’s decision to undergo or decline an offer for genetic testing to inform future discussions with women contemplating genetic testing.
- Ask women to describe how family members and friends reacted when they told them that they were contemplating BRCA1 and BRCA2 genetic testing.

References


