Breast Cancer Genetic Testing: More Than a Medical Management Tool

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Background: Knowing whether a harmful hereditary mutation exists in BRCA1 and BRCA2 can enable women to make informed decisions regarding surveillance and surgery options to manage risk. Given the attention in the media about BRCA genetic testing, nurses need to revisit how this knowledge may affect a woman’s sense of self and the forces that may influence this decision.

Objectives: This article aims to understand how complex the decision to undergo genetic testing may be for some women by exploring the impact of genetic knowledge on the self, changes to customary definitions for health and illness, and ethical issues and social forces that may influence genetic testing decisions.

Methods: A review of the literature was undertaken to understand how genetic knowledge may alter meanings attached to the breast and how health is defined, and to identify ethical concerns and social forces that may affect a woman’s decision to undergo or decline an offer for genetic testing.

Findings: An understanding and awareness of the potential benefits and harms of BRCA1 and BRCA2 genetic testing, as well as the social forces that may influence a woman’s decision to undergo or decline an offer for genetic testing and the commitment to remain open to the uniqueness of each woman’s situation, may enhance the nurse-patient relationship and result in a decision that is ethically in the best interest of the patient.

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 early detection measures, such as prophylactic surgeries 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