Decision Making After BRCA Genetic Testing: Down the Road of Transition

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The purpose of this study was to evaluate women who have completed hereditary cancer risk assessment and BRCA genetic testing to determine if they considered themselves prepared to proceed with decision making regarding cancer screening and prevention options. Levels of decisional conflict were explored, as was their preference for information delivery. The prospective, descriptive survey was conducted at a breast and clinical genetics clinic at a comprehensive cancer center in the northeastern United States. Twenty-seven female participants completed the Preparation for Decision Making scale, Decisional Conflict Scale, and a demographic questionnaire. Scores were consistent with high levels of preparation for decision making and low decisional conflict. The face-to-face approach was the preferred method for information delivery. Subgroup analysis demonstrated a difference in the measured objectives based on cancer status but not based on BRCA status. The current information delivery approach in meeting the decision-making needs of women considered to be at increased risk for hereditary breast and ovarian cancer.

Five to ten percent of all breast and ovarian cancers have an identifiable genetic component (Nelson, Huffman, Fu, & Harris, 2005). To date, the most commonly identified genetic cause of hereditary breast and ovarian cancer (HBOC) are mutations in either the breast cancer-susceptibility gene 1 (BRCA1), located on chromosome 17 (Hall et al., 1990; Miki et al., 1994), or breast cancer-susceptibility gene 2 (BRCA2), located on chromosome 13 (Wooster et al., 1994). These function as tumor suppressor genes, and a mutation in one of the BRCA genes puts the carrier at increased risk for developing certain cancers, including breast and ovarian. BRCA mutations are inherited in an autosomal dominant manner through either parent. Women considered at risk for HBOC, because of a personal or family history of disease, often complete hereditary cancer risk assessment and BRCA1 and BRCA2 genetic testing to clarify their risk of developing certain cancers and better inform subsequent decisions.

The lifetime risk of breast cancer for the general population is 12%, and the risk of ovarian cancer is 1.4% for women in the United States (Howlader et al., 2013). The risk of breast cancer increases to 45%–65% in women with a deleterious BRCA mutation, and the risk of ovarian cancer increases to 11%–59% for those women by age 70 years (Chen & Parmigiani, 2007). To date, no available genetic tests guarantee the identification of all cancer-predisposing BRCA mutations; therefore, a certain percentage of mutations may be missed (Petrucelli, Daly, & Feldman, 2011). Women who complete BRCA genetic testing (with no family history of a BRCA mutation) and receive negative test results are advised that the BRCA4 genetic test results are “uninformative.” They are still at an elevated risk of breast and ovarian cancer based on their personal and/or family history of disease. Given the uncertainties still present in genetic test results, healthcare providers face a challenge in giving women accurate and useful information and advice on which