Diff erences Between Women Who Pursued Genetic Testing for Hereditary Breast and Ovarian Cancer and Their At-Risk Relatives Who Did Not

Maria C. Katapodi, RN, PhD, Laurel Northouse, RN, PhD, FAAN, Penny Pierce, RN, PhD, FAAN, Kara J. Milliron, MS, CGC, Guipeng Liu, MS, and Sofia D. Merajver, MD, PhD

Breast cancer is a leading cause of death among women in the United States (American Cancer Society [ACS], 2011). Mutations in the breast cancer 1 and breast cancer 2 genes (BRCA1 and BRCA2) predispose carriers to hereditary breast and ovarian cancer (HBOC) syndrome. Carriers of the BRCA1 and BRCA2 mutations are at significantly higher risk for developing breast cancer (55%–85% versus 12%) and ovarian cancer (20%–60% versus 2%) compared to the general population (ACS, 2011). Twenty to 25% of new breast cancer cases are characterized as familial because they are associated with a strong family history in the absence of a known mutation (ACS, 2011).

Genetic testing identifies at-risk individuals and enables them to make informed decisions about risk management (e.g., chemoprevention, risk-reducing surgery, intensive surveillance) (Eisinger et al., 2001; Finch et al., 2006; Metcalfe et al., 2008; Schrag, Kuntz, Garber, & Weeks, 1997). Patients who already have developed breast or ovarian cancer benefit from knowing their mutation status before making surgical decisions; asymptomatic individuals might use the information to manage their risk and make informed life decisions (e.g., reproduction). Prior research indicates a high interest in genetic testing for cancer susceptibility genes; however, rates of actual uptake of BRCA1 and BRCA2 testing vary greatly, from 26%–80% among at-risk individuals (Halberg, Kessler, Stopfer, Domchek, & Wilejto, 2006; Ropka, Wenzel, Phillips, Siadaty, & Phlilbrick, 2006). Moreover, up to 60% of those who get tested may not seek their results (Pasacreta, 2003).

Studies have focused primarily on identifying individual predictors of genetic testing (e.g., perceived risk, knowledge of gene inheritance, psychological distress) (Pasacreta, 2003). However, evidence points to possible links between familial factors and decisions to pursue genetic testing (Lerman, Croyle, Tercyak, & Hamann, 2002; Peterson, 2005; Wilson et al., 2004). Little is known about the interplay of individual and familial factors and...