

# Differences 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

**B**reast 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 (Halbert, Kessler, Stopfer, Domchek, & Wileyto, 2006; Ropka, Wenzel, Phillips, Siadat, & Philbrick, 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

**Purpose/Objectives:** To (a) examine differences in appraisals of hereditary breast and ovarian cancer (HBOC), psychological distress, family environment, and decisional conflict between women who pursued genetic testing and their at-risk relatives who did not, and (b) examine correlations among appraisals of HBOC, psychological distress, family environment, and decisional conflict regarding genetic testing in these two cohorts of women.

**Design:** Descriptive, cross-sectional cohort study.

**Setting:** Two clinics affiliated with a major research university in the midwestern United States.

**Sample:** 372 women aged 18 years and older. 200 pursued genetic testing for *BRCA1* and *BRCA2* mutations (probands) and 172 of their female relatives who had a greater than 10% prior probability of being a mutation carrier but had not pursued testing.

**Methods:** After providing informed consent, probands and relatives were mailed self-administered questionnaires.

**Main Research Variables:** Perceived risk, knowledge of HBOC risk factors and modes of gene inheritance, perceived severity, perceived controllability, psychological distress, family relationships, family communication, and decisional conflict about genetic testing.

**Findings:** T tests revealed that probands perceived higher risk and had more psychological distress associated with breast cancer. Probands had more knowledge regarding risk factors and gene inheritance, and greater decisional conflict regarding genetic testing. Relatives reported higher perceived severity and controllability. No differences were observed in family relationships and family communication between probands and relatives. Pearson correlations revealed different patterns in knowledge, perceived controllability, family relationships, and decisional conflict between probands and relatives.

**Conclusions:** Significant differences exist between women who pursue genetic testing and those who do not. The family environment influences adjustment to HBOC and decisions about genetic testing.

**Implications for Nursing:** Enhancing the family communication process about HBOC can provide informational and emotional support to high-risk women and promote decision making about genetic testing.