Learning About a Twist in the Road: Perspectives of At-Risk Relatives Learning of Potential for Cancer

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The following statement illustrates the complexity of communicating newly discovered inherited breast and ovarian cancer risk in families.

We aren’t really that close . . . and she basically sent out an e-mail to the family . . . and she said, “The good news is that I am done with chemo and everything is fine, . . . the bad news is I got my genetic test back and I have this mutation, and you probably have it too, so go get tested.” . . . It came out of the blue.

More than 200,000 American women are diagnosed with in situ or invasive breast cancer every year. About 5%–10% of breast cancers are caused by inherited mutations in genes such as BRCA1 or BRCA2 (American Cancer Society, 2009). Each first-degree relative of a mutation carrier has a 50% chance of inheriting the same mutation. Women with a BRCA mutation have a 14%–87% chance of a breast cancer diagnosis by age 70 and a 10%–68% chance of ovarian cancer (National Cancer Institute, 2010). Individuals can benefit from knowing their BRCA status because the knowledge can assist them with decision making for screening and surveillance, chemoprevention, lifestyle changes, and risk-reducing surgery. In secondary prevention, the information is used for definitive treatment decisions (Schwartz, Peshkin, Tercyk, Taylor, & Valdimarsdottir, 2005) because women with BRCA mutations are at significantly increased risk of future contralateral breast cancer (Smith & Issacs, 2006–2007).

Genetic testing is not without limitations, including the potential for inconclusive results and a lack of certainty because of the variable penetrance and expressivity of BRCA mutations. Additionally, risk-reducing procedures such as prophylactic mastectomy and oophorectomy do not guarantee a future without breast or ovarian cancer. The psychosocial consequences of breast cancer genetic testing are unfolding as researchers study the impact of genetic testing on worries, depression, anxiety, emotions, and family and social relationships. Studies exploring the psychosocial consequences of genetic testing for hereditary breast and ovarian cancer (HBOC) syndrome found that most women do not experience clinically significant levels of distress, depression, and anxiety (Crotser & Boehmke, 2009).