BRCA Genetic Testing: An RN’s Personal Story

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This story begins in early December 2011. I had received a diagnosis of ductal cancer in situ (DCIS), a noninvasive cancer staged at 0 in theducts of my left breast (National Cancer Institute [NCI], 2009). The breast surgeon from whom I was obtaining a second opinion discussed lumpectomy versus mastectomy and radiation. Toward the end of the visit, the surgeon suggested that I have a laboratory workup for vitamin D levels. Vitamin D has been found in some studies to reduce breast cancer risk, and levels of vitamin D also have been found to be low in women with breast cancer (NCI, 2012b). I was directed to the laboratory area. Suddenly, the surgeon burst in and announced that no BRCA results were on file at my gynecologist’s office. She informed me that if I carried the genetic mutation, “You will need to have a bilateral mastectomy and salpingo-oophorectomy.” The BRCA1 and BRCA2 genes (breast cancer susceptibility 1 and 2) usually shield women from certain cancers; however, if these genes possess mutations, the risk of hereditary breast and/or ovarian cancer is greatly increased (NCI, 2012a). A pamphlet produced by Myriad Genetic Laboratories (2009) states that those women with BRCA mutations have:

- As much as a 50% risk for developing breast cancer by age 50 years.
- As much as an 87% risk for developing breast cancer by age 70 years.
- As much as a 64% risk for developing a second breast cancer.
- As much as a 44% chance of developing ovarian cancer by age 70 years.

I looked at her in horror and bewilderment. I practiced in the field of psychiatric nursing for most of my 40 years and I could not even remember what “salpingo” meant. She suggested performing the BRCA test immediately in her office. I consented, and after the simple blood draw was completed, I stumbled out the door in a daze.

Although I was only a 10-minute drive from my home, I turned right when I should have gone left and vice versa. I was both crying and laughing at my attempts to navigate out of the maze that my familiar trip to work had suddenly become. I recalled the last words the surgeon uttered: “I guess you are living your worst nightmare.” She was referring to the facts of my family medical history.

My mother had died of breast and ovarian cancer at age 43, when I was 16, and her mother, the grandmother I never met, had died of cancer at age 37. I also was of Ashkenazi Jewish descent (Central and Eastern European background), a factor that increased my risk of inherited breast and/or ovarian cancer (Myriad Genetic Laboratories, 2009). In spite of these factors, my sister and I had sailed through our 40s and 50s cancer-free. We had professional careers, exercised regularly, consumed healthy diets, and obtained recommended health screenings that included mammograms.

That evening, when I crawled into bed, I attempted to recall how I had mistakenly concluded that I had been tested for the BRCA mutation and the results were negative. I remembered discussing the test with my gynecologist some years before; she agreed that I should be tested. My insurance company denied the claim; one factor was the prohibitive cost of more than $3,400. I appealed that decision and won, but unfortunately my provider discontinued her care of routine gynecologic patients and I was assigned to the nurse practitioner. The nurse...