



Understanding Hereditary Breast and Ovarian Cancer

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In 1994, the first cancer susceptibility gene to be associated with hereditary breast and ovarian cancer was identified and became known as breast cancer gene 1 (*BRCA1*). In 1995, a second gene was identified and became known as *BRCA2* (Calus, Schildkraut, Thompson, & Risch, 1996). With the identification of these two genes, a new field emerged in breast and ovarian cancer treatment that is transforming how a select population of people with these cancers is managed. However, the identification of these cancer susceptibility genes also has prompted new challenges and concerns, including identifying patients who are appropriate for genetic testing, informing patients about the advantages and disadvantages of genetic testing, and determining the appropriate treatment for those who test positive for a genetic mutation (alteration). In addition, ethical, legal, and psychosocial implications for patients and their families exist.

As knowledge of cancer genetics has increased, so has awareness of cancer genetics among healthcare professionals and the public. Oncology nurses can be instrumental in informing patients, families, and the public about the implications of these findings in terms of cancer prevention, early detection, and treatment. In addition, knowledge in this area has become essential to healthcare professionals when obtaining family histories to help identify patients who may have hereditary cancers.

Recognizing the importance of this area in oncology nursing, the Oncology Nursing Society (ONS) has developed position statements regarding cancer predisposition genetic testing and risk assessment counseling and the role of oncology nurses in cancer genetic counseling. ONS stated that oncology nurses at the general and advanced practice levels must be educated in genetic

testing and counseling and continuing education programs should be developed and provided to practicing oncology nurses. In addition, advanced practice nurses with specialized training in cancer genetics should provide comprehensive cancer genetic counseling (ONS, 2002a, 2002b).

Case Study

R.J. is a 32-year-old woman who was referred to a surgeon because of a palpable lump in her right breast. Her mammogram also identified this 2 cm x 2 cm mass. R.J. had an excisional biopsy that revealed a fibroadenoma. The surgeon noted that R.J. had a positive family history of breast cancer in her mother and a maternal aunt. A maternal cousin also had ovarian cancer. R.J. was of Irish and English heritage. The surgeon referred her to a genetic counselor for possible hereditary breast and ovarian cancer testing. After extensive counseling, R.J. consented to undergo testing. The findings were positive for a deleterious mutation (a change that results in an altered or damaged protein, rendering it unable to perform its function) in *BRCA1*.

1. What percentage of patients with breast cancer is thought to have a hereditary predisposition for breast or ovarian cancer?
 - a. Less than 2%
 - b. Approximately 5%
 - c. Approximately 13%
 - d. More than 50%
2. What factor in R.J.'s history does not suggest an increased risk for hereditary breast or ovarian cancer?
 - a. History of breast cancer in her aunt
 - b. History of breast cancer in her mother
 - c. Diagnosis of fibroadenoma
 - d. History of ovarian cancer in her cousin

3. Which option currently is not being used as a possible medical intervention for women with a *BRCA1* mutation?
 - a. Surveillance measures for breast and ovarian cancer
 - b. Bilateral prophylactic mastectomy and oophorectomy
 - c. Radiation therapy to the breast and abdomen
 - d. Chemopreventive measures for breast and ovarian cancer

Discussion

Question 1: Choice b, approximately 5%, is correct. Although all cancer is genetic because it results from mutations in genes that normally control cell division, most mutations are acquired during patients' lifetimes. However, in a small subset of people, mutations may be inherited. Researchers estimate that, in 2003, more than 200,000 women will develop breast cancer. Of these women, approximately 5% are thought to have a hereditary basis for the development of the disease (American Cancer Society, 2003).

Hereditary breast and ovarian cancer are associated with mutations in *BRCA1* or *BRCA2*. Genes are pieces of DNA that are found on chromosomes and are the functional unit of heredity that express inherited traits (see Figure 1). Each gene

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