Genetic Testing

Challenges and changes in testing for hereditary cancer syndromes

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BACKGROUND: The practice of genetic testing for hereditary cancer syndromes has changed dramatically in recent years, and patients often approach oncology nurses requesting information about genetic testing.

OBJECTIVES: This article aims to explore changes in cancer genetics, the role of genetics professionals in providing comprehensive genetic care, and the implications of these new developments in genetics for oncology nurses.

METHODS: A literature review was conducted and focused on articles about the updating of genetic tests with panel testing, insurance changes, alternative genetic counseling strategies, and direct-to-consumer genetic testing.

FINDINGS: Oncology nurses play an important role in identifying and referring patients, including those who have tested negative for hereditary susceptibility genes, to genetics professionals. Genetics professionals can assist with insurance issues, interpretation of test results, clarification when a variant of unknown clinical significance is detected, and recommendations for care based on personal and family history and testing results. Oncology nurses can assist families with understanding the limitations of direct-to-consumer genetic testing.

KEYWORDS
hereditary breast cancer; genetic testing; direct-to-consumer genetic testing

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TESTING FOR HEREDITARY CANCER SYNDROMES HAS CHANGED significantly since 2013 because of legal decisions and technological advances in genetic science. Hereditary cancer syndromes account for about 10% of all malignancies, and the identification of individuals in families who have a genetic predisposition for developing cancer can guide decisions about cancer prevention and early detection, including the use of prophylactic surgery (Weitzel, Blazer, MacDonald, Culver, & Offit, 2011). The evolution of genetic testing for hereditary cancer syndromes has progressed rapidly (see Figure 1).

The implications of genetic science in clinical oncology practice have become complicated; the simple days of oncologists ordering *BRCA* testing for their patients with breast cancer is over. Multiple genes associated with varying degrees of risk for developing breast and other cancers have been identified. Individuals who tested negative for the common *BRCA* mutations in the past, for instance, may harbor a less common hereditary susceptibility gene (Daly et al., 2017; Rich, Woodson, Litton, & Arun, 2015). The availability of testing for multiple genes using a panel of genes has led to more families discovering a susceptibility gene, but expanded panel testing has also created new challenges (Marcus et al., 2015). For example, many variants of unknown clinical significance are detected, and insurance requirements for coverage of panel testing are complicated. The interpretation of expanded panel testing is similarly complex and is best managed by a credentialed genetics professional; however, the shortage of credentialed genetics professionals has necessitated the use of alternative counseling strategies. In addition, direct-to-consumer testing has increased, and oncology nurses are often confronted with patient questions about it. Taken as a whole, these developments have made understanding cancer genetics more challenging for patients and healthcare providers.

Updating Genetic Testing

With the advent of panel testing, patients who previously underwent *BRCA* or other genetic testing with negative results should be offered more extensive testing (Graffeo et al., 2016). New panel testing includes testing for high penetrance breast and colon cancer genes (e.g., *TP53*, *PTEN*) and moderate penetrance genes (e.g., *ATM*, *CHEK2*, *PALB2*) (Castellanos et al., 2017; Economopoulou, Dimitriadis, & Psyrri, 2015). The National Comprehensive Cancer Network (2016) provides recommendations for risk management for high and moderate risk genes that are associated with hereditary breast and ovarian cancer syndromes. About 9% of patients with breast cancer who had tested negative for *BRCA* mutations and later underwent panel testing were found to have a pathogenic mutation in a breast cancer susceptibility gene (Moran et al., 2017).