Genetic testing for hereditary cancer syndromes is an integral component of oncology care. Various types of common errors that occur in the genetic testing process are presented in this article with actual clinical case examples and commentary. Genetic errors are expensive and may result in poor outcomes for the patient and his or her family. Oncology nurses need to be aware of potential sources of error and advocate for comprehensive genetic care.

AT A GLANCE

- Accurate assessment of risk for developing malignancy and appropriate genetic testing can lead to improved outcomes for individuals diagnosed with cancer and their family members.
- Credentialed genetics professionals have extensive training and experience in risk assessment, genetic test selection and interpretation, and coordination of care for the entire family.
- Errors in genetic testing may be associated with significant financial costs and can be attributed to inadequate knowledge about the genetic testing process, as well as lack of time to complete a comprehensive assessment and coordinate care among all family members.

KEYWORDS
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Errors in Genetic Testing

Common causes and strategies for prevention

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Genetic tests are continually increasing in scope and complexity, and genetic testing in oncology care is no longer as simple as ordering testing for one or two genes associated with a specific cancer syndrome. Now, genetics professionals acknowledge many diagnostic and technical laboratory considerations when selecting and interpreting genetic testing for hereditary cancer syndromes. Errors in genetic test ordering and interpretation are not only financially costly but also can lead to poor patient care outcomes.

The National Comprehensive Cancer Network ([NCCN], 2017) recommends that genetic testing be ordered by a credentialed genetics professional, such as a medical geneticist, a board-certified genetics counselor, or credentialed advanced practice genetics nurse (advanced genetics nurse–board certified, or AGN-BC). The American College of Surgeons' Commission on Cancer (2015) has established that facilities should provide cancer risk assessment, genetic counseling, and testing services to patients either on-site or by referral to a qualified genetics professional. Some insurance companies now require patients to be evaluated by a credentialed genetics professional for payment to be considered (Whitworth et al., 2017). Many of these recommendations have been implemented with the goal of decreasing genetic testing errors. This article will review potential causes of genetic testing errors and provide clinical examples and case analysis.

Inaccurate Assessment of Risk

Without accurate assessment of risk, making appropriate recommendations for cancer prevention, early detection, and genetic testing is impossible. Risk assessment includes the construction of a three-generation pedigree that comprises parents, siblings, aunts, uncles, and grandparents on the maternal and paternal sides. Prior to a counseling session, a family should gather the needed information to construct a pedigree, including the current age or age at death for family members, as well as the cause of death. For those family members diagnosed with cancer, having information about the site of the cancer, pathology characteristics (when available), and the presence of second primary cancers is important. Pedigree construction takes about 15–20 minutes and must be completed prior to making risk calculations. The latter is often difficult to accomplish in a busy practice setting.

Case Study 1: Incomplete Pedigree

A 42-year-old woman was referred for assessment. Her mother had been diagnosed with breast cancer at age 58 years, and her maternal grandmother had been diagnosed with breast cancer at age 49 years. The woman's primary care provider (PCP) said the Tyrer–Cuzick model, which predicts the lifetime risk of developing breast cancer based on personal risk factors and family history, put her risk for developing breast cancer at 59% and recommended bilateral risk-reducing mastectomies based on this calculation. However, the genetics professional