Errors in Genetic Testing

Common causes and strategies for prevention

Suzanne M. Mahon, DNSc, RN, AOCN®; AGN-BC

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 testing may be associated with significant financial costs and can be attributed to inadequate knowledge about the testing process, as well as lack of time to complete a comprehensive assessment and coordinate care among all family members.

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...