Managing Families With a Hereditary Cancer Syndrome

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Oncology is one of the first subspecialties to experience the full impact of the genomics revolution; oncology nurses regularly use genomic science in prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness in cancer care (Mahon, 2009). Genetic tests are now routinely ordered to determine risk for developing and appropriate management of hereditary breast and ovarian cancer, hereditary nonpolyposis colorectal cancer, and many other hereditary cancer syndromes—often without formal genetic assessment by a credentialed professional. Two cases will be reviewed that demonstrate the complexities of providing and coordinating care for at-risk relatives with a genetic predisposition to developing cancer.

Case Study 1

The first case illustrates the potential positive aspects that can occur when a family with suspected hereditary predisposition is managed by a credentialed genetics professional. A 39-year-old woman was diagnosed with colorectal cancer after an episode of rectal bleeding. The colorectal surgeon appropriately referred her for genetic counseling based on her young age of onset. A pedigree was constructed. The proband’s (i.e., patient’s) mother died from pancreatic cancer and one great paternal aunt was diagnosed with endometrial cancer after an episode of rectal bleeding. The colorectal surgeon contacted about the possibility of adding testing is now being coordinated for their offspring as they become old enough to undergo testing. The genetics professional provided counseling and testing for the other four siblings, and three tested positive. They have since undergone appropriate prophylactic measures and testing is now being coordinated for their offspring as they become old enough to consider testing.

Commentary on the Cases

Case 1 clearly illustrates the importance and complexity of genetic testing. First, genetic testing is continually evolving. A family who has previously tested negative for common mutations should be offered testing as new mutations are identified; this is a regular component of the practice of genetics