UNDERSTANDING

Genomic and Hereditary Cancer Risk

A HANDBOOK FOR ONCOLOGY NURSES

By
Suzanne M. Mahon, DNS, RN, AOCN®, AGN-BC, FAAN

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Preface

Collecting a family medical history is a regular component of the nursing assessment process. Patients with cancer often report that multiple family members have been diagnosed with cancer or express concern that other family members might be at increased risk for developing cancer.

As the involvement of genetics and genomics in oncology continues to grow, so too does public awareness of hereditary risk for developing cancer. This increasing awareness directly affects oncology nursing care. Although true germline (inherited) risk for developing cancer is much less common than somatic (acquired) risk for developing cancer, approximately 10% of cancer diagnoses can be attributed to germline risk. The identification of these families enables those at increased risk to engage in increased surveillance and, in some cases, risk-reducing surgery and other preventive measures, which ultimately lead to decreasing the morbidity and mortality associated with a cancer diagnosis.

In the past two decades, genetic testing for germline risk has greatly advanced. Genetic testing in oncology practice is being utilized more frequently and is best managed by genetics professionals. Genetics professionals, however, rely on healthcare providers to identify and refer patients for evaluation. Each time nurses extract a family history, they have an opportunity to assess for possible genetic risk and, when appropriate, refer patients and families to a genetics professional for further evaluation.

This book provides background on basic genetic and genomic concepts, especially related to germline risk for developing cancer. Nurses need this information to know when and why to refer patients. Nurses also need to give patients reliable information about how and why genetic and genomic testing can not only guide treatment decisions but also guide recommendations for cancer prevention and early detection. Chapter 2 describes foundational information about genomics and germline testing. Important terms appear in bold throughout the book and are defined in the Glossary.

Nurses may face questions from patients about what occurs during a consultation with a genetics professional. Patients often want to know why the test

cannot just be ordered. Genetic counseling involves multiple steps, including construction and interpretation of family pedigrees, ordering of tests, and interpretation of test results, all of which may have specific challenges. Nurses must be informed on the process so that they can reassure their patients that these steps are necessary. Chapter 3 provides an overview of the genetic counseling and testing process.

The increased awareness and direct-to-consumer marketing messages about genetic testing have prompted patients to ask more questions about genetic testing. Nurses must be able to respond to basic questions about different types of genomic testing with accurate information. If patients are concerned that healthcare professionals are not paying attention to their family history, they may turn to direct-to-consumer genetic testing, which may or may not provide the information they are seeking. Direct-to-consumer genetic testing is discussed in Chapter 4.

Following genetic testing, most genetics professionals provide detailed information about recommendations for care, including recommendations for ongoing prevention and early detection. For patients who are found to have a harmful pathogenic variant (historically known as a *mutation*), these recommendations can be extensive and are based on the personal and family medical history, as well as the specific pathogenic variant. Chapter 5 provides quick overviews of many of the more common germline pathogenic variants. These overviews include information on risks associated with each variant, recommendations for cancer prevention and early detection, and when to offer genetic testing. Each overview also includes reputable resources for more information.

With public awareness of genetic testing growing, nurses need a resource that provides them with more intensive information on germline cancer risk than they would typically receive during undergraduate or graduate education. This handbook provides nurses with the essentials to understand genomic and hereditary cancer risk, to assist in facilitating interprofessional care with genetics and other oncology professionals, and to provide their patients with accurate and reassuring information.