Cancer Genomics: Advocating for Competent Care for Families

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Modest gains have been made in the prevention and early detection of cancer in the past decade. Pharmacologic agents, lifestyle modifications, and diet and physical activity changes have been linked to preventing cancer. Breast cancer detection now includes digital mammography, computer-assisted diagnosis, and, most recently, breast magnetic resonance imaging. Research of cancer prevention and early detection will no doubt continue to result in improved strategies and outcomes. The premise of cancer prevention and early detection is that cancer will either be prevented or detected when cancer therapy is most likely to be effective, cost efficient, and associated with decreased morbidity and mortality. Clearly, nurses need to be engaged in cancer prevention and early detection and explore creative ways to implement both into the daily practice of all individuals, regardless of cancer diagnosis.

A decade ago, cancer prevention practice largely centered on engaging patients in cancer prevention and early detection practices. More recently, the explosion of genetics awareness rapidly is changing oncology practice. Selecting an appropriate prevention or detection strategy requires an accurate risk assessment. Genomics has added an entirely new dimension to cancer risk assessment, and nurses need to be prepared to competently integrate the knowledge into daily practice.

Genomics in Oncology

Genomics is receiving more focus as evidence shows that most diseases and conditions have a disruption in the genetic component. Genomics differs from genetics in that genetics scrutinizes the functioning and composition of the single gene, whereas genomics addresses all genes and their interrelationships to identify their combined influence on the growth and development of the organism (World Health Organization, 2002). Nurses must understand genomics and the role it plays in oncology. Oncology is one of the first subspecialties to experience the full impact of the genomics revolution in prevention, screening, diagnosis, prognosis, selection of treatment, and monitoring of treatment effectiveness. Oncology nurses will be faced with many ethical and professional challenges because of this knowledge (Lea, 2008).

The public increasingly expects that nurses will use and understand genomic information and technology when providing care (Consensus Panel on Genetic and Genomic Nursing Competencies [CPGGNC], 2006). The expectations have direct implications for students currently pursuing an entry-level degree in nursing as well as the 2.9 million practicing nurses (CPGGNC). CPGGNC has developed proficiency levels that state the minimal amount of genetic and genomic competency expected of every nurse. They include the following.

- Elicit, verify, and construct a minimum of three-generation family health history information using standard nomenclature.
- Consider genomic and environmental influences on risk-factor disease development.
- Identify patients who may benefit from specific genetic and genomic information or services based on assessment data.
- Identify credible, accurate, appropriate, and current genomic information, resources, services, or technologies specific to patients’ needs and refer patients as appropriate.
- Provide patients with interpretation of selective genetic information or services.
- Collaborate with insurance providers or payers to facilitate reimbursement for genetic and genomic healthcare services.
- Perform interventions appropriate to patients’ genetic healthcare needs.
- Evaluate the impact and effectiveness of genetic technology, information, interventions, and treatments on patients’ outcomes (CPGGNC).

Although most would not dispute that those proficiencies are reasonable and appropriate, the professional challenge surfaces in their application. This article uses the implementation of genetic predisposition testing for breast cancer as an example of the potential problems that can arise in clinical practice.

The potential for genetic predispositions for breast cancer has received a tremendous amount of public attention in the lay press and media since the late 1990s. The media coverage and increased knowledge has led to more women requesting breast cancer screening. Many women
and healthcare providers mistakenly believe genetic testing to be a simple blood test; however, it also requires educating women about their risk, if appropriate; selecting the proper test; and ensuring women have adequate pretest counseling and anticipatory guidance regarding how to manage the potential outcomes of testing. Messages from the media, including direct-to-consumer advertising and pressure from testing companies to clinicians, suggest that tests are easy to order and interpret. As a result, genetic testing has moved away from trained genetics professionals (Masny, Ropka, Peterson, Fetzer, & Daly, 2008). Unfortunately, many aspects of genetic testing will be overlooked with that approach and may result in poor outcomes for patients and families (CPGGNC, 2006).

Many oncology professionals and cancer centers offer genetic counseling and testing. Too often, centers offer the testing without a qualified professional to provide counseling. Several organizations advocate that genetic testing should only be offered when a qualified professional is available to provide genetic counseling and education and have published guidelines on who should provide genetic cancer-risk assessment services to high-risk patients (American Society of Clinical Oncology, 2003; CPGGNC, 2006; National Comprehensive Cancer Network, 2009; Oncology Nursing Society, 2006a, 2006b, 2006c; Trepanier et al., 2004). Qualified professionals who provide cancer genetics counseling and services typically include master’s-prepared genetic counselors, physicians who have completed post-residency training in genetics, and advanced practice oncology nurses with specialized education and training in genetics. Nurses must realize that specialized training in genetics is necessary to provide competent genetic care. Such training includes formal coursework and clinical supervision. Although laudable, a one-day continuing education program on genetic testing procedures is not adequate for this role.

### Clinical Examples

The author has seen an increasing number of patients in clinical practice who seek an opinion from a genetics professional after having tested. The problems demonstrated in each case most likely would have been avoided and minimized with referral to a genetics professional prior to testing. For those patients, appropriate care has now been rendered. Of greater concern are the patients who have undergone inappropriate or incomplete testing. Those patients may not have realized the limitations of their testing.

One competency identified by CPGGNC (2006) relates to nurses constructing a minimum three-generation pedigree with standardized symbols and approach. Although many view the competency as an unnecessary step, pedigree not only suggests who might be at risk for developing a hereditary cancer but the potential hereditary cancer syndrome. Automatically ordering BRCA testing on every woman with breast cancer diagnosed at age 50 or younger (a commonly used strategy by nongenetics professionals) will lead to the inappropriate use of healthcare dollars and perhaps the selection of the inappropriate test. The pedigree may suggest a more appropriate person to test in the family or another syndrome that should be considered.

To make a decision about what genetics test to order requires knowledge of cancer genetics and hereditary cancer syndromes. Case study 1 is a pedigree of a family who presented for consultation after a patient with cancer tested negative for a BRCA mutation. Evaluation of the pedigree suggested a potential hereditary breast cancer syndrome, a hereditary nonpolyposis colorectal syndrome, sporadic transmission, or other unusual syndrome. Without the accurate pedigree assessment, a diagnosis potentially would have been missed.

Primary care providers and oncologists increasingly order BRCA testing. Recently, a woman with cancer presented in the author’s genetic counseling practice with the pedigree in case study 2. Although BRCA testing was an appropriate strategy, it was incomplete. Fortunately, the woman was still concerned about hereditary risk and sought further counseling. Although dismissed by the oncologist as having no hereditary risk, additional testing revealed a rather unusual PTEN mutation that changed the risk profile for the woman and her relatives. Once again,

![Pedigree Diagram](image)

**Case Study 1**

The patient is a 35-year-old woman who received genetic testing in the community at the time of diagnosis. She was offered BRCA testing because of young age, which was appropriate. However, she did not receive specific counseling about the implications of the testing for herself and family. She tested negative. The oncologist did not recommend further evaluation. The family was still concerned about hereditary risk and saw a genetics professional. Further evaluation of the family history, verification with pathology reports, and testing an aunt with early onset endometrial cancer resulted in the detection of an MLH1 mutation, which is associated with Lynch syndrome. The patient was tested for the MLH1 mutation and also found to be positive. The mutation is associated with significant risks for colorectal, endometrial, and ovarian cancer. A genetics professional might have been more likely to consider all of the cancer diagnoses in the family, not just breast cancer.
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Case Study 2

The patient is a 48-year-old woman with cancer who tested negative for BRCA mutation. She was still concerned about cancer risk for her children and presented for genetic counseling. Further evaluation of the family pedigree showed members with thyroid cancer, unusual skin lesions and basal cell cancer, endometrial cancer and endometrial fibroids, and multiple cases of thyroid cancer. Additional testing demonstrated an unusual PTEN mutation. The patient was not only at risk for breast cancer but thyroid cancer, endometrial cancer and issues, kidney cancer, and unusual dermatologic lesions. Follow-up and management of affected family members should be coordinated and include multiple subspecialists. Had the pedigree not been adequately evaluated, the risks to the family would have been greatly underestimated.

BCC—basal cell carcinoma; Br—breast cancer; End—endometrial cancer; Kid—kidney cancer; Thy—thyroid cancer

Note. Arrow points to patient.

Note. Numbers following diagnosis indicate age at diagnosis; all other numbers indicate age at pedigree.

referral to a genetics professional often can prevent a potential omission.

Clinicians have commented to the author that they screen with BRCA and refer all positives for further evaluation. That strategy also can lead to potential problems. The approach will potentially overlook unusual hereditary syndromes in individuals who test negative. Pretest counseling will help clarify the possibility from the start. Women who are unprepared for the potential recommended strategies made after a positive test may experience significant psychosocial distress; be unwilling to consider preventive measures, including prophylactic surgery; or have difficulties with family relationships without appropriate anticipatory guidance (Hopwood, 2005). When genetic testing is ordered as a screening tool, women do not have the opportunity to make a truly informed decision about whether genetic testing will be beneficial to them. Many women with a significant family history who test negative still may have hereditary risk and should be counseled about the availability and possibility of enrolling in a research study.

Another common limitation when patients are not referred to a trained genetics professional, particularly in the case of a positive test result, is a lack of coordinated follow-up of all at-risk family members. If a clinician orders a test for a patient, the patient may be referred for prophylactic surgery or care may be altered. However, who is responsible for helping the patient understand which family members may be at risk based on pedigree assessment and coordinating counseling for possible testing for at-risk members is not clearly delineated. Testing for at-risk family members should be arranged for those who live in close geographic proximity as well as those who live in other geographic locations by identifying genetics professionals in those areas. This aspect of the continuum of care for genetics differs from oncology.

A genetics professional has access to resources in other geographic locations to help coordinate this care.

Implications for Nurses

Oncology nurses need to consider the potential negative or even legal pitfalls of providing genetics services without adequate background or training. Referring patients to a genetics professional may be the best option. All nurses should be able to identify patients at risk for a genetics disorder (hence the family pedigree), be able to identify and refer them to a genetics professional, and assist with carrying out the recommendations of a genetics professional. Advanced practice genetic nurses (APGN) have specific training in genetics and expanded skills in pedigree construction, genetic physical assessment, risk assessment, interpretation of complex medical and family histories, genetic tests, and genetic counseling.

The Genetics Nursing Credentialing Commission (2009) has established the following as part of the credentialing process for a nurse to receive the APGN credential. A credentialed nurse has demonstrated genetics expertise through formal education, continuing education, and clinical practice.

- Proof of RN license in good standing
- Minimum of a master’s degree in nursing or equivalent with graduation from an accredited graduate program in nursing
- 300 hours of genetic practicum experience as a clinical genetic nurse with a more than 50% genetic practice component
- Completion of a log of 50 cases reflecting genetic nursing care within five years of the application
- Four written case studies reflecting International Society of Nurses in Genetics (2007) standards of clinical genetics nursing practice
- 50 hours of genetic content in the past five years through academic courses or continuing education

Credentialing is based on the submission of a professional portfolio. A well-constructed portfolio includes examples of publications, presentations, and public education pieces or publications in addition to the information required (Monson, 2005). Unfortunately, training courses and educational opportunities
are limited in the United States. Some opportunities last only a day or a few weeks and do not include direct supervised clinical training.

The question of why a nurse should be an advanced practice oncology nurse and an APGN is asked often. Advanced oncology certification either as a clinical nurse specialist or nurse practitioner reflects a great breadth of knowledge of oncology nursing. For both examinations, 4% of the test addresses issues in cancer prevention and early detection, which may include genetics (Oncology Nursing Certification Corporation, 2008/2009), but genetics is not a major component of either test. Credential renewal does not include genetics content. Renewal of the APNG credential requires at least 50 hours of genetics continuing education during the past five years. One certification is not better or more prestigious than the other; they address different knowledge areas. Cancer genetics may be an area in which subspecialty certification may be indicated to enhance and ensure appropriate care for those receiving cancer genetics care.

Conclusions

All oncology nurses should have a basic level of genetics knowledge and skills. The number of health professionals with experience and training in genetics is limited and insufficient to serve all of the patients who would benefit from the services (Masny et al., 2008). The solution may not be to offer services without the training. Patients and families could suffer the consequences of that strategy as the case studies presented in this article suggest. Oncology nurses should be challenged to obtain additional training and credentialing in genetics to better serve at-risk patients and families. Considerations for administrative nurses and those in education include, “How will an adequate number of nurses be prepared?” “How will nurses be prepared to conduct a family risk assessment?” “How can the public be ensured that the nurse is adequately prepared to offer the counseling?” and “How do nurses ensure adequate and comprehensive genetic care for patients in environments that may have limited genetics resources?”

Nurses serve as advocates for their patients. Advocating for competent and comprehensive genetics care is an important role and challenge for nurses. Nurses need to be aware that genetics is rapidly changing cancer prevention and early detection, consider the complexities of genetic risk assessment, and appropriately refer patients and families for care.

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References


Silver Spring, MD: American Nurses Association.


