Scope and Standards

Defining the advanced practice role in genetics

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Oncology nurses at all levels of practice and in all settings are in an ideal position to address issues related to genetics and genomics. This is the second of two articles (Kerber & Ledbetter, 2017) describing the implementation of the updated Genetics/Genomics Nursing: Scope and Standards of Practice in daily use (American Nurses Association [ANA] & International Society of Nurses in Genetics [ISONG], 2016). This article focuses on implications for the advanced practice nurse (APN).

The growing complexity of genetic nursing in specialties such as oncology has led to the delineation of roles and levels of practice in genetic care. Genetics/Genomics Nursing: Scope and Standards of Practice is available to help guide practitioners at all levels of practice and includes 16 standards, complete with specific details of practice, to be used as a framework for evaluating practice outcomes and goals (ANA & ISONG, 2016). The publication also features an expanded discussion of ethics specifically related to genetics and genomics based on the ANA’s (2015) Code of Ethics for Nurses with Interpretive Statements. Figure 1 provides a comparison of roles in genetics based on these standards (ANA & ISONG, 2016), whereas Table 1 lists examples in practice for the APN.

Advanced Practice Roles in Genetics: Expanding Options

APNs function in a wide range of settings. First, the APN must consider state and federal regulations, insurance regulations, and accreditation requirements. Many oncology APNs have years of experience in settings with assorted and evolving responsibilities. Significant opportunities exist to collaborate with other genetics professionals who may not have as much oncology experience. Working together will provide better quality care and access for those at risk for or affected by cancer. Nurses with APN in genetics (APNG) certification, offered through the Genetic Nursing Certification Corporation, or who are advanced genetics nurse-board certified (AGN-BC) through the American Nurses Credentialing Center may have a broader scope of practice than certified genetic counselors (CGCs®) and baccalaureate-level genetics-certified nurses (GCGNs). (The APNG credential is being retired and transitioned to the AGN-BC.) However, not all APNs have independent practice authority because of differences in state board of nursing statutes. Also, the scope of practice may vary among APNs (e.g., nurse practitioner, clinical nurse specialist [CNS]) within the same state. The Genetics/Genomics Nursing: Scope and Standards of Practice guidelines are written to accommodate independent APN practice, but individual APNs must be cognizant of and adhere to the scope of practice laws in their state.

The introduction of multigene (panel) tests and exome sequencing have greatly affected practice for providers who order and interpret genetic tests in oncology. Panel testing can provide efficiency, particularly when a patient presents with risk factors and family histories that overlap with more than one syndrome. The disadvantages of panel testing include the possibility of detecting a gene mutation that is inconsistent with the family history.
For example, CDH1 mutations are associated with a greater than a 50% risk of gastric cancer, but have been detected in people with no personal or family history of gastric cancer through panel testing (Petrovich & Ford, 2016). Another disadvantage is that the rate of genetic variants of uncertain significance (VUSs) increases with the number of genes being evaluated (Kurian et al., 2014). APNs with genetic expertise are increasingly being called to assist with patient education, support, and interpretation of results.

**Case Studies**

**Case Study I: Private Practice**

An APN has been functioning independently with more than 25 years of oncology experience in a state that supports the expanded APN role. She runs a private genetics counseling practice and closely collaborates with other genetics and oncology healthcare professionals.

"Advanced practice nurses with genetic expertise are being called to assist with patient education, support, and interpretation of results."

A 38-year-old White, non-Jewish woman was diagnosed with triple positive (estrogen receptor–positive, progesterone receptor–positive, HER2/neu-positive) breast cancer at age 28 years. Now, she presents with osteosarcoma in her thigh. Her family history is negative for cancers of the breast, ovary, and colon, as well as for sarcomas. Her mother died from a brain tumor at age 34 years. Overall, the patient has many unaffected relatives, including her two siblings and her mother’s three siblings. Her father has no siblings, but he and his parents are unaffected.

The patient’s personal and family histories include risk factors for BRCA1 and BRCA2 gene mutations (early onset breast cancer) and for TP53 mutation (early onset breast cancer, sarcoma, and brain tumor). The patient’s breast cancer phenotype is more common in TP53 mutation carriers but is also seen in BRCA1 and BRCA2 mutation carriers. In addition, BRCA1 and BRCA2 mutations are much more common than TP53 mutations in the general population. The APN ordered a panel test that evaluates 30 cancer-associated genes.

The patient’s panel test revealed that she is heterozygous for a pathogenic mutation in her MUTYH gene. Testing also revealed a VUS in her BRCA1 gene. Although the report features the MUTYH mutation and labels it as pathogenic, the APN knows that the hereditary cancer syndrome associated with MUTYH gene mutations is a recessive disorder that causes polyposis. An estimated 1%-2% of the northern European population are MUTYH carriers (Nielsen, Lynch, Infante, & Brand, 2015), so MUTYH heterozygosity is a relatively common finding with panel testing. The MUTYH mutation does not explain the

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**FIGURE 1.**
**COMPARISON OF ROLES OF NURSES IN G/G**

**ROLE OF THE G/G NURSE**
- Identify patients with increased genetic risk; evaluate family history.
- Provide basic genetic information to patients and families.
- Refer patients to credentialed genetics professionals for counseling.
- Develop collegial relationships with other genetics professionals (e.g., certified genetic counselors, physicians, geneticists, specialty counselors).
- Personalize surveillance and management with genetic information based on guidelines.
- Recognize psychosocial aspects of genetic testing and diagnosis, including the ripple effect on family and friends.
- Protect patient genetic information through Genetic Information Nondiscrimination Act and Health Insurance Portability and Accountability Act standards.
- Expand knowledge through continuing education in G/G.

**ROLE OF THE GRADUATE LEVEL—PREPARED G/G NURSE OR THE APN**
- Initiate diagnostic tests and procedures relevant to patient condition.
- Assess the influence of G/G risk and disease on family communication and functioning.
- Provide personalized G/G counseling.
- Provide anticipatory guidance to individuals, families, groups, and communities to promote health and prevent or reduce the risk of health problems.
- Prescribe evidence-based treatments, therapies, and procedures considering the patient’s comprehensive healthcare needs.
- Model expert practice of G/G health care to interprofessional team members and patients.
- Communicate consultation recommendations to patients.
- Adapt the plan of care for the trajectory of treatment according to evaluation of response.
- Keep current regarding the practice landscape for APNs on state and federal levels, and adjust practice to meet patient needs.
- Obtain and maintain professional certification as an APN in genetics.

1 Suggested guidelines include those by the American Congress of Obstetricians and Gynecologists, American Society of Clinical Oncology, College of American Pathologists, National Comprehensive Cancer Network, and U.S. Preventive Services Task Force. APN—advanced practice nurse; G/G—genetics and genomics.

**Note.** Based on information from American Nurses Association & International Society of Nurses in Genetics, 2016; Calzone et al., 2010.
### TABLE 1.
STANDARDS IN G/G FOR ADVANCED NURSING PRACTICE

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<th>STANDARD</th>
<th>EXPLANATION</th>
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<tr>
<td>1. Assessment</td>
<td>Uses family history and pedigree for detailed assessment, including impact of G/G risk and disease on medications, interactions among individuals, and family communication and functioning</td>
<td>Initiates and interprets tests and procedures relevant to the patient’s current status</td>
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<td>2. Diagnosis</td>
<td>Systematically compares and contrasts clinical findings with variations and developmental events in formulating a differential diagnosis</td>
<td>Assists staff in developing and maintaining competence in the diagnostic or problem identification process; analyzes pedigree to identify inherited predisposition to disease</td>
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<td>3. Outcomes identification</td>
<td>Identifies expected outcomes that incorporate scientific evidence, cost effectiveness, patient satisfaction, and continuity and consistency among providers</td>
<td>Differentiates outcomes that require process interventions from those that require system-level interventions (within varied practice settings); assists in finding resources for service coverage through insurance and other sources of funding</td>
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<td>4. Planning</td>
<td>Identifies assessment and diagnostic strategies, along with therapeutic interventions, that reflect current evidence, including data, research, literature, and expert clinical knowledge</td>
<td>Leads the design and development of multidisciplinary processes to address an identified diagnosis or issue meeting the multifaceted needs of complex patients</td>
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<td>5. Implementation</td>
<td>Initiates collaboration with appropriate colleagues to implement the plan</td>
<td>Uses advanced communication skills to promote relationships between nurses and patients, provide a context for open discussion of experiences, and improve patient outcomes</td>
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<td>5A. Coordination of care</td>
<td>Provides leadership, synthesizes data related to community support, and initiates appropriate referrals to facilitate continuity of care</td>
<td>Collaborates with team to facilitate integrated care delivery</td>
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<td>5B. Health teaching and promotion</td>
<td>Synthesizes empirical evidence on risk behaviors, epidemiology, behavioral change and motivational theories; and other related theories and frameworks when designing health education information and programs</td>
<td>Provides personalized G/G education and counseling; designs and evaluates educational materials for accuracy, readability, and comprehensibility</td>
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<td>5C. Consultation</td>
<td>Provides consultation to influence the identified plan, enhance the abilities of others, and effect change</td>
<td>Synthesizes clinical data, theoretical frameworks, and evidence when providing consultation; includes stakeholders in decision making and negotiating role responsibilities; communicates recommendations to patient and members of the healthcare team; provides guidance for communication with relatives; documents that communications have occurred</td>
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<td>5D. Prescriptive authority and treatment</td>
<td>Uses prescriptive authority, procedures, referrals, treatments, and therapies in accordance with state and federal laws and regulations</td>
<td>Prescribes evidence-based therapies and pharmacologic agents within state and federal regulations; provides information about costs and alternative treatments and procedures; evaluates and incorporates complementary and alternative therapy into education and practice</td>
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<td>6. Evaluation</td>
<td>Conducts systematic, ongoing, and criterion-based evaluation of the outcomes in relation to the structures and processes prescribed by the plan of care and indicated timeline</td>
<td>Evaluates the effectiveness of planned strategies; disseminates the results of evaluation to the patient, family, and others involved; documents the results of the evaluation and necessary revisions</td>
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<td>7. Ethics</td>
<td>Delivers care in a manner that preserves and protects patient autonomy, dignity, rights, values, and beliefs; participates in interprofessional teams that address ethical risks, benefits, and outcomes</td>
<td>Provides information on the risks, benefits, and outcomes of healthcare regimens, such as G/G testing, to allow informed decision making by the patient, including informed consent and refusal</td>
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<td>8. Education</td>
<td>Attains core competencies in G/G for nurses recommended in the professional guidelines (Essential Genetic and Genomic Competencies for Nurses With Graduate Degrees)</td>
<td>Keeps up to date regarding the evolving practice landscape for APNs on state and federal levels, and adjusts practice to meet patient needs</td>
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<td>9. EBP and research</td>
<td>Seeks feedback from peer evaluations of performance and patient satisfaction; promotes a climate of research and clinical inquiry</td>
<td>Disseminates G/G research findings through activities such as presentations, publications, consultation, and journal clubs</td>
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<td>10. Quality of practice</td>
<td>Designs innovations to effect change in practice; evaluates the practice environment and uses the results of quality improvement to initiate changes in nursing practice and the healthcare delivery system</td>
<td>Obtains and maintains professional certification as an APN in genetics from the American Nurses Credentialing Center</td>
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patient’s clinical or family history of cancer. The APN also knows that although BRCA1 gene mutations are associated with early onset breast cancer, most VUSs are benign (Scherr, Lindor, Malo, Couch, & Vadaparampil, 2015). Although the genetic test detected two gene abnormalities, it is clinically uninformative.

With genetic testing, as with all testing, the ordering provider is responsible for interpreting test results. Test interpretation is an important component of genetic testing and is specifically discussed in the standards and competencies. Usually test interpretation is a component of standard 5 (implementation), as well as additional standards. However, cases exist in which a patient presents with genetic test results, either his or her own or those of a relative, which must be interpreted in the assessment (standard 1) and diagnosis (standard 2). The APN must know the limitations of the test that was performed and as many details as possible about the patient and his or her family history. Both hereditary and sporadic cancers can occur in the same family, so an understanding of which phenotypes are more or less likely to be hereditary helps to guide genetic test interpretation.

**Case Study II: Consultant**

An APN has been functioning as consultant to state and community health centers and has more than 35 years of oncology experience in a state that provides title protection to CNSs. In collaboration with a physician, she provides genetics services (e.g., risk assessment, testing, assistance with coordination of care) to community health centers and works closely with other primary care, genetics, and oncology healthcare professionals across the state in rural, suburban, and urban areas.

A 32-year-old African American woman with no personal history of cancer was referred for risk assessment and counseling after a mass was found during a breast examination. Her family history includes her mother and three maternal aunts diagnosed with and deceased from breast cancer by age 33 years. No other living relatives are on the maternal side, and additional information is not available. The patient did not know her father and has minimal paternal family history, with the exception of knowing that a paternal aunt died from colon cancer at age 45 years. The daughter of this aunt recently contacted the patient about her discovery of a pathogenic MSH2 mutation.

Per the algorithm in the clinic, the patient was referred to the breast surgeon for mammography and further evaluation. Concurrently, the APN completed a risk assessment. The patient’s family history included risk factors for BRCA1 and BRCA2 gene mutations (early onset breast cancer) and for TP53 mutation (early onset breast cancer), as well as a known mutation (MSH2) in the family. The patient had serious concerns about her breast cancer risk and was interested in testing. She had no

### TABLE 1. (CONTINUED)

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<td>11. Communication</td>
<td>Communicates effectively in a variety of formats in all areas of practice</td>
<td>Assesses communication formats for preferences with patients, families, and colleagues; examines and adjusts own communication skills in encounters with others; contributes professional perspective in discussions with interprofessional team</td>
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<td>12. Leadership</td>
<td>Influences decision-making bodies to improve the professional practice environment and patient outcomes and provide equitable G/G care</td>
<td>Mentors colleagues in the acquisition of clinical G/G knowledge, skills, abilities, and judgment; networks with other G/G professionals in the promotion of quality and equitable care</td>
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<td>13. Collaboration</td>
<td>Partners with other disciplines to enhance patient outcomes through interprofessional activities, such as education, consultation, management, technological development, and/or research opportunities</td>
<td>Documents plan-of-care communications, rationales for changes, and collaborative discussions to improve patient outcomes; presents information at interprofessional conferences</td>
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<td>14. Professional practice evaluation</td>
<td>Engages in a formal process seeking feedback regarding G/G nursing practice from patients, peers, professional colleagues, and others</td>
<td>Seeks input from patients, peers, professional colleagues, and others</td>
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<td>15. Resource utilization</td>
<td>Uses organizational and community resources to formulate interprofessional plans of care; considers the cost when evaluating the risks and benefits of G/G tests in the context of achieving desired patient-centered goals</td>
<td>Recommends G/G tests associated with improved patient outcomes</td>
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<td>16. Environmental health</td>
<td>Analyzes the impact of social, political, and economic influences on the environment and G/G susceptibilities associated with human health exposures</td>
<td>Supports nurses in advocating for and implementing principles in nursing practice that promote environmental safety; participates in community outreach efforts</td>
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APN—advanced practice nurse; EBP—evidence-based practice; G/G—genetics and genomics

**Note.** Based on information from American Nurses Association & International Society of Nurses in Genetics, 2016.
insurance, but met medical and financial criteria for financial assistance. In addition, the known mutation had been detected by the same laboratory. The discussion of potential impact of the results focused on heightened surveillance and follow-up. The APN ordered a panel test that evaluates 28 cancer-associated genes.

Mammography revealed a 4 cm mass in the left breast; this was biopsied and found to be benign. Genetic testing revealed a pathogenic BRCA2 mutation, which is informative and actionable. Bilateral mastectomy and salpingo-oophorectomy were recommended because of the patient’s family history and mutation status. The APN facilitated conversation among staff, the physician, and the patient. Because the patient hoped to have a relationship and more children in the future, she opted for heightened surveillance instead of risk reduction surgery. She is being followed by the same breast surgeon.

Standards applicable to this scenario include assessment (standard 1) to determine the most appropriate genetic test or panel for the individual and the situation, coordination of care (standard 5A) to optimize outcomes (standard 3) for the patient, health teaching to assist the patient in understanding her risk (standard 5B), and promotion of the patient’s autonomy and management in alignment with her values (standard 7).

**Conclusion**

The standards outlined in *Genetics/Genomics Nursing: Scope and Standards of Practice* are designed to support and guide genetic and genomic practice as an evolving, complex, and major component of oncology practice. Oncology nurses need to incorporate these standards into the context of their experience, education, and state practice, as well as act with the goal of providing comprehensive and effective nursing care. APNs have additional competencies and responsibilities that are aligned with level of practice. All oncology nurses have the responsibility of collaboration to maintain knowledge and competencies to ensure quality of care for patients in all types of practices and locations.

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**REFERENCES**


