Genomics Education

Knowledge of nurses across the profession and integration into practice

Lisa B. Aiello, RN, MSN, AOCNS®, APRN-C

BACKGROUND: Since the completion of the Human Genome Project in 2003, the implications of genetics and genomics for health and illness have become clearer. All nurses, at all levels and across all specialties, must have a basic understanding of genetics and genomics to provide appropriate care.

OBJECTIVES: This article provides an overview of the integration of genetics and genomics into nursing education, as well as continued barriers to nurses' knowledge of genomics and genomics.

METHODS: A literature review was conducted using CINAHL®, PubMed, ProQuest, and Google Scholar. Databases were reviewed for literature published from January 1962 to November 2017.

FINDINGS: Although genetic and genomic education has been integrated into undergraduate and graduate nursing curricula, a gap exists in this knowledge among student nurses, practicing nurses, and nursing faculty.

GENETICS AND GENOMICS HAVE BECOME CENTRAL TO HEALTH CARE in all areas, specialties, and settings, particularly oncology. In addition, genetics and genomics have implications across all areas of the healthcare continuum, as well as all aspects of patient care, including assessment, prevention, screening, diagnosis, prognosis, treatment, assessment of treatment effectiveness, and drug selection (Calzone et al., 2012; Calzone & Jenkins, 2012; Pestka, Burbank, & Junglen, 2010). This integration of genetics and genomics into healthcare requires that healthcare providers, including nurses, become educated and competent in genomic care.

Background

The Human Genome Project (HGP) expanded the understanding of how genomes work and caused the rapid growth of genomic technology (Daack-Hirsch et al., 2013). Started in 1990 and completed in 2003, the HGP was an international research initiative with the goal of sequencing the entire human genome (National Human Genome Research Institute [NHGRI], 2016). This project identified the 20,500 genes in each human and their chromosomal locations, as well as provided information about the complete set of human genes and suggested that the focus of genetics (the study of single-gene disorders) should include genomics (the study of how genes interact with environmental, psychosocial-behavioral, and cultural factors). In addition, it identified the human blueprint for health (NHGRI, 2016). Subsequent research has suggested that genetics and genomics provide the basis for health, illness, disease risk, and treatment response (Clark, Adamian, & Taylor, 2013). Genetics and genomics have already moved into mainstream health care, as evidenced by the development of precision medicine and the National Cancer Moonshot Initiative.

Nurses are the largest healthcare profession, with 2.8 million RNs active in 2014 and a projection of 3.8 million RNs active in 2030 (U.S. Department of Health and Human Services, Health Resources and Services Administration, National Center for Health Workforce Analysis, 2017). Nurses practice in all healthcare settings and continue to be rated as the most honest and ethical professionals (American Nurses Association, 2016). Therefore, nurses must be at the forefront of the integration of genetics and genomics into clinical practice; however, to this point, nurses have had limited education in genetics and genomics care.

KEYWORDS

Genetics; genomics; nursing; competency; education

DIGITAL OBJECT IDENTIFIER

10.1188/17.CJON.747-753
In the 1990s, the HGP and its anticipated results became the impetus for rejuvenated concern regarding nurses’ knowledge of genetics and genomics. Nurse leaders and stakeholders in genetics and genomics began to publish articles and studies about nurses’ knowledge of the fields. Initially, the literature addressed the need for education (Godino & Skirton, 2012; Lea et al., 2011), with research focused on identifying nurses’ current knowledge of genetics and genomics and its implications related to nursing practice. The research showed that nurses were deficient in genetic knowledge and literacy (Godino & Skirton, 2012; Lea, Kapfingst, Bowen, Lipkus, & Hadley, 2011), with research focused on identifying nurses’ current knowledge of genetics and genomics and its implications related to nursing practice. The research showed that nurses were deficient in genetic knowledge and literacy (Godino & Skirton, 2012; Lea et al., 2011). Nursing faculty also reported a lack of genetic and genomic knowledge, but responded that including this content in curricula was important (Jenkins & Calzone, 2012).

Curriculum and textbooks were evaluated, and the results of this evaluation supported a lack of inclusion of sufficient genetic content. The genetic content that did exist was grouped with information about maternal and child health, and information about ethics in relation to genetic care was lacking. Hetteberg, Prows, Deets, Monsen, and Kenner (1999) recommended inclusion of a genetics course in the baccalaureate curriculum, as well as content related to the topic of ethics in genetics.

Once the baseline assessment of genetics nursing education occurred, the focus of nursing research moved to the education of faculty and integration of content into nursing curricula. Many faculty were uncomfortable with the content, and integration of this content into already content-packed curricula was difficult (Hetteberg et al., 1999; Monsen, 1984). Time constraints and an overloaded curriculum are frequently identified as reasons for continued exclusion of genetic content (Hetteberg et al., 1999; Monsen, 1984). A landmark study supported by the American Nurses Association (Scanlon & Fibison, 1995) assessed the state of nursing practice in genetics by surveying 1,000 nurses. Most nurses surveyed reported not receiving any formal education on genetics, agreed that a need existed for genetic education, and noted a lack of competence and confidence in their genetic skills. However, they also stated that current research related to mapping and sequencing human genes was not relevant to their practice (Scanlon & Fibison, 1995).

In the early 2000s, the HGP was almost complete, and findings were slowly being released. The potential implications of genetics and genomics were only just beginning to be realized. The focus of nursing researchers continued to be the assessment of nurses’ genetic knowledge and the integration of genetic and genomic content into nursing curricula. More recently, research has begun to examine the application and use of genetic and genomic competencies in nursing practice, particularly the collection and use of family histories and pedigrees (Kelly, 2011; Pestka, Meiers, Shah, Junglen, & Delgado, 2012).

**Nursing Competencies**

The research into the genetic and genomic knowledge of nurses led to many initiatives, such as the development of the *Essential Nursing Competencies in Genetics and Genomics*.
Competencies and Curricula Guidelines for Genetics and Genomics in 2006, which was updated with outcome indicators in 2008 (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). These competencies are evidence-based and define the minimal competencies expected of all nurses, regardless of educational level or specialty. They are broken down into two domains: professional practice and professional responsibilities. See Figure 1 for an overview of the competencies for the two domains. The outcome indicators specifically define how an RN can demonstrate genetic and genomic competence (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). Overall, these competencies provided nursing with a framework for identifying the educational needs of nurses. The American Association of Colleges of Nursing ([AACN], 2008) used these competencies to support their recommendation for the inclusion of genetics and genomics into nursing curricula (Jenkins & Calzone, 2012). Prior to the AACN’s (2008) recommendations, many schools of nursing did not include genetics and genomics in their curricula and instead used the blueprint of the National Council Licensure Examination for RNs (NCLEX-RN) to guide content development. The NCLEX-RN detailed test plan for 2016 includes family and genetic history in the health promotion and maintenance category of the examination, but no other mention of genetics or genomics is present in the test plan (National Council of State Boards of Nursing, 2015).

Many nursing publications elaborate on how the essential competencies for genetic and genomic nursing may be applied and integrated into nursing practice within multiple specialties (e.g., oncology, midwifery, pediatric, neonatal, critical care, transplantation, palliative care, psychiatric, perinatal, family, head and neck, hemophilia, hemoglobinopathy, gastroenterology, pharmacogenomics, neuropsychiatric, emergency care) (Bancroft, 2013; Calzone et al., 2010; Chuang, Hsieh, & Addullah Charles, 2013; Clark et al., 2013; Engstrom, Sefton, Matheson, & Healy, 2005; Jenkins, 2002; Jenkins, Grady, & Collins, 2005; Pestka, Derscheid, et al., 2010; Visovsky, 1999; Warren, 2008; Workman, 1999). Many of these specialties, including emergency care, involve illnesses and treatments that have a genetic or hereditary component, such as long QT syndrome, cardiomyopathies, hemoglobinopathies, asthma, and individualized responses to medications. Genetic and genomic competencies have been developed for graduate-level nurses and build on those intended for all nurses (Greco, Tinley, & Selbert, 2012); these are listed in Figure 2. Nursing research supports the need for advanced practice nurses to be competent in genetic care (Beery & Hern, 2004; Coy, 2005; Kerber & Ledbetter, 2017; Maradiegue et al., 2005; Whitt, Macri, O’Brien, & White, 2016; Williams & Dale, 2016; Williams et al., 2016).

Nursing Curriculum and Faculty Knowledge

In the 2000s, many educational programs were developed to educate faculty on genetics and genomics, as well as to prepare them to educate students, and they included guidelines for creating genetic curricula either as a single course or integrated across a range of courses. Daack-Hirsch, Dieter, and Quinn Griffin (2011) and Jenkins, Prows, Dimond, Monsen, and Williams (2001) provided recommendations for the integration of genomics into nursing curricula, including the tools and resources that would facilitate this move. In addition, the Faculty Champion Initiative, launched in 2008, provided genomic education and supported interventions to select nursing faculty to assist them in their role as change agents at their respective institutions (Jenkins & Calzone, 2014). The goal of this initiative was that these faculty, who were key stakeholders in their respective schools of nursing, would aid in the integration of genetics and genomics into nursing curricula. However, many barriers were identified to explain why genetics was not being added to nursing curricula. For instance, faculty did not have the educational knowledge to teach genetics and did not see the importance of this education, and practical application of genetics into practice was not easily identified (Jenkins & Calzone, 2014).

Read and Ward (2016) evaluated 495 nursing faculty members’ understanding of genomic concepts using the Genomic Nursing Concept Inventory® (GNCI®) and compared the results to a sample of 705 nursing students. The higher the score, the better the participant’s understanding of foundational genomic concepts. The mean nursing faculty score was 14.93 correct of 31 questions (48%). Nursing faculty who had taught a standalone genetics course (N = 25) scored better, with 65% of questions answered correctly, than faculty who taught genetic content within a nursing course (N = 182), with 55% of questions answered correctly. The mean nursing faculty score (48% of questions answered correctly) was not much higher than students’ mean score (47% of questions answered correctly). The majority of faculty (n = 344) reported their proficiency with genomic content as being fair to poor. Just 33% (n = 163) of the faculty sample reported having...
attended continuing education programs on genetics and genomics prior to taking the GNCI.

Donnelly, Nersesian, Foronda, Jones, and Belcher (2017) surveyed 20 faculty members from one school of nursing and found that more than half reported a lack of confidence in teaching genetics and genomics. Significant knowledge gaps were identified; most nurses reported not receiving genetic and genomic education in their own nursing program and not attending any courses with genomic content since. However, 65% indicated that the inclusion of genetics and genomics in nursing curricula was important. In response to the Donnelly et al. (2017) study, the school of nursing developed a plan to address these issues that included faculty development, identification of faculty champions, and creation of a plan for threading genetic and genomic content throughout the curriculum.

**Genomic Knowledge and Integration Into Practice**

Nurses must be competent in obtaining comprehensive family histories, identifying family members at risk for developing genetic conditions, and understanding drug reactions influenced by genomics. In addition, they must be able to educate patients about genetic and genomic tests and therapies to facilitate informed patient decision making; nurses must also know the importance of genetic counseling and be able to refer at-risk patients to appropriate genetics professionals for specialized care (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). Researchers have studied the application and use of the genetic and genomic nursing competencies in nursing practice (Kelly, 2011; Pestka et al., 2012). Most of these studies focused on the creation and use of family histories and pedigrees, because collecting this health history and constructing a three-generation pedigree is a competency expected of all nurses, regardless of education or practice setting, and is the cheapest, most accessible way of drawing or updating a family pedigree. (Calzone et al., 2012; Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). Taking a family history and constructing a pedigree is considered to be the gold standard in identifying and stratifying risk for single-gene disorders, such as cystic fibrosis and hereditary breast and ovarian cancer syndrome; chromosomal disorders, such as Down syndrome and chronic myelogenous leukemia; and complex genomic chronic diseases, such as asthma, Crohn’s disease, depression, and diabetes mellitus (Kelly, 2011; Wattendorf & Hadley, 2005).

Multiple studies evaluating the interest, knowledge, and practice of nurses related to genetics and genomics have shown a lack of knowledge and integration of genetic and genomic competencies into practice (Camack, 2016; Kelly, 2011; Munroe & Loerzel, 2016; Pestka, Derscheid, et al., 2010; Pestka, Lim, & Png, 2010; Sharoff, 2017; Spruill, Coleman, & Collins-McNeil, 2009). Spruill et al. (2009) assessed the interest, knowledge, and practice of 77 African American nurses regarding genetics and genomics, finding that most (85%) of the sample reported previously completing a family health history with a patient; however, 61% reported that they had not previously referred a patient for genetic services.

Pestka, Lim, and Png (2010) described the influence of a genomic educational intervention on practicing nurses in Singapore (N = 76). The nurses participated in a seminar, and then completed a survey. Participants were asked to provide a case study from their own practice and identify which genetic assessments and interventions they used with the specified patient. The results were as follows: 90% assessed physical findings; 86% gathered family history information; 86% discussed environmental risk factors; 72% used family history to discuss health risks; 61% assessed the genetic knowledge of the patient’s family; 55% provided education based on these assessed needs; 54% discussed preventive measures; 40% had drawn or updated a pedigree; 28% discussed ethical, legal, cultural, or social issues related to genetics; and 9% indicated they had facilitated genetic referrals. Participants reported a higher usage of competencies that involved assessment and a lower usage of competencies that involved interventions. Patient diagnoses were primarily cancer and diabetes mellitus but also included high-risk pregnancy, hypertension, and asthma.

Pestka, Derscheid, et al. (2010) duplicated Pestka, Lim, and Png’s study (2010) of Singapore nurses, instead evaluating nurses in the United States working in inpatient psychiatric units. Nurses in this study had the opportunity to attend multiple genomic educational sessions during the previous seven years because of educational initiatives offered by the facilities. Those who participated in the study were invited to complete a survey that measured the application of content from an educational session to practice. The survey was developed by the investigators based on the genetic and genomic competencies that had already been accepted by multiple nursing organizations in the United States (Pestka, Derscheid, et al., 2010). Statistical analysis of the data revealed that, overall, attendance at a greater number of educational sessions had a positive correlation with the performance of a greater number of genomic assessments (e.g., assessing family history and patient/family knowledge) (r = 0.223, p = 0.076) and was highly correlated with the performance of a greater number of genomic interventions (e.g., providing education) (r = 0.488, p < 0.001). However, only 3% performed the assessment activity of drawing or updating a family pedigree.

Kelly (2011) evaluated the family history assessment and documentation practices in three separate hospital-based gastroenterology units that performed colonoscopies. Sixteen gastroenterology unit nurses were also interviewed. Only 22% of the 88 charts reviewed included documentation of a family history of colorectal cancer. As reported by the nurses, the most common barriers to completing a family history were no hospital guidelines or protocol (reported by four nurses) and other factors (reported by six nurses), such as family history being collected in another area, the need for additional help, and language barriers (Kelly, 2011). Kelly (2011) suggested possible benefits of the use
of electronic health records (EHRs); however, most EHRs have underdeveloped family history sections that do not allow for interpretive information (Feeero, Bigley, & Brinner, 2008).

The Genetic/Genomic Nursing Practice Survey was used in multiple studies to measure the attitudes, receptivity, social systems, confidence, competence/knowledge, and decision/adoptive of nurses as they relate to genetic and genomic nursing practice (Calzone et al., 2012; Calzone, Jenkins, Culp, et al., 2013; Calzone, Jenkins, Culp, Caskey, & Badzek, 2014; Coleman et al., 2014; Saligan & Rivera, 2014). These studies indicated an overall inadequate integration of the genetic and genomic competencies into nursing practice. However, many of these studies indicated that having a higher education level or having taken a genetics course since licensure improved the use of these competencies. This information supports the continued need to further educate nurses (students and RNs who are already practicing).

The GNCI has been used in many studies assessing nursing students’ understanding of foundational genomic concepts and common misconceptions (Munroe & Loerzel, 2016; Ward, French, Barbosa-Leiker, & Ivensen, 2016; Ward, Haberman, & Barbosa-Leiker, 2014; Ward, Purath, & Barbosa-Leiker, 2016). Many such studies found that those who tended to score higher on the GNCI (indicating more knowledge of foundational genomic concepts) were male students, students who had previously taken a genetics course, students in their final semester of nursing school, and students in accelerated programs.

Additional studies have evaluated nurses’ or nursing students’ actual knowledge of genetics and genomics (Bankhead et al., 2001; Bottorff et al., 2005; Godino & Skirton, 2012; Godino, Turchetti, & Skirton, 2013; Rogers, Lizer, Doughty, Hayden, & Klein, 2017). Godino et al. (2013) found a positive correlation between level of education and knowledge. Genetics and genomics have become essential core competencies for all nurses. Nurses need to be educated, become competent, and integrate these competencies into practice to provide safe and effective quality patient care, as well as improve patient outcomes. Many educational initiatives have been implemented to educate faculty, nursing students, and practicing nurses. See Figure 3 for resources related to genetics and genomics education and literacy.

**Conclusion**

Genetic and genomic competencies, curricular standards, and the scope of genetic and genomic practice have been developed across all levels of nursing. However, among undergraduate and graduate nursing students, knowledge of genetics and genomics remains low. Many studies have addressed genetic and genomic knowledge gaps, integration of competencies into practice, and educational needs of nursing students, practicing nurses, and nurse educators. The majority of studies identified that nurses with a higher level of education and nurses who had taken a genetics course since licensure had increased their knowledge and integration of genetic and genomic competencies into practice.

Research has indicated some progress in the acquisition of genetic and genomic knowledge by nurses, but additional research can inform best practices to establish clinical genetic competencies. Research can also help to establish strategies to improve genomic nursing curricula and to ensure expertise of faculty.

Lisa B. Aiello, RN, MSN, AOCNS®, APRN-C, is an assistant clinical professor in the College of Nursing and Health Professions at Drexel University in Philadelphia, PA. Aiello can be reached at lba34@drexel.edu, with copy to CJONEditor@ons.org. (Submitted July 2017. Accepted September 11, 2017.)

The author takes full responsibility for this content and did not receive honoraria or disclose any relevant financial relationships. The article has been reviewed by independent peer reviewers to ensure that it is objective and free from bias.
REFERENCES


