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Misconceptions about Genomics among Nursing Faculty and Students

Catherine Y. Read, PhD, RN, Linda D. Ward, PhD, RN, FNP-C

Authors' Affiliations: Associate Professor (Dr. Read), William F. Connell School of Nursing, Boston College, Chestnut Hill, MA, USA; Assistant Professor (Dr. Ward), College of Nursing Washington State University, Spokane, WA, USA.

Corresponding author: Catherine Y. Read, Boston College William F. Connell School of Nursing, 140 Commonwealth Avenue, Chestnut Hill, MA, 02467
catherine.read@bc.edu

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Abstract

A comparison of 2 research studies revealed nursing faculty and students share limited understanding and specific misconceptions about foundational genomic concepts. Mean scores on the Genomic Nursing Concept Inventory were 47% for faculty and 48% for students. Identifying misconceptions is important when designing educational strategies for students who will inevitably care for patients with genomic concerns. Common clinical scenarios requiring accurate interpretation of genomic terminology, gene function and expression, and genetic counseling principles are presented.

Key Words: genomics, nursing education, genetic literacy, genetics education

Introduction

Nurses, as a primary point of contact for patients, must be able to interpret genetic and genomic information and translate rapidly advancing science into clinical practice. Screening for genomic risk, testing for the diagnosis of diseases with a genomic basis, and using genotypes to determine the most efficacious pharmacologic therapies are becoming common practices in health care. Accurate understanding of genomic science is now an essential aspect of nursing practice. Unfortunately, recent studies indicate that nursing faculty and students have limited knowledge and hold many misconceptions about basic genomic concepts. The purpose of this article is to identify those misconceptions in the hope that educators in both practice and academia will plan programs that remediate and build knowledge about genomics.

Assessing Genomic Literacy

Genomics is a relatively new science that was not part of the initial preparation of many nurse faculty. Even the terms *genetics*, which refers to the study of a particular gene, and *genomics*, a newer term that refers to the study of the entire genome of an organism,¹ are used inconsistently. In this paper, we use the broader term genomics, except when discussing a single gene, referring to established phrases or practices that have retained the term genetics, or referencing an author whose work used the term genetics.

Lack of knowledge and confidence to teach about genomics among nursing faculty are well-documented phenomena. In 2005, researchers from several countries identified lack of faculty knowledge as a primary barrier to increasing the amount of genomics content in nursing curricula.² In another study,³ less than 20% of a sample of 341 faculty from 103 US nursing schools agreed or strongly agreed that faculty at their schools generally feel capable of teaching genetics. Only 11/47 (23%) of experienced nurse educators in a 2012 study reported feeling somewhat or completely confident in their ability to counsel or refer a patient with information from a direct-to-consumer genetic test,⁴ an activity that is listed as an essential competency of a baccalaureate-prepared nurse by the American Association of Colleges of Nursing (AACN).⁵ More recently, 70% of a sample of 495 experienced educators in US nursing schools rated their proficiency with genetic/genomic content as fair or poor.⁶ A similar lack of confidence in the ability to teach essential genomics concepts was found in a small sample of nursing faculty in 2016.⁷

Low faculty confidence in teaching genomics is paralleled by a documented lack of basic knowledge. One of the first reported studies of genetic knowledge among nursing faculty and students⁸ used the Genetics Literacy Assessment Instrument (GLAI),⁹ a 31-item multiple-choice test of genetic knowledge in 6 domains (nature of genetic material, transmission, gene

expression, gene regulation, evolution, and societal implications). The faculty in that study achieved a mean score of 76%, only slightly higher than the students, whose mean score was 73%. The authors concluded that the GLAI was a promising tool for evaluating curricula and for identifying faculty and students in need of additional education about genomics. The GLAI was also used to track acquisition and retention of genomic concepts among students.¹⁰ Although GLAI scores improved with completion of genomic coursework, knowledge retention was poor; the average total score for 1 cohort of students was 70% as sophomores, 67% as juniors, and 62% as seniors.

A limitation of the GLAI is that it was developed for use with students in college science courses and does not measure concepts specifically related to the role of the nurse. The GLAI was also relatively easy for nursing faculty and students, with pre-instruction scores above 70%.⁸ That limits the ability to measure learning gains. To address these shortcomings, a 15-item multiple-choice genetic/genomic knowledge test was developed.¹¹ That test was derived from competencies outlined in the AACN Essentials of Baccalaureate Education for Professional Nursing Practice⁵ and the Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators.¹² This genetic/ genomic knowledge test evaluated performance on questions about basic genetic/genomic definitions, inheritance patterns, referral actions, pedigree development, cultural issues, and insurance issues. The mean score on the instrument, administered to 117 nursing faculty, was 53%. The 26 faculty who reported having taken a genetics course since their basic biology training scored a mean of 61.5%, which was significantly higher ($p=0.005$) than the mean score of 51% for the 91 faculty who had not had such a course.

In order to develop a more robust measure of genomic literacy targeted to nursing students, faculty, and clinicians, a rigorous concept inventory development strategy was employed to create the Genomic Nursing Concept Inventory (GNCI[®]2011).¹³ In a study of 495 faculty,⁶ the mean GNCI score was 48% correct; this was strikingly similar to the mean score of 47% correct found in a study of 1002 baccalaureate nursing students.¹⁴ Those studies provided the data used to examine common misconceptions about genomics described in this paper.

Difficulties Inherent in the Study of Genomics

Understanding of foundational genomic concepts is limited by knowledge deficit, confusion, and lack of integration. Three sources of difficulty in learning genomics have been well-elucidated in biology education research and more recently studied in nursing students.¹⁵ First, the language of genomics is intrinsically complex. Some terms (e.g., allele) are unique to the domain, requiring students to learn a new vocabulary. Other terms (e.g., dominant) are used in general conversation with meanings that do not carry over to their constrained definition in genomics.¹⁶

Second, understanding how genes influence health requires students to integrate concepts across multiple levels of organization. Genomic mechanisms occur at a molecular or cellular level and are not directly observable, yet they affect visible traits. To connect genotype to phenotype, students must integrate concepts across molecular, microscopic and macroscopic levels, which is difficult for novices.¹⁷ Together, complex vocabulary and conceptual complexity are thought to contribute to low genomic literacy demonstrated in undergraduate students.

The third source of difficulty in learning genomics stems from commonly-held wrong ideas (*misconceptions*).¹⁷⁻¹⁹ College students tend to believe they understand genomics but

endorse incorrect but firmly-held beliefs they have constructed over years of formal education and personal experiences. A substantial body of research in biology education based on cognitive learning theory describes how misconceptions impede learning, when new knowledge does not “fit” a student’s preexisting (but incorrect) conceptual framework. Integrative learning is therefore difficult and students may resort to memorization. Misconceptions may not be apparent to students or educators and are resistant to reconciliation. A number of pervasive misconceptions have been identified in studies of secondary and undergraduate students.¹⁸ Examples are that most traits are determined by a single gene, that different tissues contain different genes, that only dominant genes are expressed, and that people with a genetic disease have a “disease” gene that others lack.¹⁶⁻¹⁹

Although nursing students are likely to hold similar misconceptions as other undergraduate students, little is known about genomic misconceptions among nursing students or faculty. Genomic nursing education will benefit from identifying shared wrong ideas. Teaching can then be targeted to illuminate and reconcile misconceptions, so that education is efficient and evidence-based and results in meaningful learning.²⁰

Methods

The purpose of this study was to identify common misconceptions about genomics shared by nursing students and faculty. To accomplish this, we conducted a secondary analysis of data from 2 published studies that had utilized the Genomic Nursing Concept Inventory (GNCI). The studies were overseen by institutional review boards at their respective institutions. The first study⁶ consisted of a sample of 495 predominantly female (96%), White (93%), geographically dispersed U.S. nursing school faculty with a mean age of 53 years and a mean of 12 years teaching in a nursing program. The comparison sample¹⁴ comprised 1002 U.S. nursing students

from both BSN completion and prelicensure BSN programs; students were predominantly female (90%) and non-Hispanic (95%), spoke English as a first language (93%), had a mean age of 24 (range 18-58), and had not taken a previous standalone genetics course (84%).

The GNCI is a 31-item test of foundational concepts underlying the established essential nursing genetic/genomic competencies.¹² Evidence from extensive testing and evaluation using advanced psychometric methods supports GNCI use to measure understanding of concepts underlying genetic-genomic competencies for nurses. The scale is unidimensional and shows satisfactory internal consistency reliability, with Cronbach alpha values between 0.73 and 0.87 across studies.^{13,14,21} Content validity has been endorsed in a Delphi study.²² The GNCI was developed using a rigorous strategy common in science, technology, engineering and mathematics (STEM) education. Concept inventories use theory and research in the design of instruments to measure understanding of key concepts. An important aspect of the GNCI is that item distractors (incorrect responses) reflect the most common misconceptions identified among nursing students. Thus, GNCI data reveal “wrong thinking” that can inform curriculum development and program planning. In this study, we tabulated the incorrect responses to gain insight into common misconceptions about genomics held by nursing faculty and students.

Results

Table 1 lists the most common incorrect answers selected by faculty and students on the GNCI. Students and faculty had similar response patterns and similar mean scores (47% and 48% correct, respectively). Most misconceptions reflected poor understanding of basic genomic organization and function. The items most frequently missed included misconception number 1, where the responses of 58% of the faculty and 74% of the students reflect the belief that the insulin gene exists only in pancreatic cells; and number 2, where 56% of the faculty and 74% of

the students selected “DNA sequence” as a laboratory measure of gene expression. Items that tested understanding of commonly used terms, including genotype (misconception number 4), dominant (numbers 6, 8, and 9), and heterozygous (number 6), were answered incorrectly by a large percent of both faculty and students. Other misconceptions related to the function of a gene (number 3), the characteristics of the BRCA gene (numbers 5 and 10), and importance of genetic counseling in families with a history of heart attacks at a young age (number 7).

Discussion

Patients may suffer adverse consequences when health care practitioners lack knowledge and understanding of basic genomics. Misconceptions can easily lead to flawed clinical decision-making and erroneous or incomplete patient education. Three clinical scenarios that rely on a nurse’s understanding of genomics concepts are discussed below: decision support about risk assessment for breast cancer, counseling about carrier testing for recessive diseases, and using genotype data to select and dose medications.

Genetic testing for susceptibility to breast cancer

As genetic testing to estimate one’s probability of developing breast cancer becomes mainstream, nurses in many settings will be asked to explain the tests and help clients decide whether to be tested. Women being treated for breast cancer often get their questions answered by their oncologist or a genetics nurse or counselor. However, nurses working outside of those specialties also need to know how to respond to questions. A genomically competent nurse can potentially reduce cancer morbidity and mortality by applying knowledge of basic concepts related to genetic screening and testing for cancer.²³ This could occur in various settings where nurses provide cancer care, including ambulatory surgery centers and community or primary care clinics, where patients or families may pose questions about genetic testing for themselves or

their blood relatives. Without a solid understanding of the basic science related to genetic testing, the clinician is at risk of conveying misinformation on critically important questions.

In this study, 53% of the faculty and 40% of the students believed that if a woman tests negative for BRCA1, she must lack the BRCA1 gene. In reality, all persons, both male and female, have the BRCA1 gene, but only a small percentage have a mutated form that is associated with a predisposition to breast or ovarian cancer. Furthermore, more than one-third of faculty and students believed that different women with BRCA1 mutations had identical mutations; in fact, hundreds of different BRCA1 mutations have been identified, most of which have not been determined to be harmful.

Carrier testing for recessive diseases

Autosomal recessive conditions are sometimes referred to as “surprise” diseases because neither parent is affected. Examples include cystic fibrosis (CF), Tay-Sachs disease, phenylketonuria (PKU), and sickle cell disease (SCD). When both unaffected parents are heterozygous for the mutation, each of their children has a 25% chance of inheriting the disease gene from both parents, which would make the child homozygous for the mutation and affected with the disease. Carrier screening, also called heterozygote screening, is available for many autosomal recessive conditions, so couples may learn their risk for having an affected child prior to pregnancy.²⁴ Some of these diseases, such as Tay-Sachs, are lethal in childhood; others, such as CF, PKU, and SCD are amenable to treatment. Thus, genetic counseling and decisions about testing must consider the nature of the disease as well as the values, beliefs, and resources of the individuals being tested.

Nurses in any setting should have an understanding of the basic principles related to patterns of inheritance and carrier testing for recessive conditions. A typical scenario might

involve the grandmother of a child with SCD asking a pediatric clinic nurse about the likelihood of her other pregnant daughter having a baby with the disorder. To accurately educate this grandmother, the nurse needs to first ascertain the blood relationships among these family members and then explain the recessive nature of SCD. It is possible that the pregnant daughter is at high risk of having a child with SCD, but only if her partner is also a carrier. The results of this study (numbers 6 and 8 on Table 1) suggest that nursing faculty and students have misconceptions about the definitions of heterozygous and recessive. Furthermore, 42% of faculty and 36% of students (number 9) misunderstood the use of carrier testing.

Pharmacogenomics

Pharmacogenomics is a rapidly emerging technology that determines how an individual's genomic makeup will affect response to medications. The goals of pharmacogenomic testing are to optimize drug efficacy and decrease adverse drug reactions by identifying patients who are at greater risk. Tailoring drug selection and dosage regimens to the patient's genotype has the potential to maximize patient safety, enhance outcomes, and reduce costs. Many pharmacogenomic tests are already in use and have improved care for patients taking anticoagulant, antiretroviral, antidepressant, and pain medications.²⁵

As pharmacogenomics emerges, nurses should assist patients to understand the meaning of the tests. In this study, 36% of the faculty and 57% of the students misunderstood the meaning of "heterozygous" in a genetic test result for a mutation related to metabolism of warfarin, a common anticoagulant medication. Metabolism of drugs can vary according to the patient's genotype, so nurses need a clear understanding of basic genomic concepts to correctly and confidently communicate essential information to their patients. Practice innovation requires that all members of the interprofessional team, including nurses, understand genomic language.²⁶

The scenarios depicted above reflect misconceptions about foundational concepts uncovered in this study, but other challenges are sure to confront practitioners as genome science continues to advance in unpredictable ways. Although the GNCI evaluates a grasp of concepts that are unlikely to become obsolete, newer concepts not included in the inventory, such as gene editing technologies and epigenetic effects, will require patient counseling in the future.⁶

Implications for Teaching

Despite the critical need for nursing education that integrates genomics, this study reveals that nursing faculty and students share misconceptions about foundational genetic/genomic concepts. Such “wrong thinking” can put patients and families at risk for bad decisions based on erroneous information. For example, a first-degree male relative of a woman with a deleterious BRCA1 mutation whose nurse fails to inform him about the gene’s equal prevalence in men may neglect to encourage his children to seek early screening. Likewise, nurses caring for patients with conditions like sickle cell disease or cystic fibrosis who misunderstand the recessive inheritance pattern might miss opportunities to counsel at-risk relatives about carrier testing.

Identifying misconceptions begins with remediating one’s basic knowledge about a rapidly advancing discipline. It is imperative that nursing faculty seek out relevant educational programs for their own development to ensure that appropriate information, including basic concepts, be included in nursing curricula to foster the genomic competence of future clinicians. Fortunately, a number of such resources are available online; these are described in Table 2.

Conclusions

Identifying “wrong thinking” is important when designing educational strategies for students who will care for patients with genomic concerns. Nurse educators who recognize misconceptions are able to highlight and attempt to reconcile them; this strategy promotes

meaningful learning so that nurses are able to apply knowledge in practice. In addition, the public is likely to share the same misconceptions as nursing students and faculty. One of the essential genomic competencies calls for nurses to provide genomic information tailored to clients' knowledge level and literacy. Nurses who are aware of common difficulties in understanding genomics are better prepared to meet this competency. For these reasons, understanding genomic misconceptions provides actionable information to support the education of a nursing workforce competent to practice in the genome era.

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Table 1: Misconceptions of faculty and students about genomics concepts

| Misconception | Percent of faculty selecting (n=495) ^a | Percent of students selecting (n=1002) ^b |
|--|---|---|
| 1. The insulin gene is contained in pancreatic beta cells (vs. all nucleated cells) | 58 | 74 |
| 2. A laboratory test of gene expression might examine the DNA sequence of the gene (vs. mRNA transcribed from the gene) | 56 | 74 |
| 3. The primary function of a gene is to determine a particular trait (vs. to direct formation of a specific protein[s]) | 46 | 71 |
| 4. “Genotype” refers to the traits or characteristics determined by one’s genes (vs. an individual’s total collection of gene variants) | 58 | 53 |
| 5. A woman tested positive for a hereditary breast cancer mutation (BRCA1) but her sister tested negative. This means that the sister who tested positive has the BRCA1 gene in her DNA and the sister who tested negative lacks the BRCA gene in her DNA (vs. both sisters have the BRCA1 gene but the sister who tested positive has an altered copy). | 53 | 40 |
| 6. A person who is heterozygous for a mutation associated with response to warfarin has two copies of the gene-one is dominant and one is recessive (vs. has two non-identical copies of the gene) | 36 | 57 |
| 7. When creating a genetic pedigree, a genetic referral should be made for a person whose mother had breast cancer at age 64 (vs. a person whose father had a heart attack at age 43) | 30 | 61 |
| 8. A person with an autosomal dominant disease is equally likely to have one altered copy or two altered copies of the disease gene (vs. one normal copy and one altered copy) | 36 | 53 |
| 9. Carrier testing may be done to see if an asymptomatic individual carries either a dominant or recessive gene that could be passed to offspring (vs. a recessive gene that could be passed to offspring) | 42 | 36 |
| 10. Different women with BRCA1 mutations likely have identical mutations (vs. unique BRCA1 mutations) | 36 | 40 |

Incorrect responses selected on the Genomic Nursing Concept Inventory (with correct response in parentheses).

^a from Read & Ward, 2016 ^b from Ward, Purath, & Barbosa-Leiker, 2016

Table 2: Resources on genomics for nursing faculty

| Resource and weblink | Description |
|--|--|
| Genomic Nursing Cyberhub (Washington State University) https://genomicnursing.wsu.edu | Educational and research support for genomic nursing education. Links to genomic nursing competencies. |
| Genetics/Genomics Competency Center (National Human Genome Research Institute of the U.S. National Institutes of Health) www.g-2-c-2.org | Curricular materials for health professionals |
| National Genomics Education Programme (National Health Service, United Kingdom) https://www.genomicseducation.hee.nhs.uk | Information, links, and educational materials for health professionals. |