To the Faculty of Washington State University:

The members of the Committee appointed to examine the dissertation of LINDA D. WARD find it satisfactory and recommend that it be accepted.

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ACKNOWLEDGMENT

Like all dissertations, this one represents the outcome of an endurance event, spanning a far longer time than the four years of a doctoral program and reflecting contributions of a wide assortment of people. My natural interest in science, cultivated by my junior high science teacher Mr. Hilliard, never wavered over forty years. In 2003, the year of the genome, I found myself at the National Institute of Health’s Summer Genetics Institute. Over eight weeks, my cohort of genetic nurses insisted I would “of course” enter a PhD program, an idea I persistently renounced. My interest was in integrating genomics into nursing education; my mantra was “I don’t want to study genomic nursing education; I want to do it.” And so I did, in fits and starts, for several years. When our own university launched a nursing doctoral program, I saw an opportunity to implement the research proposal I developed at the NIH. An instrument development study would allow me to both study genomic education and “do” it, while the doctoral program provided a vehicle to complete the work I envisioned. And so I began.

Treasured friends and colleagues made my endurance event a blessing. I am grateful for the insight and unfailing support of my doctoral cohort, the wisdom and gentle rigor of my chair, Dr. Haberman, and the encouragement and expertise of my committee. I appreciate faculty and students at the College who allowed me into their classes, thoughtfully answered endless genomics questions, and good-naturedly fielded questions about methods or writing or psychometrics. Most of all, I am grateful to my family, especially my husband David (who curiously thought getting a PhD was a good idea, even as he patiently sorted 6,000 index cards and proofread 12,000 data points), and my mother, who I think might have enjoyed being in class with me. Finally, I appreciate my children, grandchildren, siblings, my dad, and friends—who all showed the good grace to (mostly) bite their tongues when I went back to school “at my age.”
DEVELOPMENT OF THE GENOMIC NURSING CONCEPT INVENTORY

Abstract

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Nurse educators worldwide are charged with preparing a workforce to deliver competent genetic and genomic based care. In the United States, a set of broadly endorsed competencies (the Essentials) define genetic and genomic competency for nurses. Certain foundational knowledge, defined as genomic literacy, is necessary for nurses to achieve genomic competency. The level of genomic literacy among nursing students is unknown, and no validated measure of that knowledge is available. Concept inventories are research-based assessments that look like multiple-choice tests. Designed to distinguish students who understand basic concepts from those who do not, concept inventories are increasingly used in science and technology education although none have been developed for nursing. The purpose of this instrument development study was to design, test, and conduct initial psychometric evaluation of a tool to measure conceptual knowledge underlying the Essentials. Key concepts were extracted from the competencies and validated with 104 genetic nurse experts. Student supplied response surveys of 96 to 134 baccalaureate nursing students elicited textual responses to open-ended questions about the key concepts. Analysis of 6,000 textual responses identified the level of understanding of key concepts and common misconceptions. A 55-item draft inventory was constructed, utilizing misconceptions as item distractors. Cognitive think-aloud interviews with 15 nursing students informed item revision. The revised inventory was pilot tested with 238 baccalaureate nursing students. Psychometric analysis informed inventory reduction, retaining the most
psychometrically robust items while preserving the initial content domain. The product of this study is a 31-item beta version of the Genomic Nursing Concept Inventory (GNCI © Ward 2011). The inventory’s psychometric characteristics are unknown pending further testing; however, estimates of scale performance are promising. Estimated scale difficulty is 53%, with item difficulty of .26 to .83. Average item-total correlation is .33, indicating adequate discriminatory power, and Cronbach’s alpha is .804, suggesting sufficient reliability. Although the GNCI requires further validation and large scale testing, it holds great promise to provide a reliable and valid ruler by which to measure genomic literacy. Such a ruler is required to inform curriculum and course design and evaluate outcomes in genomic nursing education.
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CHAPTER 1
INTRODUCTION

Statement of the Purpose

The purpose of this research was to develop and complete initial psychometric evaluation of the Genomic Nursing Concept Inventory (GNCI), an instrument designed to measure knowledge of essential genetic and genomic concepts among baccalaureate nursing students. Ausubel’s Assimilation Theory provided the theoretical framework for this study, which also utilized the assessment triangle as a psychometric model and the essential nursing genetic and genomic competencies as a conceptual framework.

Specific Aims

The specific aims of this study were to:

1) identify key genetic and genomic concepts which
   (a) are most critical to genetic and genomic literacy, and
   (b) underlie essential genetic and genomic nursing competencies;

2) identify student misconceptions about those key concepts;

3) develop and refine a concept inventory to measure knowledge of those key concepts, utilizing student misconceptions as item distractors; and

4) complete pilot testing and describe the initial psychometric properties of the concept inventory.

Significance and Rationale

Signaled by the completion of the Human Genome Project in 2003, rapid and escalating discovery in genome science has initiated a paradigm shift in healthcare and nursing science. Every disease and health condition is now known to have a genomic basis; that is, health derives
from a complex interplay of genetic and environmental factors. Consequently, health care professionals, including nurses, must begin to view health and disease through a genomic lens. As a profession, nursing has committed to developing a workforce prepared to deliver genomic-based care and endorsed a set of competencies to support that commitment. Nursing education is charged with the implementation of genomic nursing education. However, almost nothing is known about the level of genetic and genomic knowledge among nursing students at either the beginning or end of their programs, and no ruler exists to measure the knowledge underlying the genetic and genomic competencies. Developing a tool capable of measuring the body of knowledge required of nurses to deliver genetic- and genomic-based care will add to nursing science and provide a useful measure for nursing education. Such a tool is needed now.

**Arrival of the Genome Era**

The completion of the Human Genome Project (HGP) in 2003 publicly heralded the dawn of the genomic era in health care. Advances in genetics and molecular biology developed in the context of the HGP demonstrate a genetic or genomic component to all diseases and health conditions. Medical genetics, itself a relatively new field of study, has given way to human genomics. Genomics is the study of how the total DNA complement of an individual or a population, in concert with all manner of environmental factors, contributes to human health and illness. Whereas genetics focuses on relatively rare single gene disorders, genome science embraces, as well, common chronic disorders of complex and multifactorial origin. Simply stated, genomics takes a holistic view of genetics. Genome-based approaches are playing an increasingly important role all along the health continuum in disease prevention, screening, diagnosis, treatment and survivorship. Genomics has become a central science in health care (Consensus Panel, 2006).
In addition to its anticipated profound impact on health care, genomics has captured the public interest. On popular television shows, genome science is applied to solve forensic challenges. People are increasingly aware of genomic implications in common health conditions as well. On an almost daily basis, articles in the popular press announce the discovery of an association between a specific genetic variation and one disease or another. The public is encouraged to document their family health history using online tools and to present the resulting pedigree to their healthcare provider for interpretation and advice. Individuals can even order a genomic profile online to discover their personal risk for disease.

Nursing’s Response

Nearly three million nurses stand along the front lines of health care in the United States at the intersection of escalating advances in genome science and an increasingly informed and interested public. The translation of genome science into healthcare practice requires nurses to become familiar with new terminology, concepts, skills and technologies. Genomic knowledge is expanding exponentially, largely due to rapid advances in DNA sequencing technology and robotics which shrunk the cost of sequencing a genome 10,000-fold in just four years (Singer, 2009). Even though translation of research findings into clinical practice is occurring much slower than the blinding pace of knowledge acquisition, a growing knowledge-to-practice gap in genetics and genomics faces all healthcare professionals. Nurses, in particular, must prepare to provide a wide range of genomic services, such as:

- helping women understand the implications of BRCA testing on breast cancer risk,
- delivering gene-based therapies such as monoclonal antibodies,
- understanding gene polymorphisms modulate endorphin release with exercise, making it more difficult for some people to maintain regular physical activity,
- eliciting and documenting a three-generation family history,
- helping a client to understand limitations of direct-to-consumer genetic testing,
  and
- anticipating variable responses among individuals to “standard” medication doses,
  due to pharmacogenomic effects.

In fact, genome science provides a new lens through which nurses should view people. The reductionist view of medical genetics, in which inheritance of a relatively rare gene alteration directly causes disease, has given way to a much broader genomic view. To see people through a genomic lens requires the nurse to consider the collective influence of multiple gene variations and the cumulative effects of all manner of environmental factors on an individual’s health, risk for disease, response to therapies, even behaviors. The genomic era of healthcare represents a paradigm shift to which nurses must respond.

There is little doubt that nurses must achieve genomic literacy to respond to the demands of modern healthcare. Rationales for achieving genomic literacy include:

1. Genomic research occupies a prominent position in the National Institutes of Health (NIH) roadmap (http://nihroadmap.nih.gov/), and genome science is being translated to clinical practice across healthcare settings. Evidence-based practice requires nurses to apply research findings to patient care. Anticipating increasing application of genome science in healthcare delivery, nurses must prepare to provide genomically-based care.

2. Nurses are compelled to engage in lifelong learning and continual professional growth in order to achieve and maintain knowledge relevant to the current scope and standards of nursing practice (American Nurses Association [ANA], 2004). Consequently, nurses have a duty to update their own knowledge as the science of nursing changes. Nursing professional
groups and regulatory agencies agree that understanding of genome science is foundational to the scope and standards of nursing practice (ANA/ISONG, 2007; Benner, Sutphen, Leonard, & Day, 2010; Consensus Panel, 2006).

3. Nurses must respond to the changing healthcare needs of the public. Public awareness of and interest in genetic-based tests and interventions are rapidly increasing, and there are far too few genetic specialists to meet the growing demand for genetic healthcare. The public will increasingly expect nurses to use genetic and genomic information and technology when delivering care (American Association of Colleges of Nursing [AACN], 2008; ANA, 2001; Consensus Panel, 2006). In many healthcare settings, nurses interact with patients more than any other health professionals. Therefore, nurses have a particular need to understand concepts of genetics and genomics and prepare to administer and explain genomic applications, many of which are not yet developed, to patients.

4. An essential component of baccalaureate nursing education is a liberal education foundation to generate responsible citizens in a global society (AACN, 2008). Nursing education must prepare graduates to keep pace with changes driven by research and technology and provide both leadership and education as clinical, ethical, legal and social implications of genomic health care evolve. Colleges of nursing must also prepare students to provide leadership in policy-making around such issues as genetic testing, newborn screening, and gene therapy.

Like any change in the health care environment, arrival of the genome era creates uncertainty and role ambiguity for nurses (Jenkins, 2000). Nurses in all settings, including practicing nurses, nursing administrators, deans, faculty, and nursing students must recognize the relevance of genomic knowledge and then work to achieve genomic literacy. Nurse educators in clinical practice face an enormous task to deliver genetic and genomic knowledge to the nursing
workforce. Likewise, colleges of nursing must update curricula and prepare new graduates for clinical application of genomic healthcare.

**Genetic/Genomic Knowledge among Nurses**

In many ways, nurses are well prepared for the activities that comprise genomic healthcare, having specific training in history taking, multidimensional assessment, the referral process, and effective communication of complex information. However, another critical tool is lacking among nurses: genetic and genomic literacy (Wright, 2001). Research over the last three decades has consistently documented a lack of genetic knowledge among practicing nurses, nursing faculty, and nursing students (Cohen, 1979; Edwards, Maradiegue, Seibert, Macri, & Sitzer, 2006; Maradiegue, Edwards, Seibert, Macri, & Sitzer, 2005; Peterson, Rieger, Marani, deMoor, & Gritz, 2001; Prows et al., 1999; Scanlon & Fibison, 1995; Williams, 1983). Few colleges of nursing include human genetics as required content, either as a prerequisite or within the curriculum (Monsen et al., 2000). Therefore, as genome science is increasingly translated into healthcare practice, the gap between what nurses know and what they need to know grows wider. Since this gap begins at the undergraduate level, the focus of this research is on foundational knowledge of genetics and genomics among baccalaureate nursing students.

**Genetic/Genomic Knowledge of Nursing Students**

Students should come to nursing school with a fairly broad understanding of genetics. In the United States, National Science Education Standards (NSES) established in 1996 recommend specific genetics concepts be included in science education in grade and high school (Center for Science, Mathematics, and Engineering Education, 1996). Although the NSES are thought to be influential, they are only guidelines, and individual states set their own science learning
standards. Differences among teachers, textbooks, and laboratory access are likely to contribute to nonuniformity of genetic education.

In addition to the genetic content students receive prior to college, natural science courses required as nursing prerequisites offer further opportunity for genetics education. However, no recent data are available about the level of genetic and genomic knowledge among undergraduate nursing students. In fact, only two published studies of genetic knowledge among nursing students were found on literature review. One of those explored perceived genetic knowledge among advanced practice nursing students (Maradiegue et al., 2005) and the other occurred over 30 years ago (Cohen, 1979). It is reasonable to think the level of genetic knowledge among baccalaureate nursing students may be similar to that of other undergraduate college students. However, a review of the literature is unrevealing about genetics knowledge among the general college population, as well. This state of affairs may be about to change, however, as three different instruments have been recently developed to assess genetics knowledge among college students (Bowling et al., 2008; Elrod, 2007; Smith, Wood, & Knight, 2008). All three tools were developed with the primary intent of testing achievement and measuring educational outcomes.

The Essentials

In 2005, a group of nurse leaders in the United States established a minimum basis for preparing the nursing workforce for the genome era. The Consensus Panel (2006) published a set of 27 competencies (the Essentials), which translated genetic and genomic knowledge, skills and attitudes into nursing activities that collectively constitute genetic- and genomic-based care. The Essentials have been endorsed by over 40 nursing organizations and regulatory boards, including the ANA and both nursing education accrediting organizations, i.e., the National League for
Nursing Accrediting Commission and the AACN (Consensus Panel). Three years later, a second edition of the *Essentials* added outcome indicators, which explicate the knowledge and clinical practice indicators specific to each competency (Consensus Panel, 2009). The *Essentials* provide the necessary benchmark for assessing the level of genetic and genomic knowledge of nurses, whether they are nursing faculty, nursing students, or practicing nurses. In other words, the *Essentials* reflect the critical knowledge that constitutes genetic and genomic literacy among nurses. That knowledge set is the target of this assessment tool.

*Measuring Genetic/Genomic Knowledge*

A number of tools have been developed and validated to measure genetic knowledge, primarily among undergraduate biology majors (Bowling et al., 2008; Elrod, 2007; Smith et al., 2008). Those instruments tend to address genetic, rather than genomic knowledge, and many of the items focus on biologic mechanisms rather than clinical applications. Previous investigations of genetic knowledge among practicing nurses have generally utilized tools lacking psychometric validation and those, too, have had a decidedly genetic focus (Peterson et al., 2001) or have measured perceived, rather than actual, knowledge (Edwards et al., 2006; Maradiegue et al., 2005). To date, no published studies have systematically assessed genetic and genomic knowledge among nurses using the *Essentials* as the benchmark. Furthermore, although others are working on a tool for practicing nurses (personal communication, K. Calzone, November 9, 2008), no validated instrument is currently available to measure knowledge underlying the essential nursing genetic and genomic competencies.

*What This Study Will Add*

It is important to be able to accurately measure genetic and genomic knowledge among nursing students. Now that consensus has been reached regarding *what* nurses need to know, an
important next step is to develop a psychometrically rigorous tool to measure that knowledge. The goal of this dissertation is to develop an instrument to measure genetic and genomic knowledge, as identified in the Essentials, among baccalaureate nursing students. Data generated by such a tool will be useful to inform curriculum and course development, measure educational effectiveness, and support outcome evaluation for courses and/or curricula.

Theoretical Framework: Assimilation Theory and Misconception Research

The implementation of genomic nursing education can be informed by science education research, which has amassed a large body of knowledge about how students’ conceptual understanding of the world affects their learning. A good deal of this research is based on Ausubel’s Assimilation Theory, a cognitive learning theory positing that concepts held in a student’s cognitive structure are of primary importance as students learn new material (Ausubel, Novak, & Hanesian, 1978). That is, over years of formal education and personal experiences, students develop ideas and perceptions about how the world works. When those preconceptions are not congruent with prevailing scientific explanations in that discipline, they are considered to be erroneous (Cho, Kahle, & Nordland, 1985). A number of terms have been applied to such erroneous perceptions: alternative conceptions, misconceptions, commonsense alternatives, naïve beliefs, faulty mental models, and alternative frameworks (Cho et al.; Michael, 2007). The most commonly-used term in science education research is misconception; that term will be used here.

Educational research indicates that misconceptions are firmly held by a significant proportion of students. According to Hestenes, Wells, & Swackhamer (1992), misconceptions should be regarded as “reasonable hypotheses grounded in everyday experience” (p. 142) and accorded respect. Misconceptions tend to be ingrained in students’ cognitive framework and are not easily corrected by traditional educational approaches (Hestenes et al.). They often impede
conceptual understanding of new content, because students cannot easily integrate new
knowledge that doesn’t fit their pre-existing conceptual framework. In fact, misconceptions may
not even be apparent to students or educators. Students may perform well academically despite
persistent flaws in conceptual understanding by resorting to ‘recipe learning’ of factual
information (Mazur, 1992). Identification of student misconceptions not only uncovers targets
for instructional remediation, but also provides anchors for designing instructional strategies

**Physics Education and the First Concept Inventory**

A substantial body of research has accumulated about misconceptions, primarily in
science education. The physics education community led the way in applying misconception
research to education. In the late 1980s, David Hestenes at Arizona State University discovered
that academically strong physics students often lacked conceptual understanding of Newton’s
laws of gravity. They could apply formulas to solve problems and had historically performed
well on exams despite lacking understanding of basic physics concepts (Hestenes et al., 1992).
Hestenes went on to develop a method to measure conceptual understanding. His tool, the Force
Concept Inventory (FCI), has been credited with initiating substantial reform in physics
education, and its use has been tied to positive educational outcomes (Richardson, 2004). The
FCI became the prototype for a number of instruments designed to measure conceptual
understanding in undergraduate science, technology, engineering and mathematics (STEM)
education. Concept inventories (CIs) have been developed for broad domains such as biology,
statistics, and astronomy, as well as narrow content areas such as natural selection, diffusion and
osmosis, and star formation. Concept inventories are based on cognitive learning theory and a
model known as the assessment triangle.
The Assessment Triangle

The assessment triangle is a psychometric model applied broadly in educational assessment, the discipline charged with determining how well students are learning and providing outcome measures of educational effectiveness. Educational assessment applies recent advances in cognitive and measurement sciences in order to (1) identify the best evidence of learning, (2) develop accurate means of measuring that evidence, and (3) maximize the interpretation of those measures (National Research Council, 2001). The model ties together three components, or pillars, of assessment: the mechanism of learning (cognition), the activities by which learning is demonstrated (observations), and the evaluation of those activities to reflect learning outcomes (interpretation). Application of the model involves applying the best available knowledge about how people know and learn, i.e., contemporary theories of learning and knowing, during the design and interpretation of assessments. The assessment triangle has guided the development of both large-scale standardized tests and smaller classroom assessments to find out what students know (National Research Council). Concept inventories are among the research-based assessment tests developed using the assessment triangle.

Concept Inventories

Concept inventories are instruments designed to measure conceptual, rather than factual knowledge. Based on misconception research, CIs have been shown to discriminate between students who understand basic concepts and those who simply memorize unconnected ideas (D’Avanzo, 2008). The design and use of concept inventories are guided by the assessment triangle model. Concept inventories look like multiple choice tests, however the distractors are written to reflect common misconceptions about the concept of interest. When the goal of education is to promote the application of information to perform problem solving and other
relevant tasks, a concept inventory provides a useful tool for documenting concept-based learning (Michael, Modell, McFarland, & Cliff, 2009). This is most certainly the case in genomic nursing education. The rapid pace of genome research and constant evolution of didactic information demand conceptual understanding rather than rote memorization. Therefore, the strategy must be to provide students with conceptual knowledge along with critical thinking skills required to provide safe and effective genomically-based patient care as clinical applications unfold.

Concept inventories are constructed for a target population. Here, that population is baccalaureate nursing students. Concept inventories are most useful when a body of knowledge is well defined. Typically, a set of key concepts or “big ideas” are gleaned from learning objectives in a particular course or discipline (Michael et al., 2007). In the case of genomic nursing education, the Essentials provide the overarching framework from which the “big ideas” most relevant to genomic healthcare were extracted.

At least three genetic concept inventories have been developed and validated in recent years; each is targeted toward undergraduate college students (Bowling et al., 2008; Elrod, 2007; Smith et al., 2008). However, no CI specific to either healthcare or nursing, or having a genomic focus, has yet been developed.

The National Research Council (2001) cautions “the more purposes a single assessment aims to serve, the more each purpose will be compromised” (p. 2). Nevertheless, a validated inventory offers versatile support to the implementation of genomic nursing education. As a diagnostic tool, the inventory will reflect student misconceptions about genetic and genomic concepts to inform teaching. As an evaluative tool, it can be used as a pre- and post-test to measure learning outcomes. It can be used as a placement exam. A CI can even be used by
students to track their own progress in understanding concepts and identify content areas in which further study is indicated (Garvin-Doxas & Klymkowsky, 2007). In the future, it may be validated for nursing faculty and/or practicing nurses to inform faculty and staff continuing education. A genomic content inventory for nursing has immediate appeal for the basic education of baccalaureate nursing students and future applications to bring all of nursing into the genome era.
CHAPTER 2
LITERATURE REVIEW

Literature across multiple disciplines informed this research and is described here. First, concepts underlying genomic nursing education are explicated, in order to create a clear foundation for discussion. Next, attention is turned to the changes in medicine and nursing that over the last two decades have led to the need for genomic nursing education. The current state of genomic nursing education is then explored, to understand the context in which the Genomic Nursing Concept Inventory was developed. Finally, the fields of educational philosophy and assessment are tapped for the best current information about how students learn and how a tool might be designed to measure evidence of learning. This broad review of the literature is necessary to understand, in context, the nature and extent of genomic nursing education and how to measure achievement of learning that supports genomic competency.

Conceptual Underpinnings

A number of concepts are embedded within genomic nursing education and must be explicated. Imogene King described concepts as “abstract representations of persons, objectives, and things in the world” (1986, p. 14). The more abstract a concept is, the greater its need for clarification. David Ausubel, whose theory of concept assimilation provides the theoretical framework for this study, defines concepts as “objects, events, situations or properties that possess common criterial attributes and are designated by some sign or symbol” (Ausubel et al., 1978, p. 56). Conceptual understanding is not static. In the last decade, the concept of genetics has changed with acquisition of new knowledge borne of the Human Genome Project, and during that time the term *genomics* has permeated both professional and popular writing. Unfortunately, the meanings of *genetics* and *genomics* are often conflated, even in professional
Efforts to resolve concept ambiguity around genetics and genomics must occur early in the process of implementing genomic nursing education. Other –omics have emerged during recent years, as well; these warrant explication, as do concepts of genomic health care, genomic literacy and competency, and genomic nursing education.

**Genetics**

Genetics is the study of individual genes and their protein products (Guttmacher & Collins, 2002). Genes, as functional and physical units of DNA, are located at specific sites along chromosomes. Collectively, the approximately 23,000 genes that make up the human genome account for only about 1.5% of human DNA; the vast majority of human DNA does not encode protein (Green, 2010). The role of a gene is to direct the formation of one or more functional proteins in amounts adequate to support normal physiologic activities. A change in DNA sequence within a gene (known as a polymorphism or mutation) may cause the formation of a defective protein with altered function. Thus, a typical genetic condition occurs when a single gene, or both copies of gene alleles, carries a mutation that interrupts normal gene function. This is the basis for traditional genetic conditions.

Medical genetics, the study of heritable diseases, is a relatively new discipline, born after the end of World War II on the heels of the ignominious eugenics movement. The heritable disorders addressed by medical geneticists are fairly rare, mostly single-gene disorders of predictable, or Mendelian inheritance. Traditional genetic diseases include phenylketonuria, cystic fibrosis, sickle cell anemia, and Huntington disease. While they inflict enormous burden on affected families, all Mendelian disorders together account for only 5% of the total healthcare burden in developed countries (Yoon et al., 2001).
Genomics

Although the term genome has been used for nearly a century to indicate the entire DNA sequence of an organism, genomics is a much newer concept. The roots of genomic medicine lie in the decoding of the human genome, accomplished in 2003. Genomics is the study of the total DNA complement of an individual or a population, including environmental effects on gene expression. Genomics involves not just the small fraction of DNA contained in genes, but also DNA that lies between genes and does not encode protein. About 3% of noncoding DNA is preserved across mammalian species and thought to be functionally important, although specific functions of intergenic DNA are not well understood (Green, 2010). Some noncoding DNA sequences are known to control gene expression, i.e., whether a gene is actively making protein, which protein it is making, and in what quantity. Genomics addresses these and other contributors to gene expression, including interactions between genes as well as effects of environmental factors. Therefore, whereas a typical genetic condition is related to malfunction of a single gene, genomic conditions develop due to contributions of multiple genes and are often modulated by environmental effects. Evidence of genomic expression is accumulating for virtually all diseases and health conditions. Common chronic disorders such as hypertension, diabetes, asthma, heart disease, and cancer have genomic causes. That is, genetic traits (nature) in concert with multiple environmental factors (nurture) contribute to gene expression and possible alterations in health.

The relative contributions of genes and environment vary widely among health conditions. For example, infectious diseases such as AIDS and tuberculosis have long been considered to be of environmental origin: Individuals who avoid acquiring the pathogen simply don’t develop the disease. However, genetic factors are also involved in both disease acquisition
and progression. A commonly-cited example is alteration in a gene designated CCR5, which encodes a human immunodeficiency virus (HIV) co-receptor. Individuals with two altered copies of the gene do not acquire HIV despite repeated exposures (Nussbaum, McInnes, & Willard, 2007). At the other end of the spectrum are classic genetic disorders such as cystic fibrosis (CF). The cause of CF, mutation of the calcium channel gene CFTR, is well described and purely genetic. However, morbidity and mortality associated with CF are affected by environmental factors. Consider a CF clinic in Minneapolis where the mean survival of people with CF exceeds the national average by over 14 years. Their remarkable success is attributed to diligent management of a collection of modifiable environmental factors (Gawande, 2005). In short, all diseases and health conditions have both genetic and environmental contributors: All diseases are genomic.

Jenkins and Calzone (2007) caution that the definitions of genetics and genomics must remain works in progress since ongoing research will change the understanding of genome science. Their caution is salient. As an example, a widely-used definition of genomics offered in 2002 by Drs. Alan Guttmacher and Francis Collins of the National Human Genome Research Institute (NHGRI) is, in light of new knowledge, inadequate. Their definition of genomics as the “study of the functions and interactions of all the genes in the human genome, including their interactions with environmental factors” (2002, p. 1513) fit the state of the science at that time, one year prior to the completion of the Human Genome Project. Just three years later, however, in the same journal, Guttmacher and Collins (2005) reported that about two thirds of biologically important DNA sequence lies not within but between genes and does not encode protein. Discovery that noncoding DNA has important roles in health requires revision of the definition of genomics to include all DNA sequence, not just that represented by genes.
Proteomics

The history of genetic discovery can be described as a process of drilling down to the root causes of health and disease. The concept of traits first described by Mendel was followed by the discovery of DNA, the physical “stuff” by which traits are inherited and expressed. Traits were mapped to chromosomes, then genes, and finally to variations as trifling as a single nucleotide substitution at a critical site somewhere in the 3.2 billion nucleotides comprising the human genome. DNA is a long double helix polymer of repeating units called nucleotides. The four base nucleotides found in DNA are adenine, thymine, cytosine and guanine. The molecular biology of most single-gene disorders is well understood: A glitch in DNA sequence, such as a change in the order of nucleotide bases, is faithfully copied as a protein encoded by that gene is formed. The result is a protein that functions abnormally or not at all, leading to disease. Or, a glitch in a noncoding stretch of DNA causes a protein to be produced in abnormal amount, or at the wrong time, or in an inappropriate tissue, thereby altering health. Although the cause of single-gene disorders can be traced to an exact DNA locus, the mechanism of disease occurs at the protein level. In order to understand disease, one must back away from a reductionist focus on DNA sequence to consider the protein products of genes.

Proteomics is a relatively new discipline investigating the structure, function, and interactions of a full complement of proteins in a cell or an organism (Childs & Valle, 2000). Estimates of the size of the human proteome range from about 300,000 proteins (Pierce, Fakhari, Works, Pierce, & Clancy, 2007) to as many as a million (Green, 2010). Clinical proteomics focuses on identifying and studying proteins that are medically relevant (Pierce et al.). Nurses must understand basic concepts of proteomics in order to provide genomically competent care. It is not the nucleotide sequence per se that impacts health; rather, it is the downstream effect of
variations in the nucleotide sequence on protein production, structure and function (Sulston & Ferry, 2002). Proteins form the structural and functional elements of cells, tissues, organs and organisms, and virtually all physiologic functions are mediated by proteins. Drug receptors, ion channels, neurotransmitters, enzymes, mediators of intercellular communication, antibodies, transport molecules—all are proteins, whose structure and hence function depend on the DNA sequence in their encoding gene. One’s proteome is the collection of proteins that can be produced according to the instructions encoded in one’s genome. Proteomes differ between individuals largely to the same extent that their genomes differ.

Other -omics

Genome science has rapidly branched into a number of fields of study that share not only ties to the genome but a common suffix. As such, -omics has become something of a neologism, or at least a neo-suffix. Proteomics is perhaps the best established, but other -omics are appearing more commonly in the literature. For example, metabolomics is the study of chemical processes that involve cellular metabolites. Several –omics are of particular relevance to nursing.

Pharmacogenomics is a genomic application of pharmacogenetics, the study of how gene variations affect drug metabolism and drug effects. Proteins are involved in all the molecular actions of pharmacokinetics and pharmacodynamics—they constitute the enzymes, receptors, transporters, channels, and cell communication molecules that mediate both therapeutic and adverse drug effects. Variations in genes that encode these proteins are responsible for individualized responses to medications, both in terms of efficacy and adverse reactions. The promise of pharmacogenomics is personalized prescribing, adding yet another “right” to several pre-existing rights of medication administration: the right medication for the right genotype.
Transcriptomics is the study of the messenger RNA of an organism or a tissue. A transcriptome reflects the small percentage of a genome that is actually transcribed in order to form proteins. As such, it reflects gene expression. Clinical applications of transcriptomics are perhaps most common in oncology, where microarray technology is used to discover which growth regulatory genes are being over- or under-expressed in a particular tissue. Genomic profiling of cancers supports the development of targeted therapies and guides the selection of chemotherapeutic agent based on the profile of specific tumor cells.

Microbiomics is the study of the collective genomes of microbes that populate a host. In a human body, microbial cells outnumber human cells by a factor of ten to one, and the proteins produced by microbial genomes may be implicated in health and illness. The Human Microbiome Project (HMP) is a NIH-funded effort to examine the genetic makeup and gene expression of microbes that inhabit healthy adults. The HMP is an example of a metagenomic study, i.e., a study of a collection of genomes from a mixed community of organisms (NHGRI, 2009).

A diseasome is a network of gene-disorder pairs in which a mutation of a particular gene is correlated with one or more disorders. The hypothesis suggests that certain essential human genes are expressed in many tissues, and proteins with critical functions are likely to play a role in multiple disease processes. Mapping of the human diseasome is ongoing, with a goal of creating a conceptual framework that reveals common genetic origins for many disorders and allows greater understanding of the human interactome (Goh et al., 2007).

Not all of the –omics necessarily hold bright promise for healthcare. Within the genomics community, “recreomics” is a name assigned to the practice of obtaining information about disease prediction and prognosis via direct-to-consumer (DTC) genetic testing, without critical
evaluation of the accuracy or usefulness of the information (Oetting, 2009). While DTC genetic testing has the potential to inform individuals about their personal risk for common disease, lack of regulation of testing and reporting raises concern about individual interpretation of test results. Nurses are likely to have a role in helping people understand the implications and limitations of DTC genetic testing.

**Genomic Healthcare**

By the time the Human Genome Project had been completed, the human genetic code lay available to all in a public database (http://www.ncbi.nlm.nih.gov/projects/mapview/map_search.cgi?taxid=9606/). However, the HGP provided more than the human DNA sequence. An additional bequest was refinement of technology necessary to mine that code for variations associated with human health and illness. Optimistic forecasts called for genome science to transform the prediction, prevention, diagnosis and treatment of disease with a promise of personalized healthcare. Genomic knowledge would allow the prediction of specific disease risk among individuals, families and populations, encompassing not only genetic influences on health, but environmental factors as well (Collins, Green, Guttmacher, & Guyer, 2003). Health screenings would be targeted to those at highest risk to develop disease. Diagnostic methods would be refined to uncover specific molecular signals associated with early disease, and treatments would be directed to precise molecular targets, avoiding the “collateral damage” of treatment side effects. Even adverse drug effects would be avoided by applying pharmacogenomic principles. The genome era was broadly proclaimed to signal a healthcare revolution, and contagious enthusiasm led to forecasts which, in retrospect, were perhaps overly optimistic. As early as 1998, Francis Collins, NHGRI Director, predicted that pharmacogenetic testing would be “routinely” performed “in the very near future” by
primary care physicians before writing prescriptions (Collins, 1998). Predictions about availability of the $1000 personal genome profile have been made and then deferred repeatedly over the last decade. Discussions of ‘hope’ versus ‘hype’ have become common in genome literature, as clinical applications of genome science have been far outpaced by dazzling progress in genome research. While the transformation of healthcare that was predicted upon completion of human genome sequencing may yet occur, genome science has encountered similar delays in translation to practice as other areas of research. And why should genome science be any different? Citing delayed and limited uptake of aspirin therapy for patients with coronary artery disease, Lenfant (2003) noted “Let’s be realistic: If we didn’t do it with aspirin, how can we expect to do it with DNA?” (p. 873).

Although nursing has not yet been radically transformed by genome science, effects of genome science are beginning to permeate all aspects of healthcare (Khoury et al., 2009). Genomic healthcare is simply care tailored to an individual based on genetic information (Green, 2010). For nurses, genomic healthcare incorporates genomic tools into nursing practice and is operationalized in the Essentials (Consensus Panel, 2006). Genomic tools are not all high-technology molecular manipulations: On the contrary, one of the most fundamental, the three-generation family history, is almost disappointingly mundane. Nurses are accustomed to collecting family history of highly heritable, traditional genetic conditions; one competency calls for nurses to expand that history to include common chronic (genomic) conditions and display the information in the form of a pedigree. However, genomic healthcare involves more than a set of skills. It also requires nurses to adopt a new world view or paradigm and see patients through a genomic lens. Current knowledge of genomics incites, at the bedside, an acute awareness of the interface of heredity and environment. Individual patient characteristics including race and
ethnicity, risk for disease, response to therapy, and even behaviors are influenced by genes and environmental factors acting together. Is the person who experiences uncontrolled postoperative pain following a standard dose of narcotic a drug seeker? Or might s/he be a non-addicted person in misery, harboring a genetic variation that prevents metabolism of the drug to its active form? Is the obese person with diabetes who just won’t exercise lazy and nonadherent to the provider’s admonishment to be active? Or might s/he have altered genes that block endorphin release with physical activity? Does a family share a predilection for oxycodone abuse because of common learned behaviors? Perhaps, but they could also share a particular form of opioid receptor that produces a highly exaggerated sense of euphoria with each narcotic dose. To view a patient through a genomic lens is to recognize and consider the interplay of nature and nurture on health and illness.

*Genomic literacy and competency*

For more than half a century, a handful of nurses have championed the addition of genetic content to nursing education and worked to define exactly *what* nurses need to know about genetics. As Lashley (1997) pointed out, “. . . not every nurse needs to be a genetics expert, but there is content nurses must know in order to provide competent nursing services” (p. 15). Several content outlines have been proposed over the years, and the *Essentials* document represents current consensus on the set of knowledge and skills required by nurses to deliver genomically competent care. However, it is crucial to note that the *Essentials* are written as competencies and do not specifically delineate the fund of knowledge required for nurses to achieve genomic proficiency. In a number of domains, terms such as literacy and competency are used to describe knowledge or proficiency. In general, literacy is more closely aligned with
knowledge, while competency infers the ability to apply that knowledge. The terms are, however, sometimes conflated and warrant examination to provide conceptual clarity.

Literacy

Literacy in general is understood to be content and context specific (Ratzan & Parker, 2006). Much has been written about the body of knowledge that constitutes genetic or genomic literacy. Kaphingst (2009) considers genomic literacy in terms of health literacy, applying the definition that was used in both Healthy People 2010 and the 2004 Institute of Medicine (IOM) report on health literacy: “Health literacy is the degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions” (IOM, 2004; Ratzan and Parker, 2000, p. vi). This is a functional definition, describing a level of understanding necessary to support a specific role. So defined, literacy cannot be achieved by rote learning alone but requires a level of knowledge that falls at the application level or higher in Bloom’s taxonomy (Anderson & Krathwohl, 2001).

Ratzan and Parker (2000) describe health literacy as the currency needed to navigate an increasingly complex health care system. Genomic literacy might be considered similarly, i.e., as the currency necessary to apply genomic principles in the context of one’s personal and/or professional roles. The degree of understanding that constitutes genomic literacy varies according to the context in which it is applied. For example, Jennings (2004) suggests that citizens require genetic literacy sufficient for informed consumerism, i.e., to be able to provide informed consent for a genetic test or gene-based therapy. For nurses, however, the literacy requirement is greater. Nurses have a role in the delivery of genetic services and management of genetic information and a responsibility to advocate for clients within the healthcare system (ANA/ISONG, 2007). Nurses also have a civic duty to provide leadership in healthcare policy
decision making, which may increasingly address genetic and genomic issues (AACN, 2008). The various activities by which nurses provide genomic health care are subsumed in the Essentials competencies. For nurses, then, genomic literacy requires knowledge sufficient to carry out the activities that make up those competencies.

According to Kaphingst (2009), genomic literacy requires certain conceptual knowledge along with oral and print literacy skills and numeracy skills. Nurses therefore require conceptual knowledge that underlies the Essentials competencies, along with the specific language required to understand and articulate issues in genomic healthcare. Surely the delivery of genomic healthcare does not require nurses to have detailed knowledge of genetic mechanisms. It does, however, require an understanding of genetic and genomic terminology and a solid grasp of the underlying concepts of genome science. Nurses must, for example, understand the central dogma of genetics, i.e., that DNA is transcribed into RNA, which is then translated to form proteins, and that proteins represent the collection of enzymes, receptors, neurotransmitters, signaling molecules, and structural units that mediate all physiological activities. Nurses must understand that, while variations in DNA and hence in proteins play fundamental roles in disease development and therapeutic response, the expression of genetic variation is modulated by environmental effects. Nurses must understand that genetics also has a role in driving patient behaviors, since studies in behavioral genomics have linked gene polymorphisms with substance abuse, anger, risk-taking behaviors, domestic violence, optimism, even gambling (Plomin, Defries, Craig, & McGuffin, 2003). Genomic literacy supports nursing activities such as genetic assessment and testing, gene-based therapies, pharmacogenomic and nutrigenomic applications, and a greater understanding of the role of genetics in human behavior. The scope of genomic literacy required to support these professional activities is subsumed by the nursing Essentials
which outline, in the most recent edition, specific areas of knowledge to support each competency (Consensus Panel, 2009).

**Competency**

In the last half century, competency-based educational frameworks have become dominant in health professional education, largely replacing knowledge-based models (Carraccio, Wolfsthal, Englander, Ferentz, & Martin, 2002). The competency approach has paralleled and perhaps been driven by the development of national educational and practice standards, which delimit the set of knowledge, skills and attitudes thought to represent competence in a particular domain. The shift to competency-based education has raised important questions about what it means to be competent and how (or even if) competency can be measured.

The Centers for Disease Control, in developing competencies for public health professionals, defined competencies as “applied skills and knowledge (blended with behaviors)” that enable effective practice (Centers for Disease Control [CDC], 2001). Based on a medical education meta-analysis, Carraccio et al. (2002) defined a competency as “a complex set of behaviors built on the components of knowledge, skills and attitudes” and defined competence as “personal ability” (p. 362). Whitcomb (2002), writing about medical residents, pointed out that competence implies the ability to provide services “in accord with practice standards established by members of the profession and in ways that conform to the expectations of society” (p. 359). He suggested that knowledge, skills, and attitudes are necessary but not sufficient to achieve competency. Also required is the ability to translate knowledge, skills and attitudes into a set of complex behaviors that result in the delivery of high-quality care. This translation requires critical thinking, decision making and interpersonal skills. Benner (1982) noted that competency
implies not only skill application in real situations, but also desired outcomes: “Competency . . . is the ability to perform [a] task with desirable outcomes under the varied circumstances of the real world” (p. 304).

Evaluation in competency-based education requires a functional analysis of an occupational role, whereby elemental activities that make up the role are identified and translated into outcomes, and proficiency is evaluated on the basis of demonstrated actions related to those outcomes (Leung, 2002). While the movement toward competency-based education in health care has been inexorable, it has been plagued by difficulty in developing appropriate evaluation tools (Benner, 1982; Carraccio et al., 2002). This should not be surprising, given the transactional nature of health care practices. If the end point of an evaluation is a patient outcome, then evaluation must take place in the real world of clinical practice. As Benner (1982) points out, the performance of even basic nursing skills (e.g., insertion of a nasogastric tube) requires one set of actions when it is performed on a student volunteer or a manikin, and quite another when it is performed on a real patient. In fact, Husserl (as cited in Benner, 1982) saw the task of listing criteria for competencies in human activities as endless, given the infinite variety of human responses, each requiring the addition of new sub-competencies. Whether the task is endless or just impossible, the fact remains: Knowledge and skill acquisition are simply easier to measure than knowledge and skill application. However, Benner cautions that important competencies must not be ignored just because of lack of technology to measure them.

Fortunately, the aim of this research is to develop a tool to measure knowledge, not competence. The specific fund of knowledge comprising the target of this tool is drawn from a set of well-defined competencies and represents a relatively high order of knowledge, i.e., conceptual knowledge of fairly complex mechanisms and applications. Still, the task of
identifying key concepts subsumed by the nursing *Essentials* and developing an assessment of those concepts is much more straightforward than developing an assessment of proficiency in the delivery of genomic care. If genomic literacy is the level of understanding necessary to achieve genomic competence, then the goal of this tool is to assess genomic literacy among nursing students. Genomic literacy is necessary but not sufficient for genomic competence. As Benner (1982) reminds us, nursing competence develops over years, not over a semester. However, the development of genomic literacy might very well occur during a semester, or a year, or a curriculum. Only by having an appropriate, psychometrically rigorous assessment tool, can we begin to understand systematically the genomic literacy of nursing students.

*Genomic Nursing Education*

The concept of genomic nursing education is operationalized in the *Essentials* (Consensus Panel, 2009), which explicates not only the genetic and genomic competencies expected of professional nurses, but also the underlying knowledge, attitudes and skills necessary to provide genomically competent care. According to the *Essentials*, “Each nursing curriculum preparing registered nurses for practice . . . should include genetic and genomic learning experiences sufficient for all registered nurses to be proficient in the essential competencies” (p. 38). The sum of the 27 essential competencies, each of which is described along with underpinning knowledge and clinical practice indicators, constitutes the gold standard for genomic nursing education.

*Genetics and Genomics in Healthcare*

Genomic education represents an innovation to be diffused into not just nursing, but all health professional education (Core Competencies Working Group, 2001). Diffusion is affected by a broad collection of factors, including but not limited to social constructions around the
innovation (Rogers, 1995). Therefore, it is important to investigate the broader context in which an innovation diffuses. Because nursing, medicine and healthcare are inextricably bound, consideration of genetics and genomics in the context of medicine in particular and healthcare in general is necessary. Such consideration uncovers two important issues: that the genome era represents a paradigm shift within healthcare, and that medicine as a discipline will not be able to meet the genomic needs of patients, families and populations.

There is little doubt that genomics has initiated a paradigm shift in health care. As described by Kuhn (1996), a paradigm is an “implicit body of intertwined theoretical and methodological belief” (p. 16) which provides an accepted model or pattern to guide scientific research. Reflecting current best knowledge within a discipline, a paradigm supplies a “preformed and relatively inflexible box” (Kuhn, p. 24) into which nature is attempted to be forced. As current best knowledge changes, a paradigm will no longer fit the science, and for science to develop, the paradigm must be replaced. Kuhn describes the development of science as a “successive transition from one paradigm to another via revolution” (p. 12). Paradigm changes are often driven by advances in technology. Such is the case with genomic medicine, in which the sequencing of the human genome signaled the beginning of the revolution (Guttmacher & Collins, 2005).

In 1999, anticipating the completion of the human genome project, Barton Childs depicted the “new” genetics as a trigger for a new way of thinking in medicine, which he labeled a new “logic of disease.”

But now genetics is exposing medicine to concepts that provide new ways to think about disease, its causes, and its pathogenesis. We have no choice but to examine how these ideas are influencing medical thinking today, how they should do so, and how we might use them to make medical education more relevant to what is happening in communities no less than in laboratories. In short, we should ask how genetics might be integrated with medicine. (1999, p. ix)
That the new logic of disease described by Childs represents a paradigm shift has been recognized widely in healthcare (Ginsburg & Willard, 2009; Guttmacher & Collins, 2005; Jenkins, Grady, & Collins, 2005; Singh, 2003; Weiner, Thomas, Goodspeed, Valle, & Nichols, 2010). A paramount moment in the shift occurred on April 14, 2003, with the announcement of completion of the Human Genome Project (Collins et al., 2003). True to Kuhn’s description, the paradigm shift was driven by advances in technology and has been described by many experts as a revolution (Couto & Cardiff, 2008; Parikh, Johnson, & Merchant, 2008; Wilkinson & Targonski, 2003).

Writing in the late 1990s, Childs did not ascribe the term genomics to the new logic of disease. He did distinguish between traditional genetics (which he called medical genetics) and what he called genetic medicine. As described by Childs, genetic medicine is based on the interplay between genes and environment and is synonymous with what is currently called genomics.

It is the genes that are transmitted from one generation to the next. Then, after providing the organism with an outline for its structure and development and after specifying the molecules that are the engines of cells, organs, and individuals, the genes become the servant of all of these, responding to signals from within and without that promote choices among the options inherent in the outline and that enable uniquely individual open systems to live in congruence with the environment. (1999, p. 6).

The paradigm shift described by Childs suggests a new way to view health and illness, seeing patients, families and populations through a genomic lens. While he was addressing physicians, one need only replace “medicine” with “nursing” to create an equally relevant message. In fact, for nurses, who are well versed in holism, the paradigm shift may be smaller in scale.

There is no argument that the discipline of medicine will fall far short of meeting the public’s genomic health care needs. This was recognized as early as 1994 in an Institute of
Medicine report, which predicted that nurses and other healthcare providers would have to play a critical role in providing genetic services (Andrews, Fullarton, Holtzman, & Motulsky, 1994). Since then, the need for genetic and genomic services has expanded exponentially, far beyond the capability of the current genetic workforce in the United States, which includes approximately 3000 genetic counselors, 1250 medical geneticists, and 38 certified genetics nurses (Wicklund, 2008). Nurses, representing the largest group of health care professionals and practicing in a variety of settings, will increasingly be called upon to provide genomic health services (Consensus Panel, 2006). If nurses cannot meet the challenge, then other disciplines will step forward and fill the void. Nursing already missed a similar opportunity in the 1970s, as described below, when the expansion of genetic testing created a need for healthcare professionals to perform genetic assessments and help patients understand test results.

*Genetics in Nursing: An Historical View*

The call for nurses to prepare for the impact of genetics on health care dates to 1962. That same year, as Massachusetts initiated the first state-mandated newborn metabolic screening program, two nurse educators wrote of the need for nursing curricula to include basic genetics education. Brantl and Esslinger (1962) specifically recommended the inclusion of “basic genetics concepts and their application to nursing” in nursing curricula (p. 92). In intervening years, their message has echoed repeatedly in the literature, but nursing has been slow to respond. In 1997, Lashley reported that nursing had not yet committed to genetics and consequently missed opportunities in that growing field. She referred to the birth of the new profession of genetic counseling in the 1970s, when burgeoning use of genetic testing created a need for professionals who could provide education and guidance to patients: “... [N]urses should have been the natural group to fulfill this role and they were not” (p. 15). By the time of Lashley’s writing, the
Institute of Medicine report had already called for nurses and other healthcare professionals to prepare to help patients understand the genetic implications of common chronic illness such as heart disease, diabetes, cancer, and mental illness (Andrews et al., 1994).

In 1984, nurses with a shared interest in genetics came together to form the Genetics Nurse Network, an informal group that by 1988 was incorporated into a nursing professional organization, the International Society of Nurses in Genetics (ISON). Today, ISONG consists of nearly 350 members (B. Kassalen, personal communication, June 16, 2009), many of whom have provided critical leadership in genetic healthcare.

In 1996, The American Medical Association, the ANA, and the NHGRI came together to support genetics education for all health care professions and create the National Coalition for Health Professional Education in Genetics (NCHPEG). Novel in its multidisciplinary approach, NCHPEG’s goal was to encourage healthcare professionals to integrate genetic knowledge, skills and attitudes into routine health care and provide a framework for educators to respond to emerging demands for genetics education (NCHPEG, 2005). The coalition represents over 100 diverse organizations including health professional societies, genetics organizations, consumer and voluntary groups, private businesses, managed care organizations and government agencies.

The NCHPEG chose to utilize a competency, rather than a knowledge or process based framework, with a focus on the application of genetic knowledge, attitudes and skills to routine health care. In 2001, they published a report entitled Core Competencies in Genetics Essential for All Health-Care Professionals, listing 35 basic competencies to represent the collection of knowledge, skills and attitudes recommended for health professionals in all disciplines in order to provide genetically competent care (Core Competencies Working Group, 2001). Nine additional skill-based competencies were recommended for health professionals who provide
direct genetic counseling services. The working group pointed out that the competencies must represent a work in progress, anticipating rapid advances in genetics would accompany the nearly-completed Human Genome Project. Indeed, the NCHPEG competencies have been revised twice since their initial publication in 2001. Of interest, the language in the original document revealed a clear emphasis on genetic science; the word genome was mentioned just once (in the context of pharmacogenomics). In a second edition, NCHPEG (2005) reported on the impact of the core competencies, documenting their dissemination through professional literature and describing their use in health education. While the competencies remained unchanged and the language retained a distinct genetic focus, discussion of the purpose, background, and implementation of the competencies reflected advances in genome science during the intervening four years. A third edition in 2007 collapsed the 35 basic competencies into just 18 and omitted competencies specific to genetic counseling (NCHPEG, 2007). The language remained strongly genetic.

Nursing had a strong presence in the development of the core competencies; in fact, an ISONG member, Jean Jenkins, was the chair of the NCHPEG working group. The NCHPEG core competencies received significant, but far from overwhelming support from nursing organizations. In 2002, a strongly supportive position statement was issued by the American Academy of Nursing (AAN), a member organization of NCHPEG. The AAN recommended that baccalaureate and higher degree nursing programs adopt the NCHPEG competencies and integrate supporting genetic content into their curricula. The AAN further recommended the NCHPEG competencies be included in accrediting criteria for baccalaureate and higher degree nursing programs and continuing education requirements for professional nurses (Lea, 2002).
In the meantime, the AACN had revised *The Essentials of Baccalaureate Education for Professional Nursing Practice* (1998), a document outlining the expectations of baccalaureate-degree nursing graduates. Although the Human Genome Project was still years from completion, the AACN anticipated its effect on health care and included genetics as an important knowledge area. The ability to take a genetic history and assess genetic risk factors were stipulated as core competencies. The ANA, which had already provided leadership in the founding of NCHPEG, was not far behind. Its house of delegates voted in 1999 to support genetics in basic, graduate, and continuing education for all nurses (Badzek, Turner, & Jenkins, 2008). In 2000, an expert panel convened by Health Resources and Service Administration (HRSA) called for expanded genetics education for nurses. Acknowledging genetics to be the central science for all health professions, the report offered nine recommendations for improving genetic competence among nurses. Emphasis was placed on interdisciplinary efforts to increase genetic education for all healthcare professionals in preparation for expanded genetic applications predicted to evolve from the Human Genome Project. While the language in the report has a genetic (rather than genomic) focus, HRSA emphasized preparing a nursing workforce for the future and noted that genetic knowledge includes genomics.

On April 14, 2003, completion of the Human Genome Project was announced and the era of genomic medicine took a giant leap forward. The paradigm began to shift on that first DNA Day from traditional genetics to genomics (Collins et al., 2003). Guidelines, recommendations and competencies prepared after that date increasingly reflected the need for nurses and other health care professionals to develop genetic and genomic literacy.

By 2004, collaboration had begun to develop a set of genetic and genomic competencies specific to nursing. Factors prompting these efforts included a pressing need for oncology nurses
to develop genetic and genomic proficiency and concern about the limited diffusion of the NCHPEG competencies into nursing education. Some nursing leaders believed the NCHPEG competencies, which numbered 35 at the time, were too comprehensive and not realistically achievable in already packed curricula. There was interest in a streamlined genomic competency model that was being developed in the United Kingdom (UK) (Jenkins & Calzone, 2007). Using the NCHPEG competencies as a template, nurses in the UK identified just seven competencies believed most relevant to nursing and developed learning outcomes and practice indicators for each (Burke & Kirk, 2006). In that setting, nurses from the NHGRI and the National Cancer Institute set about to define a set of essential genetic and genomic competencies specific to registered nurses (Jenkins & Calzone). A steering committee of nurse leaders from clinical, research and academic settings as well as from professional nursing organizations reviewed and compared competency recommendations that had been developed for diverse groups of health professionals. The consensus process was completed by 2005, and the resulting set of essential nursing competencies in genetics and genomics was quickly endorsed by the ANA and 47 other nursing organizations (Consensus Panel, 2006). Divided into professional responsibilities and professional practice domains, the Essentials include 27 role activities that represent genetically and genomically competent care. They represent the minimal amount of competency expected of every registered nurse, regardless of academic preparation, practice setting, role or specialty (Consensus Panel). Although the Essentials share much with the NCHPEG core competencies, they take a nursing perspective and, developed in the wake of the Human Genome Project, have a decidedly genomic flavor. The phrase “genetic and genomic” is repeated throughout the document, and consideration of the combined effects of genetics and environment on health is emphasized.
Given the exponentially increasing pace of genome research, the body of knowledge that denotes genetic literacy is destined to change. Therefore, the Essentials, like the NCHPEG competencies, are presented as a work in progress. However, for nurses in the United States, the Essentials represent a definitive benchmark for nursing proficiency in delivering genetically and genomically competent care.

The fact that the Essentials were written as competencies led to multiple attendant difficulties in creating evaluation tools to measure genetic and genomic literacy. While the competencies were clearly described in the 2006 document, the body of knowledge required of nurses to achieve competence was not explicated until 2009, when a second edition of the Essentials added outcome indicators for each of the 27 competencies. Outcome indicators were designed to guide didactic and clinical education by outlining genetic and genomic learning experiences sufficient for the development of genomic proficiency (Consensus Panel, 2009). The outcome indicators deconstruct each competency, identifying specific areas of supporting knowledge and suggesting clinical activities (written as performance indicators) that reflect proficiency for that competency. Much of the hard work involved in measuring genetic and genomic literacy among nurses has therefore been completed by the Consensus Panel. Collectively, the specific areas of knowledge identified for the 27 essential competencies should represent the knowledge set that denotes genetic and genomic literacy for nurses.

Strategic planning is ongoing to integrate the essential competencies into nursing curricula, with strong support from the AACN. Baccalaureate education is charged with preparing nurses to understand concepts of science and keep pace with changes driven by research and technology (AACN, 2008). The current AACN Essentials of Baccalaureate Education for Professional Nursing Practice includes sixteen specific references to genetics
and/or genomics in the curricular recommendations for preparing students for a generalist nursing role. Genetics and genomics are listed among the core concepts required for nurses to practice in today’s “ever-changing and complex healthcare environment” (AACN, p. 3).

**Current State of Genomic Nursing Education**

The historical view described above illustrates some real strengths in nursing’s ability to adopt genomic education. Nursing leadership in genomics is strong. Dr. Jean Jenkins led the NCHPEG core competency task force, and Dr. Elizabeth Thomson is the clinical genetics program director of the Ethical, Legal and Social Implications (ELSI) research division at the NHGRI; both are nurses. The international genetic nursing professional organization, ISONG, has a strong presence in the United States. Genomics enjoys a prominent position on the NIH Roadmap for Medical Research (NIH, 2009). Nursing advisory and regulatory boards formally endorse genetic and genomic nursing education, although that support has not yet been translated into the inclusion of genomic-based questions on licensing and certification exams. For example, the 2010 NCLEX-RN® Detailed Test Plan includes a single mention of genetics, i.e., listing *collection of a genetic history* among exam content areas (Wendt, Kenny, & Schultz, 2010). A vast collection of rich educational resources is widely available for anyone, including nurse educators, who wish to become genomically competent (Maradiegue, 2008). Free web-based tutorials, downloadable presentations and video clips are available, as well as residential and online programs such as those sponsored by the National Institute of Nursing Research (http://www.ninr.nih.gov/training/) and Cincinnati’s Children’s Hospital Medical Center (http://www.cincinnatichildrens.org/ed/clinical/gpnf/). The availability of these resources reflects the open, nonproprietary sharing of information modeled by the HGP. Finally, nurse researchers are publishing important findings in genetics and genomics, evidenced by over 900 articles.
indexed in CINAHL by genetics or genomics and nursing since Brandt and Essinger first sounded the call in 1962.

There remains, however, much work to be done. Preparing nurses to practice in tomorrow’s healthcare environment, while mandated by the AACN and HRSA, is particularly challenging in the face of rapid technologic change. Regardless of the level of genetic and genomic knowledge of students entering nursing school, understanding genetics and genomics in the context of nursing practice requires the implementation of genomic nursing education. That in turn requires nursing faculty to achieve a certain level of genetic and genomic literacy.

Although few studies have assessed genetic knowledge among nursing faculty, experts agree that faculty are likely to know little about genetics or genomics. Alarmingly, no published studies describe faculty knowledge from a genomic view. Anderson, in a 1996 state-of-the-science paper, summarized the body of research around genetics education in nursing in the United States and identified ‘need for knowledge’ as the primary recurring theme. Since then, a scant body of research has consistently documented very limited perceived or actual knowledge of genetics among practicing nurses (Peterson et al., 2001; Scanlon & Fibison, 1995), nursing faculty (Edwards et al., 2006; Prows et al., 1999), and advanced practice nursing students (Maradiegue et al., 2005). No published studies report genetic or genomic knowledge among undergraduate nursing students. A realization that nurses and nursing faculty have limited genetic knowledge should not be surprising. With an average age of 43 years for practicing nurses and over 50 years for nursing faculty (AACN, 2009; Buerhaus, 2008; HRSA, 2010), neither group is likely to have received substantial genetic content or any education about genomics in their own formal or continuing education.
Because faculty cannot teach what they do not know, it follows that nursing programs likely include limited genetic/genomic content. Indeed, studies of both undergraduate and graduate nursing programs have revealed little coverage of genetic or genomic issues (Edwards et al., 2006; Hetteberg, Prows, Deets, Monsen, & Kenner, 1999; Prows et al., 1999). Of particular interest, Hetteberg et al. found no increase in genetic content from 1980 to 1996, despite significant advances in molecular biology and genetics during that time period. An audit of nursing textbooks found wide variation in content, with pediatrics and maternal child health texts accounting for most of the genetic content (Monsen et al., 2000). Concentration of genetic content in pediatric and childbearing texts reflects a traditional genetic focus on birth defects and single-gene disorders.

Efforts to increase genetic and genomic knowledge among nurses will surely fail if nurses don’t believe the content is important to nursing practice. For years, nursing educators have struggled with content overload, and genomics is one of several evolving content areas vying for limited space. Findings about perceived relevance of genetics and genomics among nurses are limited and conflicting, with wide variation (Cragun, Couch, Prows, Warren, & Christianson, 2005; Edwards et al., 2006; Hetteberg et al., 1999; Peterson et al., 2001; Scanlon & Fibison, 1995). For example, a group of senior nurses in the United Kingdom felt they didn’t need to know very much about genetics currently or “for some time“ (Pfeil & Chi-Mei, 2005, p. 1130), while 95% of a group of advanced practice nursing faculty found genetics to be very relevant to nursing education (Edwards et al.). Some faculty consider science already to be overrepresented in nursing education, favoring more emphasis on the art of nursing, i.e., caring and holism (Jordan, Davies, & Green, 1999).
Barriers to integrating genetic and genomic content into nursing education have been investigated in multiple studies, and those data are consistent. Limited knowledge of genetics and genomics among nursing educators, packed curricula with no room for new content, limited time to update knowledge and course content, questionable perceived relevance to nursing education, lack of genetic questions on the NCLEX exam, and lack of faculty resources have been repeatedly cited as impeding the integration of genetic and genomic content (Anderson, 1996; Edwards et al., 2006; Hetteberg et al., 1999; Prows et al., 1999). State boards of nursing and nurse credentialing organizations have provided little incentive for colleges of nursing to address genetics education. As of 2005, only 10% of states required colleges of nursing to include genetics content (Prows, Glass, Nicol, Skirton, & Williams, 2005). Little information is available about the presence of genetic or genomic questions on nursing certification exams. The most recent study, published over a decade ago, indicated negligible coverage of genetic conditions and almost no genomic questions on registered and advanced practice nursing licensing and certification examinations (Lea, Jenkins, & Monsen, 1999).

The level of genetic and genomic knowledge among nursing students is unknown and likely unexplored: No assessments of genetic knowledge among undergraduate nursing students are published. There is cause to believe nursing students likely have some understanding of fundamental genetic concepts. Genetic content is specifically included in the National Science Education Standards, recommendations put forth in the United States in 1996. Life sciences standards stipulate that students in grades 5 through 8 receive instruction in about the “facts, concepts, principles, theories and models” of heredity and that students in grades 9 through 12 receive similar instruction in the molecular basis of heredity and biological evolution (Center for Science, Mathematics, and Engineering Education, 1996, p. 106). The Standards specifically
include DNA structure and function, genetic change, and genetic variation in the recommended content. In addition, nursing students should receive genetic content in their prerequisite college biology course. Hott et al. (2002), in a noteworthy review of 357 college biology instructors, found an average of 10.8 hours (range 0 to 48) spent on genetic content in introductory biology courses for nonmajors. It is quite possible, although not demonstrated, that nursing students may have a higher level of genomic literacy than their faculty.

The implementation of genomic nursing education logically begins with assessment. It is necessary to assess student and faculty understanding of genomic concepts, as well as their attitudes about the relevance of genetics and genomics to nursing education and practice. Findings will inform both curricular planning and faculty development.

The population of interest for this dissertation is nursing students, and the domain of interest is genetic and genomic knowledge. Learning what students know about genetics and genomics is critical in order to plan an appropriate curricular approach. The knowledge of interest is genomic literacy, i.e., the set of genetic and genomic concepts underlying the essential competencies. That knowledge set is both content specific and complex. The content must correlate closely with the Essentials, and the complexity of understanding lies at the analysis, synthesis and evaluative levels of Bloom’s taxonomy.

Fortunately, educators in science, technology, engineering and mathematics have spent years developing tools to measure conceptual knowledge in well-defined dimensions. The products of their research are assessments known as concept inventories, and the methodology for creating concept inventories is well described. Development of a genomic concept inventory for nursing will provide a means to learn what students know about the genetic and genomic concepts that underlie the essential nursing competencies.
Concept Inventories

The current approach to educational improvement involves setting challenging academic standards and then measuring students’ progress toward meeting those standards (National Research Council [NRC], 2001). Applying this approach to genomic nursing education, it becomes apparent that the academic standards have already been established with the essential competencies. Missing, however, is a method to measure student learning in this domain. The lack of a means to evaluate student learning is not trivial. Educational assessment is integral to improved education as a means of providing feedback to all stakeholders about the effectiveness of educational services. High-quality educational assessment tools are needed, including both large-scale and classroom assessments, and their design should be informed by contemporary theories of cognitive science and current psychometric modeling (NRC). Concept inventories are educational assessment tools that meet both these criteria. Ausubel’s Assimilation Theory, a cognitive learning theory, represents the underlying learning theory for concept inventories, and a conceptual framework known as the assessment triangle provides the psychometric model (Richardson, 2004). Concept inventories have the benefit of being easy to administer and score, thereby allowing efficient data collection about conceptual understanding within a domain (Dufresne, Leonard, & Gerace, 2002).

The first concept inventory was developed about twenty years ago in response to concern about the failure of physics students to develop understanding of basic physics concepts and their reliance instead on rote memorization of isolated fragments of information (Hestenes et al., 1992). The assumption of concept inventories is that every student approaches physics (or biology or genetics) with a set of preconceptions or well-established beliefs about how the world works. In cognitive learning theory, that set of established beliefs represents the student’s
preexisting cognitive structure (Ausubel et al., 1978). Derived from years of personal everyday experience as well as formal education, these beliefs tend to be commonsense and are well ingrained (Hestenes et al.). Effective instruction must take these preconceptions into account or risk being ineffective in changing them. A considerable body of misconception research indicates that misconceptions persist among even advanced students and graduates of physics programs (Hestenes et al; McDermott, 1984) and in other science disciplines as well (Cho et al., 1985; Odom & Barrow, 1995).

Hestenes and colleagues (1992) developed the first concept inventory (the Force Concept Inventory, or FCI) to measure conceptual understanding, rather than rote knowledge, of Newtonian physics. Their goal was to develop an instrument to probe student beliefs about force, the central concept in Newtonian mechanics, and the various conceptual dimensions that interconnect to make up that greater concept. The FCI attracted considerable attention when it was endorsed by a well-known Harvard physicist, Eric Mazur. Mazur’s reaction when he first saw the FCI was that the questions, which are qualitative questions about basic concepts of mechanics, were too easy for his students. Nevertheless, he began pairing conceptual questions from the FCI with quantitative questions (or problems) addressing the same concept on exams in his undergraduate physics classes. He was surprised to find that 40% of students did better on the quantitative problems than the conceptual problems that Mazur himself found to be simple and basic, even intuitive. Mazur (1992) described slowly coming to understand that bright students were blindly applying problem-solving strategies with little understanding basic physics concepts. This revelation led Mazur to implement a constructivist teaching approach in physics education which was emulated widely, and the FCI has been broadly adopted as an important assessment in physics education (Richardson, 2004).
In the last two decades, concept inventories have been developed for several content domains within physics (e.g., electromagnetic waves and heat transfer) as well as in other disciplines including statistics, astronomy, chemistry and biology (Richardson, 2004). In fact, three concept inventories have been developed for genetics (Bowling et al., 2008; Elrod, 2007; Smith et al., 2008). Concept inventories have been shown to discriminate between students who understand basic concepts and those who simply memorize unconnected ideas (D’Avanzo, 2008).

Concept inventories (CIs) are research-based instruments that look like multiple-choice exams but in which the distractors are crafted to represent common misconceptions in that particular domain. For example, a common misconception in biology is that antibiotic exposure causes bacteria to mutate in a way that produces antibiotic resistance. In truth, mutations are random and sporadic, and the antibiotic-containing environment simply selects for resistant strains. Identifying the misconception allows the CI developer to include the misconception among distractors for an item on the inventory. Therefore, a CI measures not only how many students do not understand a concept, but which alternative conceptual picture they hold instead (Garvin-Doxas et al., 2007). The content addressed by a CI is derived from a framework constructed of the big, overarching themes or core principles that span a course (Michael, 2007). The goal of a CI is to identify students’ conceptual weaknesses in that framework (Garvin-Doxas et al.). Concept inventories are often administered as a pre-test at the beginning of a course or curriculum and again as a post-test. Results are useful to measure student proficiency, evaluate teaching effectiveness, and inform the design of learning strategies to improve conceptual understanding.
A concept inventory is particularly fit for the task of assessing genetic and genomic knowledge among nursing students for two reasons. First, when the goal of education is to develop student ability to apply information for problem solving and other relevant tasks, rather than to encourage recipe learning, a concept inventory provides a useful tool (Michael, 2007). Certainly this is the goal in genomic nursing education. The rapid and escalating pace of genome discovery will lead to increasing but as yet unknown clinical applications. Nurses must play a critical role in implementing those applications as well as helping patients to understand the implications of genomic healthcare. These activities require conceptual knowledge. For example, a nurse who is helping a cancer patient interpret his or her microarray test must understand that the test measures RNA levels, which indicate gene expression in specific tissues. In this case, the nurse’s understanding is based on two important genomic concepts: (a) the central dogma of genetics, i.e., DNA → RNA → protein, and (b) the concept that gene expression varies among cells, even when the DNA in those cells is identical. Simply acquiring factual knowledge about microarray testing is not sufficient to the task.

A second characteristic of CIs also makes them particularly fitting for genomic nursing education. Concept inventories are most useful when the key concepts in a domain are well defined (Treagust, 1988). It is likely no accident that the first concept inventory was developed in physics education, as content boundaries are well established and remarkably uniform across programs in first-level undergraduate physics courses. In genomic nursing education, the essential competencies provide a well-defined and widely-adopted set of underpinning concepts around which an inventory can be designed.

Most concept inventories are designed to be completed in about 30 minutes and include about 25 questions. As a result, concept inventories are limited to a relatively small number of
concepts and provide a narrow view of students’ conceptual knowledge (Richardson, 2004). Developers of concept inventories agree that the identification of concepts most closely aligned with the content domain is a critical step. They also agree that concept inventories should be designed to measure understanding rather than fact-based knowledge. Nevertheless, the nature of those key concepts varies across inventories. Some CI developers argue that key concepts (alternatively called core principles, core ideas, or big ideas) are different than topics (Garvin-Doxas et al., 2007; Michael, 2007; Michael et al., 2009), while others use the terms interchangeably (Wage, Buck, Wright, & Welch, 2005). The difference may be semantic, but a description of core ideas by Duschl, Schweingruber, & Shouse (2007) is useful when considering the scope of target concepts. Duschl et al. describe a core idea as being “well tested, validated and absolutely central to the discipline” and add: “Each [core idea] integrates many different findings and has exceptionally broad explanatory scope” (p. 223). This description makes particular sense when one notes that most concept lists are organized into main concepts and related subconcepts. Duschl’s criteria will be applied during the process of identifying target concepts for the genomic nursing concept inventory.

Richardson (2004) warns that construction of a concept inventory is a “multiyear process involving a team of professionals with expertise in both teaching the subject and in educational assessment” (p. 24). However, all projects must begin somewhere, and the important work of integrating genomics into nursing education cannot wait. This dissertation takes up the challenge of developing a concept inventory to systematically measure genomic literacy of undergraduate baccalaureate nursing students. The project represents an important early step in developing a ruler by which student knowledge can be measured.
Assimilation Theory

Concept inventories are built upon cognitive learning theory, more particularly Ausubel’s Assimilation Theory, and development of a CI necessitates some understanding of that supporting theoretical framework. In fact, the greater task of implementing genomic nursing education might benefit from application of educational psychology principles. Ausubel et al. (1978) describe educational psychology as an applied science with the purpose of identifying “properties of learning that can be related to effective ways of deliberately bringing about stable cognitive changes that have social value” (p. 10). Ausubel adds that the goal of educational psychology is probably the development of a network of related principles that together constitute a comprehensive theory of classroom learning, while recognizing that application of those principles in specific situation is likely more an art than a science.

While a number of learning theories have been described, cognitive learning theory predominates in adult classroom learning in general, as well as in science and nursing education. Cognitive learning theory posits that adult learning involves the assimilation of new content into an individual’s pre-existing cognitive structure, which is a hierarchically organized collection of pre-existing ideas (Ausubel et al., 1978). The organization of cognitive structure is not trivial, because learning occurs when content is linked to a specifically relevant aspect of the existing cognitive structure; this relevant pre-existing idea serves as a critical anchor to the new content (Ausubel, 2000).

Ausubel et al. (1978) describe the goal of education to be meaningful learning, defined as the “long-term acquisition and retention of a complex network of interrelated ideas characterizing an organized body of knowledge that learners must incorporate into their cognitive structures” (p. 12). They contrast meaningful learning to rote learning, the “short-term
acquisition of single, somewhat contrived concepts, the solution of artificial problems, or the learning of arbitrary associations” (p. 12). As new content is assimilated during meaningful learning, a complex bond forms between the pre-existing and new content, and the meanings of both ideas are subject to modification. Meaningful learning has occurred when the component concepts are available in the learner’s cognitive structure with a sufficient degree of clarity (Ausubel et al.).

Meaningful learning has three prerequisites: (a) the material has potential meaning (i.e., is not arbitrary); (b) the learner employs a meaningful learning set (i.e., is predisposed to learning); and (c) the learner’s cognitive structure has relevant anchoring ideas to which the new content can be related (Ausubel et al., 1978). While all these requisites are important in designing and implementing genomic nursing education, the third is particularly germane to concept inventories in general and assessment of genomic literacy in particular.

A learner who lacks relevant anchoring ideas to link new content may resort to rote learning (Ausubel et al., 1978). Rote learning utilizes short-term memory, which in most humans is exceedingly limited in comparison to the far greater capacity of long-term memory (NRC, 2001). More important, rote learning does not result in modification of the existing cognitive structure or provide new anchoring ideas to support acquisition of more complex concepts (Ausubel et al.). Erik Mazur’s graduate students had developed proficiency in utilizing rote memory and applying formulas without evidence that their cognitive structures had ever assimilated basic concepts of Newtonian physics. Concept inventories are designed with the very specific goal of measuring conceptual understanding, as opposed to rote learning. In fact, developers of CIs describe the identification of concepts, rather than topics, within a particular
domain as a particularly difficult step in inventory development (Garvin-Doxas et al., 2007; Michael, 2007).

Success in genomic nursing education requires students to achieve meaningful learning of the body of knowledge that is required for genomic competence. According to Ausubel et al. (1978), the most crucial and variable determinants of potential meaningfulness of learning materials are found in “the availability of . . . relevant content in different learners’ cognitive structures” (p. 44). The existence of relevant anchoring content varies in turn with prior educational background, along with age, IQ, occupation, social class and cultural membership. It is for these reasons that genomic nursing education must begin with assessment. To be meaningful, genomic content must be anchorable to the cognitive structures of nursing students. While students are likely to enter nursing school with some knowledge of genetics and genomics, almost nothing is known about the level of that knowledge or prevailing genetic or genomic misconceptions. Assessing genetic and genomic content within the cognitive structure of beginning nursing students is a necessary first step in planning curricula.

Educational Assessment and the Assessment Triangle

As noted above, the conceptual framework for a genomic nursing concept inventory lies with the essential competencies, and the theoretical framework that guides the current study is that of concept assimilation. Also required is a psychometric model to inform development of a tool capable of assessing genetic and genomic literacy. The assessment triangle is such a model. All concept inventories are educational assessments, and the assessment triangle represents the psychometric model upon they are designed. As such, their development has benefitted from recent work in the field of educational assessment.
In 1998, The Committee on the Foundations of Assessment was convened by the National Research Council with support from the National Science Foundation. A major charge of the committee was to explore how advances in cognitive sciences might best be applied to improve educational assessment, particularly in math and sciences. Identifying a particular need for assessments “that help all students learn and succeed in school by making as clear as possible the nature of their accomplishments and the progress of their learning” (NRC, 2001, p. 18), the committee chose to focus on assessment of student achievement rather than predictive tests of aptitude or ability.

The committee’s work was based on a central tenet of education, i.e., that curriculum, instruction and assessment should be aligned. Effective teaching requires these three primary components of education to reinforce each other and be directed toward the same ends. The committee’s three-year study resulted in a set of recommendations for educational research, policy and practice, published in a report entitled, “Knowing What Students Know” (NRC, 2001). Central to their recommendations was the need to merge the best current science in learning and psychometrics to inform the design of assessments capable of measuring complex knowledge required by contemporary academic standards. The work of this committee led to the development of the assessment triangle. The assessment triangle applies the best current knowledge about (a) how students learn and (b) how to measure evidence of that learning to guide development of assessment tools capable of measuring achievement of curricular objectives (NRC). The model supports the development of large-scale assessments such as national achievement exams as well as smaller classroom assessments. Concept inventories are among the tools based on the assessment triangle.
In the assessment triangle model, the term “assessment” describes both a process and a tool. The process of assessment is defined as “reasoning from evidence” to provide an estimate of what a person knows or can do (NRC, 2001, p. 36). The assessment tool, then, is an instrument “designed to observe students’ behavior and produce data that can be used to draw reasonable inferences about what students know” (NRC, p. 42). The model has two important assumptions. First, like any measurement, assessment always involves some degree of measurement error or imprecision, so that educational assessments provide only estimates of student knowledge or skill. Second, assessments may be used for multiple purposes: assisting learning, measuring student achievement, or evaluating educational programs. However, the more purposes a particular assessment is designed to serve, the more each purpose will be compromised (NRC). Therefore, an assessment should be designed with its primary purpose(s) in mind.

The assessment triangle ties together three foundational elements that underlie all assessments. These elements—cognition, observation, and interpretation—must be explicitly connected during the design of an assessment, or the inferences drawn from the assessment will be compromised (NRC, 2001). See Figure 1.

_Cognition_ refers to the thinking and learning approach that students employ within a given content domain. For this study, cognitive learning theory is applied to a content domain described in the *Essentials*. Over the years, several learning models have been developed, as differential and behavioristic perspectives gave way to more recently-described cognitive and situative views. Specific theories of learning are more or less useful to describe the learning of different types of subject matter, and cognition and learning are better understood in some domains than others. In general, the complex knowledge and skills required by
contemporary educational standards are best considered through a lens of cognitive learning theory, which takes a constructivist approach to education. Likewise, cognitive theory offers the best fit for both nursing and science education. Nursing students must acquire conceptual understanding of knowledge that underlies the essential competencies and then apply those concepts to nursing care. Similarly, the goal in science education is the understanding of basic concepts to provide a foundation upon which more complex content can be added. This level of learning, which lies at the application level of Bloom’s taxonomy and above, is consistent with the competency framework of the Essentials. While factual knowledge is important, conceptual understanding is essential. Ausubel’s concept assimilation theory provides the best model for designing assessments of conceptual understanding and is the favored model for concept inventories.

**Figure 1**
The Assessment Triangle

*Observations* are the tasks students perform to demonstrate knowledge and skills. Observations may take many forms, such as assignments, concept maps, exams, papers, presentations, and real or simulated skill demonstrations. Observations provide evidence that learning has occurred, and assessments must be designed so that the observations reflect, to the
greatest possible degree, student achievement. However, the mental representations and processes that comprise complex learning are not easy to measure. The types of observations capable of reflecting a student’s ability to apply knowledge and skills in an authentic task invariably raise issues of cost, feasibility and psychometric concerns (NRC, 2001). The Committee on the Foundations of Assessment describe the design of an assessment as an opportunity to “structure some small corner of the world” to make observations that reflect student understanding. The goal is to create an assessment in which what students do reflects, as accurately as possible, what they know. Therefore assessments must be carefully designed, through a lens of evidence about how students learn in the given domain, to provide the best possible estimate of proficiency. In the case of genomic nursing education, a concept inventory will represent the “corner of the world” in which evidence of genetic and genomic conceptual understanding is made visible.

*Interpretation* is the process of evaluating observations and describing patterns of response associated with varying levels of student competency. Interpretation leads to inferences about student achievement and relative rankings. For large-scale assessments, statistical models are used to interpret observations, while smaller classroom assessments may utilize a grading rubric or a more informal intuitive or qualitative model. In all cases, interpretation makes the link between observations of student performance and assumptions about student knowledge or skills. Some interpretation models attempt to uncover students’ reasoning behind particular observations, and rules of interpretation describe how a given pattern of responses indicate a student’s grasp or misunderstanding of a particular concept. In the assessment triangle model, methods of interpreting the observation of student performance must be aligned with cognition
and observation, lest the interpretation fail to accurately detect student strengths and weaknesses (NRC, 2001).

In order to design an effective assessment and sound inferences, each element in the assessment model must make intrinsic sense and must connect to each of the other two elements in a meaningful way. The design of such an assessment is therefore an iterative process in which the content domain, teaching and learning theory, assessment method, and interpretation model are integrated and refined, weeding out mismatches and improving consistency.

Benefits of the Current Study

Eight years have passed since completion of the Human Genome Project fueled optimistic predictions about a new era of personalized health care tailored to individual genomes. While the translation of research to practice has been tempered compared to original forecasts, there is little doubt that nurses, like all healthcare professionals, must achieve genetic and genomic competency in order to practice effectively in the genome era. Although barriers to genomic nursing education persist, a good deal of work has already been accomplished. Nursing has provided leadership among healthcare professions in the promotion of genetic and genomic education. Nursing has committed to developing genetic and genomic competency within its own profession and has achieved consensus around what nurses need to know. Nursing has been instrumental in providing a rich collection of resources to support genetic and genomic education. However, the integration of genetic and genomic content into nursing curricula has been patchy, and much work remains to be done to ensure graduate nurses are prepared to practice in a world of genomic healthcare.

Developing plans to achieve any task should begin with assessment, and that principle provides the basis for this dissertation. Virtually nothing is known about the genetic and genomic
literacy of students entering baccalaureate nursing programs. Furthermore, no ruler exists to measure that knowledge. This dissertation applies cognitive theory, an educational assessment model, and a nursing conceptual framework to guide the creation of such a ruler. A genomic concept inventory for nursing will provide a useful tool to assess essential genetic and genomic knowledge of entering students. Application of the tool will produce data to inform course and curricular development and gauge educational effectiveness.
CHAPTER 3
METHODOLOGY

This chapter describes the methodology for the development of the genomic nursing concept inventory (GNCI). The process was synthesized from approaches used to create other concept inventories (Anderson, Fisher, & Norman, 2002; Bowling et al., 2008; Garvin-Doxas & Klymkowsky, 2007) and generally followed the method of Treagust (1988). The resulting approach comprised four sequential steps, each employing a different methodology (Table 1). This study was granted exempt status by the Washington State University Institutional Review Board (Appendix A).

Overview of Methodology

First, key concepts were extracted from the current nursing genetic and genomic benchmark (the Essentials) and validated by 104 experts. Secondly, the understanding of those concepts among baccalaureate nursing students was investigated using student-supplied response surveys ($N = 96 - 134$). Inventory items were then written as multiple choice questions, utilizing common misconceptions as distractors. Thirdly, cognitive interviews with 15 first year nursing students informed item revision. Fourth, the survey was pilot tested with 238 nursing students to measure psychometric characteristics. The instrument was then reduced to the most parsimonious scale possible, retaining the most psychometrically robust items while preserving coverage of the intended content domain. The product of these steps is a 31-item, empirically-derived inventory designed to assess understanding of concepts most fundamental to genomic nursing. The GNCI © Ward 2011 represents a preliminary but novel assessment tool ready for further testing and refinement.
Step One: Extraction and Validation of Key Concepts

Development of the GNCI began with identifying the initial inventory content domain. Although establishing the content domain began during Step One, the process was iterative and spanned all steps of inventory development as the domain was repeatedly focused, refined, and reorganized. To begin, the Essentials were deconstructed to identify embedded concepts. Based on consensus of 104 experts, that list of concepts was prioritized according to relevance to nursing practice at the baccalaureate level and reduced to include the most important concepts. The product of Step One was a list of 21 key concepts comprising the initial content domain for the GNCI.

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<tr>
<th>Step</th>
<th>Description</th>
<th>Method</th>
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<tr>
<td>1</td>
<td>Extraction and validation of key concepts</td>
<td>1. Extract supporting concepts from Essentials.</td>
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<td>2. Validate relevance to nursing with 104 experts.</td>
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<td>3. Rank order concepts.</td>
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<td>4. Select cutoff to establish 21 key concepts.</td>
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<td>2</td>
<td>Exploration of student understanding and inventory drafting</td>
<td>1. Write open-ended questions to explore understanding of each key concept.</td>
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<td>2. Administer questions to beginning nursing students (N = 96-134) using Student Supplied Response (SSR) surveys.</td>
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<td>3. Apply content analysis to SSR data to identify level of understanding and common misconceptions.</td>
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<td>4. Write multiple choice questions to assess understanding of each key concept, utilizing misconceptions as item distractors, and assemble into draft inventory.</td>
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<td>3</td>
<td>Pretesting and inventory refinement</td>
<td>1. Administer draft inventory to 15 first-year nursing students using cognitive think-aloud interviews to check clarity of items, readability, and student reasoning.</td>
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<td>2. Refine inventory based on interview findings.</td>
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<td>4</td>
<td>Pilot testing and inventory revision</td>
<td>1. Administer pilot inventory to 238 nursing students utilizing Scantron methodology.</td>
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<td>2. Analyze student demographic information and item and survey psychometrics.</td>
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<td>3. Reduce inventory to beta version, retaining most robust items to cover key concepts.</td>
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Table 1
Steps in Inventory Development
Nunnally and Bernstein (1994) allege once a test domain and test format are agreed upon, “arriving at an acceptable instrument is mainly a matter of technical skill and application” (p. 296). A test domain includes both the content and the population to be tested. For the GNCI, the target population (baccalaureate nursing students) and the test format (concept inventory) were unambiguous. However, the concepts most foundational to genomic literacy for professional nurses needed to be delimited. While the Essentials provide a detailed benchmark, they were written as competencies for which a set of foundational concepts had not been clearly explicated. Furthermore, the breadth of knowledge subsumed by the Essentials exceeds the scope of a concept inventory. Therefore, the goal was to identify genetic and genomic concepts that are (a) included in the essential competencies, and (b) most salient to nursing practice. During Step One, a group of experts ranked 85 concepts identified in the Essentials according to salience to nursing practice. Twenty-one key concepts were identified and represented the initial content domain for the GNCI.

**Extraction of Concepts**

In the second edition of the Essentials, the Consensus Panel (2009) identified specific areas of knowledge along with clinical performance or practice indicators for each competency. Both the knowledge areas and the clinical performance indicators were deconstructed to identify supporting genetic and genomic concepts, i.e., basic foundational knowledge required to achieve each competency.

Wide variance is evident in the knowledge necessary to achieve various competencies. Certain professional responsibility competencies require little understanding specific to genetics or genomics, while various competencies in the professional practice domain necessitate understanding of multiple concepts. For example, fairly broad knowledge about how genotype is
translated into phenotype is necessary to achieve the competency of understanding “the relationship of genetics and genomics to health . . . .” (Consensus Panel, 2009, p. 11). Certain concepts, such as the use of family history, were embedded in more than one competency. In all, 85 concepts were extracted from the Essentials. To reduce the magnitude of the list, those concepts were reviewed by the investigator to identify concepts (a) most foundational to competency achievement; (b) supporting multiple competencies; and (c) reflecting basic conceptual, rather than task-oriented “how-to” knowledge. Concepts failing to meet those criteria were omitted, reducing the list to 65, an ambitious but more reasonable number of concepts to move forward to the validation step. The 65 concepts were grouped into 14 topical categories (Appendix B) for inclusion in the expert survey.

Validation of Concepts

An online survey was then designed to elicit expert opinion about the importance of each concept to nursing practice. The survey was developed by the investigator in collaboration with staff from Washington State University and Maplewood Software and administered using a Maplewood Software product, Survey Assistant™ (see Appendix C for survey). The survey opened with an informed consent statement, the first question requested acknowledgement of that statement, and clicking “yes” designated consent to participate. Two demographic questions explored professional roles of the experts and length of time in genetic nursing. The rest of the survey consisted of the concept list and a four-point Likert scale, so respondents could rate each concept as critical to know, important to know, nice to know, or not important. Respondents could also mark no opinion. The survey displayed concepts from each topical category on a single page. Answers were forced, so that the survey could not be advanced if a question was unanswered. Free text areas at the end of each page solicited comments or
suggestions about additional important concepts in that category. The survey was pretested for troubleshooting purposes with a convenience sample of ten individuals without specific expertise in genetics.

Next, genetic nurse experts were identified and recruited to complete the survey. Three groups of nurses were recruited: nurses who served on the steering committee for the Essentials Consensus Panel \( (N = 17) \); nurses in education, research or practice in the United States who held membership in the International Society of Nurses in Genetics (ISONG) \( (N = 277) \); and nurse educators in the National Human Genome Research Institute Community of Genetic Educators \( (N = 23) \). Membership in these three groups overlapped broadly, and the final expert list numbered 289. E-mail addresses for ISONG members were provided by that organization as a Microsoft Excel file according to their listserv sharing policy, and e-mail addresses of the other experts were known by the investigator. Each expert was assigned a numeric identifier in order to track responses.

Invitations to participate (Appendix D) were sent by e-mail to the 289 experts. Invitations stated the purpose and sponsorship of the study and included an implied consent statement, instructions for survey completion, and a link to the survey. An attachment (Appendix E) provided more details about the survey. Eight invitations were undeliverable, resulting in 281 contacts. Issues with email blocking programs were addressed individually, and in one case the survey was faxed to a participant. After eleven days, a follow-up email invitation (Appendix F) was sent to 234 non-respondents. The survey was closed four weeks after it was launched with a final sample of 104.

Survey results were stored on a database server housed at Maplewood Software, Inc., in Spokane, Washington, utilizing an encryption process managed via a Secure Sockets Layer
(SSL) certificate on the SurveyAssistant™ website. Data were exported from the database server into a Microsoft Excel file and transferred electronically to the investigator. The Excel file was imported into SPSS version 17.0 for analysis. Survey results included categorical responses to the demographic questions and Likert-type concept rankings, and textual comments.

Demographic data were simply tallied for descriptive purposes. Textual data from comments or suggestions were collated and reviewed to identify common themes to guide inventory development. For each of the 65 concepts, responses were converted to numerical values as follows: critical to know = 4, important to know = 3, nice to know = 2, not important = 1, and no opinion = 0. The mean value was calculated for each concept, and the concept list was rank-ordered according to those means. The concept list was then reduced. The goal was to retain crucial concepts, as identified by the expert group, while maintaining a fairly broad content domain. Therefore, concepts were examined in the context of their topical categories. Within each category, concepts falling below natural breakpoints in relevance scores were omitted, and the mean relevance of the remaining concepts was calculated. The remaining concepts were then resorted according to content, in some instances combining concepts. The process was repeated until an optimal set of 21 concepts had been identified, organized into five topical categories, and labeled. Those key concepts comprised the content domain for Step Two.

Step Two: Exploration of Student Understanding and Inventory Drafting

In the second step in inventory development, student understanding of the key concepts identified in Step One was explored. The purpose was to identify (a) concepts poorly understood by a significant number of students, and (b) misconceptions held by students about those concepts. Inventory items were then written for each poorly-understood concept, using the most common misconceptions as item distractors.
Student Supplied Response Surveys

Student-supplied response (SSR) surveys were utilized to learn what students knew about each of the 21 key concepts. Following the method of Bailey (2008), one or more open-ended questions were written by the investigator to explore understanding of each concept. The questions were brief and stated in simple language. As recommended by Conrad and Blair (1996), author intent was explicitly stated for each question and served to tie the question to the concept of interest. Author intent is a clear statement identifying the purpose of an item as conceived by the survey designer. As an example, one SSR question was: *To the best of your knowledge, what is the function of a gene?* The author intent for that question was to identify whether a student could correctly identify gene function to be the encoding of one or more gene products – in particular, proteins.

The SSR surveys were conducted in classrooms of first-year students in a baccalaureate nursing program at Washington State University (WSU). Convenience sampling was utilized. The goal was to collect at least 100 textual responses to open-ended questions about each key concept. First-semester students in a nursing research class participated in ten short survey sessions. Each session consisted of four or five questions and lasted eight to ten minutes. In addition, second-semester students participated in two similar survey sessions. All classes included students at three sites. Approximately 90 students were in a Spokane classroom with the investigator, and two groups of approximately 20 students each were located on WSU distant campuses in Richland and Yakima, Washington. All sites were connected via live video streaming, and an instructor was present in each classroom. Altogether, students provided responses to 50 open-ended questions.
Surveys were conducted over twelve weeks during regularly scheduled classes. At the beginning of each session, an informed consent statement was read aloud and displayed on classroom video monitors. Voluntary consent was implied by participation. Students were provided with blank index cards. Each question was read aloud to the class, repeated once, and displayed on classroom monitors. Students were asked to mark each card with a number identifying the question and provide a short (one or two sentence) written response. No more than two minutes were allowed for each response; when most students finished writing, the next question was introduced. Students were asked not to write their name or any other identifying information on the cards.

To the extent possible, questions were posed to students prior to the presentation of related content in their nursing classes. For example, questions related to cancer genetics were asked before that content was covered in class. However, it was assumed that misconceptions would be uncovered whether or not students had studied a particular content area. In a seminal study, Hake (1998) administered the Force Concept Inventory to over 6,000 physics students and found students who received content-related class lectures acquired understanding of about 25% of the concepts listed in the inventory. The persistence of misconceptions was also demonstrated by Wage et al. (2005), who administered the Signals and Systems Concept Inventory to engineering students at the beginning and end of a traditional lecture course. Normalized gain analysis indicated students acquired understanding of 20% of concepts they did not comprehend at the beginning of the course.

Analysis of Student Supplied Response Data

Responses from the SSR survey were analyzed using content analysis, applying the method of Krippendorff (1980) and a procedure described by Garvin-Doxas and Klymkowsky.
The goal was to generate a picture of student understanding for each of the 50 questions about the key concepts.

Content analysis is a method of objective, systematic and quantitative description of manifest and latent content of communication (Graneheim & Lundman, 2004). Content analysis enables a researcher to systematically and efficiently identify trends or patterns in a large volume of data (Stemler, 2001). For this study, manifest, i.e., visible or obvious content, was of predominant interest, and responses were explored for conceptual understanding rather than underlying or latent meaning.

The SSR cards were not transcribed. The collection of cards from each of the 50 questions represented a unit of analysis, and each set was analyzed separately. The goal was to identify the level of student understanding of each concept and the most common misconceptions. First, each stack of cards was read three or more times to “obtain a sense of the whole” (Gebru & Willman, 2010), i.e., to gain a sense of the range and patterns of conceptual understanding. This method represents emergent coding, as the codes were not predetermined but emerged from the data (Stemler, 2001).

As each stack of cards was reviewed, a set of textual units emerged. Textual units are groups of words with similar meaning. Each textual unit described a student perception about the concept of interest. Some textual units reflected accurate understanding, and others revealed misconceptions, defined as conceptions incongruent with prevailing scientific explanation. The investigator’s expert knowledge of genetics and genomics provided the basis for identifying misconceptions.

Labels were applied to the textual units. For example, no misconception was the label assigned to responses reflecting an accurate understanding of the concept. Various descriptive
labels were assigned to responses indicating different misconceptions. Blank cards and other responses indicating insufficient knowledge to answer a question were labeled don’t know, and nonsensical or uninterpretable responses were coded unable to interpret.

Each set of cards was sorted according to the textual units embedded in each response. Responses revealing more than one conception were assigned accordingly to multiple textual units. A fairly coarse degree of coding was sufficient to identify the most common misconceptions, and responses revealing unique perceptions were often coded together in a miscellaneous category. Throughout analysis, constant comparison ensured that each textual unit was aligned with a single perception and that textual units were mutually exclusive.

Response cards were analyzed as soon as they were received. This proved useful when unanticipated questions arose about student understanding. For example, a question about autosomal dominant inheritance revealed considerable misunderstanding, leading the investigator to wonder whether the source of confusion was the meaning of autosomal or dominance. The next week, responses to separate questions clarified the picture: most students correctly described dominance, but only 12% could define autosome.

All cards were coded by the investigator. In addition, an undergraduate nursing student whose honors thesis involves genomics and nursing education was instructed in content analysis and independently sorted cards for five SSR questions (10% of SSR data). Coding by the investigator and the student were compared. Variations in coding were reviewed and resolved to reach consensus on the level of understanding and the most prevalent misconceptions for each of the five SSR questions. This step followed the recommendation of Krippendorff (2004).

The output of SSR content analysis was a codebook of student perceptions uncovered during the SSR surveys. The codebook listed each SSR question, the related concept, author
intent, and the number and percent of student responses assigned to each textual unit. The codebook provided a convenient aggregation of data to support inventory drafting.

**Item Development and Inventory Drafting**

Item development began with examination of data in the SSR codebook to identify the level of student understanding of each key concept as well as common misconceptions. For concepts revealing a significant level of misunderstanding, one or two multiple choice questions were composed, utilizing the most common misconceptions as item distractors. No inventory questions were written about the SSR questions which either appeared to be well understood or for which responses failed to reveal specific misconceptions. The goal was to develop an inventory of 50 to 60 items covering the content domain identified in Step One. This number was derived from Nunnally’s (1978) recommendation that an initial item pool contain 1.5 to 2 times as many items as the final instrument (which, for the GNCI, was 25 to 30 items). The resulting draft inventory was then mapped to the content domain to ensure all concepts were represented.

During item writing, recommendations found in the item-writing literature were applied (Frey, Petersen, Edwards, Pedrotti, & Peyton, 2005; Haladyna, Downing, & Rodriguez, 2002; Hufnagle, 2000; Martin, Mitchell, & Newell, 2003; Nunnally & Bernstein, 1994; Taylor & Smith, 2009), as were recommendations specific to concept inventory development (Richardson, 2004). Each item was designed to measure knowledge of a single concept. Items were written in simple language to maximize the ability of the instrument to detect conceptual understanding rather than test-taking skills. Item distractors reflected common misconceptions and were stated in language similar to that used by students in the SSR responses. When possible, questions were written to allow formulation of an answer without reading the response options. Items with direct clinical application were attempted to be placed within a nursing framework.
Step Three: Pretesting and Inventory Refinement

In the third step of inventory development, the draft inventory was pretested with 15 first-year nursing students using individual cognitive think-aloud interviews. The purpose of the interviews was to troubleshoot and improve inventory items prior to pilot testing. An iterative process was utilized, making revisions as problems were identified and retesting items in subsequent interviews. New items were added when gaps in content coverage were identified. The output of this process was a 52-item inventory ready for pilot testing.

_Cognitive Interviewing Methodology_

Cognitive interviews involve “the administration of draft survey questions while collecting additional verbal information about the survey responses” (Beatty & Willis, 2007, p. 287). Sound interpretation of aggregate survey data requires consistent understanding of the meaning of survey items between the survey designer and survey respondents, as well as across respondents (Conrad & Blair, 1996; Willis, Royston, & Bercini, 1991). Survey designers use cognitive interviews to identify variance in meaning by exploring clarity of items, readability, and respondent reasoning (Drennan, 2003). Omission of important aspects of phenomena being examined may also be identified (Desimone & LeFloc’h, 2004). Interview findings inform revision of survey items to enhance instrument reliability and validity (Knafl et al., 2007).

Cognitive interviews most often employ a “think aloud” process: Participants verbally report their mental activity as they complete a draft survey, while the interviewer collects information about how responses are formulated (Conrad & Blair, 1996). Cognitive interviews may occur concurrently with survey completion or retrospectively after the survey is completed (Beatty & Willis, 2007; Drennan, 2003). In a pure think aloud interview, the interviewer assumes the role of an unobtrusive observer, speaking only when necessary to remind the respondent to
keep talking. Alternatively, the interviewer may ask additional direct questions (called probes) to elicit information (Beatty & Willis). The use of probes varies: They may be scripted prior to the interview or created extemporaneously during the course of the interview (Conrad, Blair, & Tracy, 1999).

Cognitive interviews are used extensively in the development of surveys and questionnaires collecting autobiographical data such as information about behavioral frequency. Responding to such a question typically requires recall and sometimes manipulation or calculation of information from autobiographical memory (Willis, Royston, & Bercini, 1991). The use of cognitive interviews in the development of tests of knowledge, which require a different sort of cognitive task, is less well described in the literature. However, cognitive interviewing provides a close view of how respondents interpret questions, and developers of other concept inventories have used various permutations of cognitive interviewing to evaluate and improve items (Bowling et al., 2008; Garvin-Doxas & Klymkowsky, 2007; Nelson et al., 2007; Smith, Wood, & Knight, 2008).

Cognitive interviewing methodology has been criticized for subjectivity and inadequate guidelines for analyzing, interpreting, and using data generated during the interviews (Drennan, 2003; Knafl et al., 2007). In order to objectify and systematize data collection and analysis, this study utilized a method of interview coding and analysis described by Conrad and Blair (1996). Their method is based on a response model generally accepted in cognitive psychology and is summarized below.

Response Theory and the Respondent Problem Matrix

Conrad and Blair (1996) describe a survey question as a set of instructions given to a respondent about a task they are asked to perform. In general, response theory suggests a four-
stage response process. However, Conrad and Blair suggest verbal report methods lack the sensitivity to distinguish four steps. They propose instead a three-stage model, in which responses are formulated in three, usually sequential stages: understanding, task performance and response formatting. A respondent may encounter difficulty during any stage. The interview coder identifies the stage of the response process in which the problem occurred and then categorizes the nature of the problem. This process of staging and categorizing provides a context to understand the problem and promotes the development of promising solutions during survey revision. Conrad and Blair developed a matrix to illustrate response stages and problems and support the coding process (Figure 2).

<table>
<thead>
<tr>
<th>RESPONSE STAGE</th>
<th>Understanding</th>
<th>Task Performance</th>
<th>Response Formatting</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lexical</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Temporal</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Logical</td>
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<td></td>
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</tr>
<tr>
<td>Computational</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Omission/Inclusion</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Figure 2
Respondent Problem Matrix
(Conrad & Blair, 1996)

The first response stage is the understanding stage, when the respondent determines what information is requested and derives an approach to provide that information. The understanding stage requires both a literal interpretation of the question and a determination about the instructions subsumed in the question. Once the respondent has interpreted the instructions and conceived of an approach to develop an answer, he or she is ready to execute the second
response stage, which is to perform the primary task. The task involves executing the mental operation required to produce the data on which the response will be based. The last response stage entails formatting or mapping the response to the options included in the questionnaire. Although respondents typically move sequentially through these steps, the steps may overlap, or a respondent who encounters difficulty with a step may revisit a previous step (Conrad & Blair, 1996).

Consider, for example, how a student might approach the following question: What is the relationship between genes and chromosomes? The first response stage requires the student to interpret the question and understand the instructions. Difficulty may occur with understanding whether the question addresses a particular gene and its corresponding chromosome, or genes and chromosomes in general. The second response phase requires the student to perform a mental process (such as comparison, deduction or evaluation) and formulate an answer. Here, difficulty could occur if the respondent identifies more than one relationship between genes and chromosomes, for example both a physical and a functional relationship. In a knowledge-based inventory such as the GNCI, difficulty with task performance may stem from a knowledge deficit. In that case, there may be nothing for the inventory developer to fix, since the purpose of the scale is to assess knowledge. The third response stage requires mapping of the response to the options provided. Difficulty during this stage could occur if a respondent has a specific answer in mind, but that answer does not correspond to any of the provided answers.

Five types of response problems, identified by Conrad and Blair (1996) and further described by Drennan (2003), claim to characterize most problems encountered by respondents during think aloud protocols. Lexical problems stem from difficulty understanding the meaning or context of language in the questionnaire. Temporal problems occur when respondents have
difficulty answering questions about time frames, such as time spent doing an activity. Logical problems often occur when connecting words, connecting concepts, or presuppositions occur in questions. Logical problems can also stem from information that is shared by different questions in the interview, a distinct possibility with the GNCI. The computational category is designated for problems related to mental calculations. Computational problems can compound problems in any of the aforementioned categories, or they can occur independently. However, the computational category also serves as a residual category for problems that do not fall into another category. Conrad and Blair recommend that coders assign problems to this category after all other categories have been considered. Omission/inclusion problems occur when the scope of language is confusing to a respondent. An example might be when a question refers to gene function, and a respondent’s answer depends on whether they consider the function of one gene or multiple genes.

Conducting the GNCI Cognitive Interviews

For this study, concurrent cognitive interviewing was conducted by the investigator. A mixed approach was utilized. Students were asked to read each question aloud and verbalize their thoughts as they contemplated and answered each question. In addition, the investigator utilized verbal probes to clarify student thought processes. Because thinking aloud adds to the burden of questionnaire completion and slows down the process (Conrad, Blair, & Tracy, 1999), interviews were anticipated to take 60 to 90 minutes for the 55-item draft GNCI.

Junior baccalaureate nursing students in their second semester of nursing school were invited to participate in the cognitive interviews. The invitation was extended during a regular class session of medical-surgical nursing. To meet inclusion criteria, students were required not to have progressed beyond the first year of nursing school, and to be available for a 90-minute
interview at the home campus during a designated three-week period. These students were likely to have participated in one or two SSR sessions earlier in the semester, but they were not the students who completed the bulk of the SSR sessions. Cash awards of $30 were offered as incentive for students to participate and provide thoughtful responses.

Students who expressed an interest in participating were sent further information by email and invited to schedule an interview at their convenience during the designated time frame, which was near the end of the semester. Several students chose to complete their interview in the days following final exams. As students scheduled their interviews, they were sent informed consent forms to review (Appendix G). Ultimately, the sample comprised the first 15 students to schedule and complete an interview.

All interviews were conducted by the investigator in an interview room in the research suite at the WSU College of Nursing. Before each interview began, the investigator reviewed details of the informed consent and answered any questions. When the consent form had been signed, participants were given a paper copy of the draft inventory, a pencil, and a blank paper.

Verbal instructions were provided. Participants were asked to complete the inventory, marking each answer on the paper test and verbalizing their thoughts as they considered each question. Students were asked to cover the responses to each question, so that they could only see the stem, and then read each stem aloud. They were encouraged to state whether the question seemed clear and then “think aloud” as they formulated an answer. Participants were asked to articulate an answer before uncovering the responses, when possible, and to continue verbalizing their thoughts as they selected a response. Participants were reminded that the purpose of the interview was not to measure their knowledge but to troubleshoot and improve inventory items. Consequently, responses were not forced. When students had difficulty selecting a response, they
were allowed to guess, “rule out” one or more responses, or simply decline to answer. In that event, however, students were often asked if the responses were understandable and seemed distinct. During the interviews, other probing questions were asked by the investigator to clarify student reasoning. The interviews were audio recorded. Recordings were labeled only with the date and time of recording and a numeric identifier to match the paper interview forms.

During the interviews, the investigator wrote short textual notes directly onto a coding form. The form was printed with each inventory item, a brief statement of author intent, Conrad and Blair’s (1996) respondent problem matrix, and space for notes. The author intent served as a reminder about which key concept each question was designed to explore.

Notes were written when a student expressed difficulty understanding a question, formulating an answer, or mapping their answer to the provided responses. In addition, the interviewer marked the matrix to indicate the nature of a student’s difficulty with any item. For example, when a student struggled with the language in the item stem, the lexical cell in the understanding column was marked and the nature of the problem annotated in a note. Similarly, if a student articulated a correct response to a question but was unable to find that response among the options, the appropriate cell in the response formatting column was marked and a note written. Responses that did not fit into one of the predefined categories were coded as computational problems and described with an anecdotal note. Occasionally a student would offer a suggestion for improving the item; those suggestions were noted and sometimes implemented but not necessarily coded as problems. When a response was particularly insightful or revealing or warranted further consideration, it was flagged for later review.

Each interview was coded as it occurred. Both copies of the survey (i.e., that completed by the student and the annotated copy used by the interviewer) were identified only with a
numeric identifier for later reference. At the end of the interview, each student was provided an opportunity to discuss any of the inventory items and was given a $30 cash award. When all interviews had been completed, the paper forms and the audio recordings represented collective data about each survey item.

Analysis of Cognitive Interview Data

Data analysis occurred following each interview or occasionally after a pair of consecutive interviews. The matrix and anecdotal notes for each inventory item were reviewed to identify and correct, if possible, the source of difficulties encountered by students. In addition, the audiorecordings were frequently reviewed during data analysis.

Throughout data analysis, inventory items were revised on a rolling basis and retested in their new format with the next interview. The process was therefore iterative, with some items being revised several times. On occasion, consecutive interviews utilized the same draft inventory, but in general each inventory reflected cumulative changes.

When data analysis revealed gaps in coverage of key concepts, SSR data was reviewed and new items were developed and added to the draft inventory. Such recognition of significant omissions has been described by others (Desimone & LeFloh, 2004). At the time of the last interview, the inventory contained 60 items.

When all the interviews had been completed and analyzed, collective data regarding each inventory item were reviewed. The method for this step was adapted from Conrad and Blair (1996) and Knafl et al. (2007). Each item comprised a unit of analysis. Pooled interview data about each item was merged with data from the codebook created during Step Two. At this point, the codebook contained comprehensive data about each survey item, including the question in its current form, the concept tested by the question, author intent, and notes summarizing collective
problems encountered during the 15 interviews. This codebook reflected the relationship of each item to the content domain and displayed data in a convenient format to support critical analysis of each inventory item.

**Inventory Refinement**

Utilizing the codebook, all 60 inventory items were reviewed and last revisions made prior to pilot testing. The goal was to test a sufficient number of items for optimal inventory development while not overtaxing students. Because of concern about whether students could reasonably complete 60 items in the hour available for pilot testing, eight items were omitted. Decisions about item omission were based primarily on preservation of the breadth of the content domain. The resulting 52-item pilot version was pretested with four volunteers, who completed it in 40 to 50 minutes. The inventory was then ready for pilot testing in Step Four.

**Step Four: Pilot Testing, Psychometric Analysis, and Final Inventory Revision**

In the fourth step of this research, the 52-item Genomic Nursing Concept Inventory (GNCI) pilot version was tested with 238 baccalaureate nursing students. The psychometric properties of the entire inventory as well as individual items were examined. Based on those data, the inventory was reduced to a parsimonious inventory of the most robust questions that provide the broadest possible coverage of the content domain. The resulting 31-item GNCI beta version is ready for further validation.

**Conducting the Pilot Test**

Pilot testing should be conducted in the same population for which a scale is designed and should utilize a sample of sufficient size to limit the effects of variance in scale scores; a sample size of at least 200 is recommended (Nunnally and Bernstein, 1994). Two cohorts of students at Washington State University College of Nursing provided a convenience sample of
approximately 250 and were invited to participate. These students had not participated in either the SSR surveys or the cognitive interviews. One group had just entered nursing school, beginning the first semester of the junior year; the other group was beginning the final semester of the senior year. The survey was conducted during regularly scheduled classes in two courses: a professional development class taken by students in their first semester of the nursing program, and a community health nursing course taken by students in their last semester. Enrollment was 120 students in the professional development course and 133 students in the community health nursing course. Both classes were conducted at three sites, with approximately 90 students at the main campus in Spokane, Washington, and approximately 20 students at each of two distant sites in Richland and Yakima, Washington. Classrooms were connected via live videostream and a faculty proctor was in attendance at all sites.

Participation was voluntary, however incentives were provided to encourage students to participate, complete all inventory items, and give careful thought to their responses. Incentives were awarded to all students who completed every inventory item. In the professional development class, students who participate in a research project were awarded extra credit points, and this pilot test fit that requirement. In the community health nursing class, students had a choice of completing the survey or participating in a course discussion board. In addition, to encourage students to provide thoughtful answers, $10 cash awards were offered to ten students in each class with the highest scores.

Informed consent was provided to each student group prior to the pilot test by reading a consent statement while displaying information on classroom monitors. Consent was implied by participation. The survey was administered as a paper-and-pencil test with a Scantron form. Students were asked to write their name and student identification number on both forms to
facilitate Scantron scoring and awarding of cash prizes. One hour was allowed to complete the survey, which included three demographic questions and 52 knowledge-based items. Students were asked answer the questions in the order presented. To capture maximal data from any student who ran out of time, students were asked to complete their paper survey prior to filling in their Scantron.

Both the paper surveys and the Scantrons were collected as students completed the inventory. The paper surveys were checked for missing demographic data and students were asked to complete unanswered questions. Any missing data on the Scantron forms was retrieved later from the paper surveys. There were consequently no missing data at the time of data analysis.

Analysis of Pilot Test Data

Pilot testing generated data about the student sample, individual scale items, and psychometric properties of the entire survey instrument. Data about the students were reported without analysis. Psychometric data about individual scale items were examined to identify and revise or eliminate items of poor quality and reduce the total item number to between 25 and 30. Data about the psychometric features of the entire scale were evaluated to assess scale difficulty and internal consistency reliability.

Characteristics of the Student Sample

Three demographic questions elicited information about respondent age, gender, and prior genetics courses. Students wrote responses to those questions on the paper survey, and raw data were entered into SPSS Version 17 to generate descriptive statistics.
Item Analysis

Item analysis was conducted to identify the best item set, defined as the optimal collection of psychometrically robust items to sample the intended content domain (Ferketich, 1991). Item analysis was based on aggregate data from both student groups regarding item difficulty, discrimination, and correlation, as well as response frequency. The first three criteria informed decisions about retaining, revising or deleting individual items, while response frequency was used to make similar decisions about item distractors. Psychometric features of individual items as well as logical arguments about content validity were balanced when making decisions about item retention.

Initial item analysis data were derived from the Scantron reports; however, subsequent calculations were more conveniently made using SPSS. Therefore, student responses from the Scantron report were manually entered as binomial (correct-incorrect) values into SPSS Version 17, recording 1 for correct responses and 0 for incorrect responses to each inventory item. The database included 238 responses for each of 52 items. Forty percent of the data was rechecked for accuracy.

Key psychometric values for each of the 52 items were entered into a table to facilitate item comparison. The Item Analysis Table displayed pilot test data about item difficulty, discrimination, and correlation, and provided a convenient resource to inform decisions about item retention, omission and revision.

Item difficulty. Item difficulty reflects the proportion of students who answer a scale item correctly (Kaplan & Saccuzzo, 1997), with a high value indicating low difficulty. In general, items answered correctly by more than 85% of respondents are considered too easy, items answered correctly by less than 25% of respondents are considered too difficult, and both types
of questions limit scale validity (Statistical Analysis of Multiple Choice Exams, n. d.) Within those parameters, including items of varying difficulty improves a scale’s ability to discriminate between students with varying levels of the measured attribute (Nunnally & Bernstein, 1994). Kaplan & Saccuzzo suggest optimal item difficulty lies halfway between 100% and the chance a student could choose the right answer by guessing. Applying their recommendation to the GNCI, which includes items with three, four, and five response options, optimal difficulty would fall in the range of 60% to 66%. However, concept inventories focus on difficult concepts and typically exhibit lower difficulty indices (indicating greater difficulty) than traditional achievement tests (Nelson et al., 2007).

Item difficulty was reported as a percentage on the Scantron report and entered as a p value in the Item Analysis Table. Items with values below .25 or above .85 were flagged for consideration of revision or deletion. Throughout inventory reduction, an effort was made to retain items of varying difficulty while considering the overall scale difficulty.

**Item discrimination.** Nunnally and Bernstein (1994) recommend applying item discrimination as the primary criterion during item analysis. The simplest measures of item discrimination compare an item score to the total scale score. In this study, item discrimination was reported variably by the two software programs. The Scantron software reported item discrimination as a point-biserial value ($r_{pb}$), a correlation between each item score as a binary variable (i.e., correct or incorrect) and the total score. The Scantron value was uncorrected, i.e., the item in question was not removed from the total when the correlation was calculated. In SPSS, item discrimination was calculated as a corrected item-total correlation, removing each item from the total. Therefore, the values for each item were not identical. However, ranking items by discriminatory power corresponded closely, irrespective of which value was used.
Like all correlations, point-biserial and item-total values range from -1 to +1; the larger the value, the more discriminating the question. Very easy and very difficult items have poor discrimination. Generally, a value below +.2 indicates poor discriminatory power, a value between +.2 and +.4 reflects fair discrimination, and a value above +.4 indicates optimal discrimination. A minimum acceptable value of +.3 is suggested (McGahee & Ball, 2009; Nunnally & Bernstein, 1994). However, the acceptable level of item-total correlation depends on the number of scale items and scale homogeneity. Knapp and Brown (1995) caution “no magic cutting point” exists for item-total correlation (p. 467); Crocker & Algina (1986) suggest an item correlating less than +.3 may be considered for retention if the value exceeds two standard deviations above zero; and Kehoe (1995) suggests restructuring any item that correlates less than +.15 with the total test score.

Point-biserial values from the Scantron report and corrected item-total correlations calculated in SPSS were entered into the Item Analysis Table and considered when evaluating item discrimination. Although the goal was to retain items with values greater than +.3, initially items with values less than +.2 were flagged for review. Discriminatory power was assigned significant weight when making decisions about retaining or omitting items.

Following the first step of inventory reduction, corrected item-total correlation provided the only measure of item discrimination, because Scantron data was not recalculated. Each time a set of items was omitted from the inventory, corrected item-total correlation values were recalculated using SPSS to measure the correlation between each retained item and the new total item set. Those values were used to evaluate and compare the discriminatory power of retained items.
An ideal test question has a high positive \( r_{pb} \) value for the correct answer and a negative \( r_{pb} \) value for each of the distractors (Martin et al., 2003). The Scantron software calculated the \( r_{pb} \) for all item responses. Therefore, during response frequency analysis, the set of \( r_{pb} \) values for all response options was reviewed. Negatively-correlated distractors were reviewed for omission, revision, or replacement.

**Inter-item correlation.** If the GNCI represents a unitary central factor defined as genomic literacy, the frequency of correct responses on one item should correlate positively and significantly with the frequency of correct responses on other items. Inter-item correlation indicates how well scale items correlate to each other and directly relates to both item-total correlation and the coefficient alpha for the entire scale (Ferketich, 1991). In general, items displaying inter-item correlations below +.3 are considered insufficiently related to the hypothetical core factor, while items that correlate above +.7 may be redundant (Ferketich). However, inter-item correlation varies with breadth of the content domain and the number of items, and the same parameter cannot reasonably be applied to all scales (Knapp & Brown, 1995). Boyle (1991) suggests an inventory capable of measuring knowledge across a broad content domain requires a degree of item diversity that limits item intercorrelation, and Nunnally and Bernstein (1994) advise content-validated tests do not require high internal consistency. Finally, as Ferketich pointed out, a test comprising ten or more items with an average inter-item correlation as low as +.2 may achieve an acceptable alpha of .71.

The broad content domain and variable item difficulty of the GNCI limit achievable inter-item correlation, and limiting either breadth of content or variability in item difficulty to achieve higher measures of correlations would not be advantageous. Therefore, inter-item
correlation was considered during item analysis but assigned less weight than item difficulty or discrimination.

The 52 by 52 item correlation matrix generated in SPSS displayed correlations between all item pairs. When a scale contains so many items, the correlation matrix is large and unwieldy, and the variance among inter-item correlations may be limited (Robinson et al., 1991). The matrix was reviewed, and item pairs that correlated in the preliminary target range of +.3 to +.7 were flagged for review during item analysis. In addition, inter-item correlations were imported into Microsoft Excel to calculate the average corrected inter-item correlation coefficient. This value was calculated for each item, omitting the item’s correlation with itself. As predicted, the values showed little variance, making them difficult to compare. Consequently, the 52 items were ranked according to average corrected inter-item correlation, and the rankings were added to the Item Analysis Table.

The last piece of information about item correlation was the estimate of Cronbach’s alpha if any item was deleted from the scale. Values were calculated in SPSS and, for ease of interpretation, converted to reflect the effect on alpha if any item were deleted. The converted values were entered into the Item Analysis Table to support decision making. The completed table provided the basis for inventory reduction.

Inventory Analysis

Data reflecting attributes of the entire inventory were also analyzed to investigate the psychometric qualities of the GNCI. Included were descriptive statistics about student scores on the scale and indicators of scale difficulty, reliability and validity.

Descriptive statistics. Descriptive statistics about total GNCI score were calculated to explore group differences according to age, gender, campus, program progression, and previous
genetics course. Data included range of scores, measures of central tendency, and standard deviation.

*Scale difficulty.* As previously discussed, optimal difficulty for multiple-choice questions with three to five response options is approximately 65%. However, concept inventories logically exhibit a greater level of difficulty because they are designed to measure difficult concepts. Increased difficulty also affords inventory versatility in evaluating students with varying levels of knowledge and supports the use of the scale as a pre/post-test. Review of 13 CIs across disciplines revealed mean pretest difficulty of 43%, and mean pretest difficulty for three genetic concept inventories were 41% (Smith, Wood, & Knight, 2008), 43% (Bowling et al., 2008), and 50% (Elrod, 2007). Target difficulty for the GNCI was initially set at 40 to 55%.

*Scale reliability.* Cronbachs alpha represents the most common measure of scale reliability, and Nunnally & Bernstein (1994) recommend a modest alpha of .70 as a reasonable goal for a new scale. However, the value of alpha varies with the number of scale items and the degree of internal consistency, so relying on a fixed standard of reliability is implausible.

Cronbachs alpha was not reported by the Scantron software but was included in scale reliability analysis using SPSS. The Cronbachs alpha was calculated for the 52-item pilot test and recalculated each time a set of items was omitted. Other things held equal, reducing the item number should decrease alpha (Ferketich, 1991); however, eliminating items with poor psychometric characteristics can mitigate or overcome this effect.

The Kuder-Richardson formula is the measure of internal consistency most often applied to multiple choice examinations. The K-R 20 value represents a special application of the alpha coefficient to tests with dichotomous (here, correct-incorrect) items and is calculated by the Scantron software. Values range between 0.0 and 1.0, with a higher number indicating greater
internal consistency. Parameters for K-R 20 vary according to the context, purpose, and length of an exam, but in general values above .5 or .6 represent good reliability (McGahee & Ball, 2009; Test and Item Analysis, n.d.). The K-R 20 value parallels that of coefficient alpha, and although K-R 20 was reported for the 52-item pilot test, only the Cronbachs alpha was recalculated during inventory reduction.

Additionally, a rough estimate of test reliability was derived by comparing the range of scores to the standard deviation. In general, a higher ratio indicates a test that is better able to discriminate between students of different levels of ability. For a group of 100 students, a score range broad enough to include five standard deviations would be optimal; for 500 students the range of scores would optimally include 6 standard deviations (Statistical Analysis of Multiple Choice Exams, n.d.). For the GNCI pilot test, therefore, a range of scores containing five to six standard deviations would provide an additional indication of inventory reliability.

Scale validity. The validity of the GNCI reflects the scale’s ability to measure a construct (genomic literacy) in a particular target audience (baccalaureate nursing students). Content validity rests primarily on rational grounds rather than statistical measures (Nunnally & Bernstein, 1994). Efforts to strengthen content validity permeated all steps of inventory development. For example, the initial content domain was drawn directly from the nursing Essentials and validated by experts. During the SSR surveys and initial inventory drafting, an explicit statement of author intent tied each item to its supporting concept. Item writing was based on that author intent and student misconceptions uncovered during SSR surveys. Throughout item analysis, decisions to retain, revise or omit items were made by balancing statistical indicators of psychometric rigor against the criticality of an item to the entire content
domain. Other measures of validity, such as concurrent and discriminant validity, represent future steps in the development of the GNCI and will be addressed in Chapter Five.

*Inventory Reduction and Revision*

The Item Analysis Table displayed data to inform decision making during inventory reduction. The primary criteria for item retention were difficulty, reflected as a p value in the table, and discrimination, which was expressed as a point-biserial (r_{pb}) value and a corrected item-total correlation. Cronbachs alpha for the entire scale if any item was dropped was also considered, and ranked average inter-item correlation was considered as a secondary criterion. Individual parameters for item retention were described previously.

In an iterative process, items with poor psychometric features were eliminated, always considering the effect of each item omission on the entire content domain. In the first rounds of inventory reduction, items that fell below parameters for multiple measures were omitted. As the inventory was reduced, scale characteristics were recalculated using SPSS. Recalculation of scale psychometrics was necessary because omitting even one item affects average inter-item correlation and the scale alpha. With each iteration, the Item Analysis Table was revised to reflect retained items and new values. Because the recalculations were generated using SPSS, point-biserial values were not recalculated, and the corrected item-total correlation became the sole measure of item discrimination. Less-robust items were occasionally retained based on rational grounds. At the end of this process, an optimal set of retained items had been identified by balancing psychometric rigor and content domain coverage. Retained items with suboptimal psychometric features had been flagged for revision or replacement during inventory revision.

Following inventory reduction, each retained item was reviewed one final time. The underlying concept, author intent, and the item’s relationship to the whole content domain were
reexamined. At this point, analysis of response frequency data from the Scantron report was conducted on retained items.

Response analysis data indicates how many students selected each response option, as well as the point-biserial value for each response. Response frequency analysis is useful to reveal distractors that are unselected or selected by few students. Unselected distractors add to the burden of test taking but do not contribute to test reliability and should be omitted, revised or replaced (Nunnally & Bernstein, 1994). The data were reviewed and, applying an arbitrary cutpoint, distractors selected by fewer than 3% of students were flagged for consideration of omission or replacement. Because each incorrect response on the GNCI represented a misconception identified in the student population, few unselected distractors were anticipated.

Inventory items should have a positive point-biserial value for the correct answer and negative values for each distractor. Therefore, the point-biserial values were reviewed, and any distractor with a positive point-biserial value was flagged for omission, revision or replacement. As response data were analyzed, data from the SSR surveys and cognitive interviews were occasionally reviewed to inform decisions about rephrasing or removing item distractors.

The resulting item set represented the GNCI beta version. Once again, inventory items were mapped to underlying key concepts and final revisions made to the content domain. The Genomic Nursing Concept Inventory, with 31 items in its beta version, represents the product of this research.
CHAPTER 4
RESULTS

The results of the analysis previously described in Chapter 3 are now discussed. The discussion is organized by the four steps used to develop and pilot test the Genomic Nursing Concept Inventory (GNCI © Ward 2011).

Step One: Extraction and Validation of Key Concepts

The goal of Step One was to identify the most important concepts within the domain of knowledge necessary for nurses to achieve genetic and genomic competency. The methodology of concept extraction and reduction was described in Chapter 3. Eighty-five concepts were extracted from the Essentials, reduced to 65 foundational concepts, and imported into an online survey. Genetic nurse experts were asked to rank those concepts according to relevance to nursing practice.

Response Rate

Of the e-mail invitations sent to 289 genetic nurse experts holding membership in three specialty groups, eight invitations were undeliverable, resulting in 281 distributed invitations. When the survey was closed four weeks later, 104 experts had participated, representing a 37% response rate. The response rate was 35% (6 of 17) for members of the Consensus Panel Steering Committee, 37% (102 of 279) for ISONG members, and 38% (8 of 21) for nurse educators in the NHGRI Community of Genetic Educators.

Demographic Data

Data from the online survey were imported into SPSS version 17 for analysis. The sample of experts reflected considerable longevity in genetic/genomic nursing, with the largest group of respondents having been in the field for longer than ten years and 68% of respondents for longer than five years (See Figure 3).
Most of the 104 respondents reported more than one role in genetic nursing. Fourteen members of the Essentials Consensus Panel participated in the survey; six of those served on the steering committee. Half of the respondents held faculty positions, 40% were advanced practice nurses in clinical practice, and 34% were involved in clinical research. Smaller numbers of respondents were registered nurses in clinical practice (11%) or reported other roles in genetic nursing (17%). Among those roles were study coordinators, managers of genetic programs or clinics, and graduate students in genetics or genomics. Five respondents were involved in laboratory research in addition to their other roles (see Figure 4).

*Data Analysis: Establishing the Initial Content Domain*

Although the survey was designed to force responses, five experts did not complete the entire survey, and the number of responses to each question ranged from 99 to 104 ($M = 102, SD = 1.2$). Missing data were simply excluded from the calculation for each concept. Based on a
four-point Likert scale where four represented “critical to know” and one represented “not important,” mean relevance values were calculated for each concept. Relevance values ranged from 3.90 to 2.34 ($M = 3.39$, $SD = 0.22$). Sorting the concept list according to those mean values resulted in a prioritized concept list (Appendix H). No natural breakpoint in the mean values signaled a clear cut-off point for selecting concepts important enough to retain for further inventory development, however ten concepts with mean relevance values below 3.0 were omitted at this point. An iterative process of comparing the relative rankings of concepts within topical categories, omitting lower-priority concepts, and regrouping remaining concepts eventually revealed a natural breakpoint at a mean of about 3.4. Three entire categories of concepts (proteins, test characteristics, and race/ethnicity) fell below this threshold. Some categories had been reduced to a single concept. Retained concepts were reorganized into topical categories. The resulting concept list (Table 2) included 21 concepts in five topical categories:
human genome basics (3 concepts), mutations (4 concepts), family history (3 concepts), genomic healthcare applications (6 concepts), and genetic testing (5 concepts). These concepts represented the content domain for Step Two of inventory development. Considering the limited capacity of a concept inventory, retention of 21 concepts seemed appropriate at this stage, since some concepts were anticipated to be eliminated during further inventory development.

<table>
<thead>
<tr>
<th>Five Topical Categories</th>
<th>Concept Description (21 concepts)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Human genome basics</td>
<td>Genome structure and organization</td>
</tr>
<tr>
<td></td>
<td>Genome function and information flow</td>
</tr>
<tr>
<td></td>
<td>Genome size and homogeneity</td>
</tr>
<tr>
<td>Mutations</td>
<td>Nature of a mutation</td>
</tr>
<tr>
<td></td>
<td>Germline and somatic mutations</td>
</tr>
<tr>
<td></td>
<td>Penetrance</td>
</tr>
<tr>
<td></td>
<td>Variable expression</td>
</tr>
<tr>
<td>Family history</td>
<td>Role of family health history</td>
</tr>
<tr>
<td></td>
<td>Identifying genetic red flags</td>
</tr>
<tr>
<td></td>
<td>Inheritance patterns and pedigree interpretation</td>
</tr>
<tr>
<td>Genomic healthcare applications</td>
<td>Potential harms in genomic care</td>
</tr>
<tr>
<td></td>
<td>Pharmacogenomics</td>
</tr>
<tr>
<td></td>
<td>Genomic care and lifestyle changes</td>
</tr>
<tr>
<td></td>
<td>Genomics and disease</td>
</tr>
<tr>
<td></td>
<td>Genetic risk assessment</td>
</tr>
<tr>
<td></td>
<td>Cancer genetics</td>
</tr>
<tr>
<td>Genetic testing</td>
<td>Genetic exceptionalism and privacy issues</td>
</tr>
<tr>
<td></td>
<td>Screening tests</td>
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<tr>
<td></td>
<td>Diagnostic tests</td>
</tr>
<tr>
<td></td>
<td>Carrier testing</td>
</tr>
<tr>
<td></td>
<td>Predictive testing</td>
</tr>
</tbody>
</table>

Table 2
Step One: Initial GNCI Content Domain
21 concepts in 5 topical categories

Textual Comments

Review of textual comments on the surveys failed to reveal additional concepts thought by more than a few participants to be important. In several cases, suggested concepts were in fact
included in a different part of the survey. The nurse experts did offer a few common comments.

For example, several experts pondered reasonable expectations of baccalaureate nurses,

*Regarding [genetic] risk assessment: a major issue is the level of expertise needed for practicing nurses. I believe that nurses need to have an understanding of these concepts, but genetic counselors or nurses specializing in genetics should receive the referrals for thorough assessment and explanation of genetic risk.*

and of baccalaureate programs.

*Even as an educator, I worry that the concepts are at too high of a level to facilitate their integration into undergraduate nursing education. This may be particularly difficult at institutions that lack educators with specialized knowledge of genetics.*

Other comments acknowledged that genomic literacy is a moving target, due to uncertainty about whether and when rapidly-developing knowledge and technology will be translated to practice.

*Epigenetics is very hot right now, but I don’t know if it will turn out to be a direction that the science of genetics will take seriously.*

A few respondents reflected on the difficulty in defining a fundamental knowledge domain for all professional nurses, both because knowledge requirements vary with practice area, and because of blurred lines between practice domains of basic and advanced practice nurses. The example most frequently cited by experts was the interpretation of genetic information.

*I struggle with expecting BSN grads [to quantify] risk because if done, then they should be expected to provide the counseling but I am not convinced they have the time to do this effectively while meeting all their current role expectations. Nurses function within a multidisciplinary team for a reason.*

*I do not think the BSN needs to be able to interpret a pedigree but should identify when a person should be referred to a genetics professional.*

The degree to which all professional nurses should be able to interpret a pedigree is therefore unclear. A proposed distinction suggests all nurses should be able to identify a client at increased risk and initiate a referral, while quantifying that risk and developing an appropriate plan of care may be the role of a genetic specialist or advanced practice nurse.
From the textual comments emerged two issues that warrant particular consideration. Those were defining reasonable expectations for the practice domain of baccalaureate-educated nurses, and how to assimilate rapidly-evolving genetic and genomic knowledge into nursing education and practice. Both issues are addressed in Chapter Five.

**Step Two: Exploration of Student Understanding and Inventory Drafting**

The second step of the research process entailed investigating student knowledge about the concepts identified in Step One. As described previously, open-ended questions about each of the 21 concepts were asked of first-year nursing students. Content analysis of their textual responses indicated the level of understanding of each concept as well as prevailing misconceptions. Based on those data, multiple choice questions were written, using the most common misconceptions as item distractors. Those questions were organized into a draft survey.

*Student Responses*

Two classes of first-year nursing students participated in SSR surveys. Enrollment in each class was approximately 130 students, and the number of response cards varied from 96 to 134 ($M = 119$). Class attendance diminished as the semester progressed and was significantly lower for the second-semester class ($M = 101$) compared to the first-semester class ($M = 123$). Altogether, 12 SSR sessions generated nearly 6,000 student response cards.

For 19 of the 50 questions, every student wrote an answer; for the remaining 31 questions, between 1% and 48% ($M = 12\%$, $SD = 11$) of students either left the card blank or reported they did not know the answer.

Analysis of student responses using the card-sorting method (described earlier) revealed widespread knowledge deficit about the key genetic and genomic concepts. Of the 50 questions asked, 40 generated responses specific enough to allow calculation of level of understanding and
identify common misconceptions. Understanding, represented by the percentage of correct responses to each question, ranged from 7% to 97% \((M = .35, SD = .211)\). Responses to 37 questions revealed between one and nine different misconceptions reported by at least 3% of students \((M = 3.84, SD = 2.10)\).

Ten questions were exploratory in nature or had multiple correct responses. For those questions, no calculation of level of understanding was made, however common misconceptions were identified. A few SSR questions provided insight into student beliefs about concepts such as genetic testing, but did not provide data leading to development of an inventory item.

A second coder analyzed data from 10% of the SSR questions. Initially the codes identified by both coders matched for 85% of cards, with both coders identifying the same most common misconceptions for each of the five questions. Variations in coding were discussed and resolved, reaching 100% consensus on the level of understanding and the most prevalent misconceptions for each of the five SSR questions.

**Inventory Drafting**

A draft survey was constructed by writing one or two items for each SSR question that reflected a significant level of misunderstanding and for which student responses suggested plausible distractors. No items were written about the few SSR questions which reflected a high level of student understanding. The resulting draft survey (Appendix I) initially consisted of 55 items, although 5 items were later added during Step Three. The number of items was consistent with Nunnally’s (1978) recommendation for an initial item pool containing 1.5 to 2 times as many items as the final instrument.

An example of the process of item development is provided. One SSR question asked students: *To the best of your knowledge, what is the function of a gene? What does a gene do?*
The author intent for this question was to investigate student knowledge that a gene encodes one or more proteins. All 134 students answered this question. The responses of 57 students (43%) did not reveal any misconception: 33 students (25%) correctly reported that a gene encodes or directs the formation of proteins; 13 students (10%) wrote that a gene stores information (which is not incorrect but rather nonspecific); and 11 students (8%) described reproduction or inheritance as gene functions (which is also not incorrect). The most common misconceptions were as follows: 76 students (57%) wrote that a gene determines a physical trait, 11 (8%) said a gene directs tissue development, 10 (7%) said a gene drives a physiologic function, 5 (4%) said a gene encodes DNA, and 2 (1.5%) said a gene directs cell replication. Three responses described nonspecific gene functions. Because a number of responses included more than one textual unit, the total exceeds 100%. These data led the investigator to write the following inventory item:

The primary function of a gene is to
a. determine a particular trait
b. allow cell division
c. direct the formation of specific protein(s)
d. direct a particular physiologic function
e. regulate tissue development

When the draft inventory had been created, items were mapped to the content domain to ensure representation of all concepts. Three concepts (potential harms in genomic care, genomic care and lifestyle changes, and genetic exceptionalism and privacy issues) were relatively underrepresented in the draft inventory, due to difficulty writing items based on SSR data. However, no changes were made in the 21-concept content domain at this time.

Step 3: Pretesting and Inventory Refinement

The third step in inventory development was to refine the draft inventory by pretesting it with student nurses during cognitive interviews. Problems encountered by students as they
considered each inventory item were identified, and items were revised to improve clarity and readability. In addition, gaps in the content domain were uncovered during the interviews, prompting the creation and testing of new items.

*Interview Sample and Administration*

A convenience sample of 15 first-year nursing students participated in cognitive interviews over a three-week time period. Three male and 12 female students were interviewed. No other demographic data was collected. Two of the students spoke English as a second language. Interview duration ranged from 48 to 96 minutes ($M = 74, SD = 13.3$). One student completed only 52 questions in the available time; all other students completed all 55 - 60 inventory items.

*Cognitive Interview Data*

Coding of the interview data using Conrad and Blair’s (1996) Respondent Problem Matrix revealed 57 problems with items on the draft inventory (see Table 3). Fifteen items were easily navigated by students. Between one and four problems were identified for each of the other items: 27 items had a single identified problem, 10 items had two problems, two items had three problems, and one item had four problems. Most problems were either lexical or logical in nature, and the greatest number of problems occurred during the understanding stage.

Lexical problems were recorded when students did not understand the meaning of words or word combinations in either the item stem or response options. Lexical problems were coded to the understanding stage when they interfered with a student’s ability to form a response. During the interviews, lexical problems often occurred when the respondent did not understand genetic terminology such as *allele* or *heterozygous*; such problems were attributed to knowledge deficit in the test domain and were not coded. Approximately half of coded lexical problems
were caused by single words with multiple meanings or nuances, particularly common words such as *form, different, and new*. As an example, some students wondered if the phrase *produce new genes* meant to make an identical copy of a gene, or to produce a novel gene. Other lexical problems occurred when students did not understand the meaning of combinations of words. Examples are *personalized prescribing, flow of genetic information* and *family planning*. In one instance, a lexical problem occurred at the response formatting stage when a student correctly identified that DNA sequence refers to the order of base pairs, but was not able to map that answer to the response options (the intended correct response was *nucleotides*, which form base pairs).

<table>
<thead>
<tr>
<th>Problem Type</th>
<th>Response Stage</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Understanding</td>
<td></td>
</tr>
<tr>
<td>Lexical</td>
<td>21</td>
<td>22</td>
</tr>
<tr>
<td>Temporal</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Logical</td>
<td>7</td>
<td>26</td>
</tr>
<tr>
<td>Computational</td>
<td>7</td>
<td>7</td>
</tr>
<tr>
<td>Omission/Inclusion</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Total</td>
<td>37</td>
<td>57</td>
</tr>
</tbody>
</table>

|                     | Task performance   |       |
| Lexical             | 0                  |       |
| Temporal            | 0                  |       |
| Logical             | 11                 | 26    |
| Computational       | 0                  | 7     |
| Omission/Inclusion  | 0                  | 2     |
| Total               | 11                 | 57    |

|                     | Response formatting|
| Lexical             | 1                  |
| Temporal            | 0                  |
| Logical             | 8                  |
| Computational       | 0                  |
| Omission/Inclusion  | 0                  |
| Total               | 9                  |

Table 3
Step Three: Respondent Problems Identified on GNCI Pretesting

No temporal problems were identified. This was anticipated, because unlike many surveys the GNCI does not involve recall of autobiographical information in a given time frame. Conrad and Blair (1996) included a specific category for temporal problems in the matrix because so many surveys address behavioral frequency.

Logical problems comprised almost half of problems identified during cognitive interviewing. These were fairly evenly distributed between the different response stages. In the understanding stage, logical problems occurred due to negative-framed questions and questions
about individuals with similar names (e.g., Jack and Jake) or names that were perceived to be gender-neutral when the respondent felt it important to know the gender. A few students found one question to be difficult because the stem did not naturally lead them to think about the response options. Most of the logical problems during the task performance stage occurred when one inventory item informed another. This was not unexpected, because concepts were interrelated and because more than one question was written for some concepts. Logical problems occurred during response formatting when more than one distractor was correct, when distractors had two parts and the student thought only one part was correct, and when an illogical distractor was included.

No computations were required to complete the GNCI. However, Conrad and Blair (1996) suggested using the computational category as a residual category for problems that don’t fit well elsewhere. Computational problems related to complicated syntax were identified during the understanding stage, when wordy stems and/or response options proved difficult for students to navigate and remember. Questions framed in the context of a specific disease were occasionally problematic. On a few occasions, students reported anxiety that a lack of knowledge about that specific disease would be a barrier to answering the question; at other times students based their answer on personal knowledge about someone who had the disease in question.

Omission/Inclusion problems occurred just twice, both times related to questions about gene function. These were understanding problems and occurred when students questioned whether an item referred to the function of a single gene or of genes in general.

When a problem with an inventory item was identified, a solution was devised to resolve the problem and the item was revised and retested. Some items were revised multiple times across the 15 interviews and therefore underwent limited testing in their final form. During the
course of the interviews, gaps in content domain related to carrier testing and mutations were identified. Such recognition of important omissions during cognitive interviewing has been described by others (Desimone & LeFloch, 2004). In each case, SSR data in those content areas were reviewed, and new items were added to the draft inventory. Therefore, the number of items on the draft inventory increased from 55 to 60 across the course of interviews.

Following analysis of cognitive interview data, final revisions were made to inventory items. To accommodate time constraints during pilot testing, eight items were omitted, resulting in a 52-item inventory ready for pilot testing (Appendix J). In all cases, omitted items addressed concepts covered by retained items. Based on those 52 items, the content domain was again reviewed. Interview data informed minor changes in verbiage for concept titles and descriptions. The concepts were also reorganized to better reflect the content domain of the pilot inventory. The new concept list included 16 concepts in 5 topical categories. Five concepts were broad enough in scope that they were parsed into subconcepts. This organization of concepts and subconcepts, which has been utilized in other concept inventories, was anticipated to be useful during item analysis during Step Four. The resulting 16-concept content domain, which included 20 subconcepts, provided the basis for the pilot inventory (Table 4).

Although the primary focus of the interviews was to troubleshoot the inventory rather than measure knowledge, item difficulty was of interest to the investigator. Because items on the draft inventory were revised between interviews and because responses were not forced, a precise measurement of item difficulty was not possible. However, an estimate of inventory difficulty indicated a mean score of 57% correct responses (range 36 to 80, SD 13.3), indicating slightly lower difficulty than the targeted range of 40 – 55%.
<table>
<thead>
<tr>
<th>Category</th>
<th>Concept</th>
<th>Subconcept</th>
<th>Items</th>
</tr>
</thead>
<tbody>
<tr>
<td>Human genome basics</td>
<td>Genetic composition and organization</td>
<td>DNA composition</td>
<td>1,8</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Genome organization</td>
<td>3,7,15</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Alleles</td>
<td>4</td>
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Table 4  
Step Three: Pilot Inventory Content Domain  
16 concepts and 20 subconcepts in 5 topical categories  

Step Four: Pilot Testing, Psychometric Analysis, and Final Inventory Revision  
In the last step of inventory development, the 52-item GNCI pilot version was tested with 238 baccalaureate nursing students. Psychometric characteristics of the entire inventory as well as individual items were explored. Based on those data, the inventory was reduced to the most
parsimonious form possible, retaining the most robust items while striving to maintain coverage of the content domain. The product of this research endeavor is a 31-item inventory ready for further validation and refinement.

Pilot Testing

During pilot testing, the 52-item GNCI was completed by two cohorts of students who were beginning their first and their last semesters of a baccalaureate program. All students completed both the paper survey and the Scantron in the time allotted. Any missing data on the Scantron forms were retrieved later from the paper surveys. There were consequently no missing data at the time of analysis.

Analysis of Pilot Test Data

Three kinds of data were generated by pilot testing: information about the student sample, psychometric information about the entire inventory, and psychometric measures of individual inventory items. Those data are described below.

Student Sample

All 238 pilot test participants were enrolled in a baccalaureate nursing program: 117 were junior students in their first week of nursing classes, and 121 were seniors just beginning their final semester. There were 44 male students (18%) and 194 female students (82%). Ages ranged from 19 to 55 years ($M = 24.87$, $SD = 6.25$). Twenty students (8%) had taken a previous course in genetics.

Scores on the 52-item pilot test ranged from 11 to 46 ($M = 26.3$, $Mdn = 26.0$, $SD = 6.93$). Score distribution approximated normality (skewness = .303, kurtosis = -.584, see Figure 5). Scale difficulty was 51%. Comparison of mean scores between students groups showed a statistically significant effect only for progression in the nursing program and completion of a
genetics course. Scores of senior students ($M = 29.3$, $SD = 6.34$) were higher than those of junior students ($M = 23.2$, $SD = 6.14$), $t(236) = 7.49$, $p < .001$; and scores of students who had taken had taken a previous genetics course ($M = 31.3$, $SD = 7.04$) were higher than students who had no course ($M = 25.9$, $SD = 6.75$), $t(236) = 3.40$, $p < .001$. There were non-statistically significant differences in score between three campus sites, $F(2,235) = .202$, $p = .903$, and between genders, $t(236) = .43$, $p = .668$, and there was a statistically non-significant correlation between test score and age, $r(236) = .03$, $p = .605$.

Figure 5
Score Distribution for GNCI 52-item Pilot Test ($N = 238$)

Scale Analysis

Psychometric features of the pilot inventory included measures of difficulty and internal consistency reliability. Scale difficulty on the 52-item pilot test was 51%, which fell within the target range of 40 to 55% but is less difficult than most concept inventories. A broad range of item difficulty was noted. The Cronbachs alpha and the K-R 20 value were .79, indicating
acceptable reliability. Of note, the K-R 20 was higher for senior students compared to junior students (.77 and .73, respectively). An additional indication of reliability was derived by comparing the range of scores to the standard deviation. The resulting ratio of 5.05 suggested the inventory may be able to measure across a broad range of student knowledge (Statistical Analysis of Multiple Choice Exams, n.d.).

*Item Analysis*

Item analysis was based on aggregate data from both student groups. Psychometric data reflecting item difficulty, discrimination, and correlation was entered into an Item Analysis Table (Appendix K) to facilitate comparison. Applying the iterative process previously described, the psychometric characteristics of each item were examined in the context of the entire content domain to inform decisions about item retention, revision or omission. Data about the 52 pilot test items are discussed below.

*Item difficulty.* Item difficulty, reported as a percentage of students who answered each item correctly and reflected in the table as a p value, ranged from .11 to .92 ($M = .51$, $SD = .21$). Applying criteria described in Chapter 3, six items answered correctly by fewer than 25% of students were considered too difficult, and two items answered correctly by more than 85% students were considered too easy. Those eight items were flagged for omission or revision.

*Item discrimination.* As previously described, the two software programs utilized in this study calculated different measures of item discrimination. The absolute values of the point-biserial correlation and the corrected item-total correlation varied for a given item; however, the relative discriminatory power of the items was nearly identical, irrespective of which value was used. Point-biserial values reported by the Scantron software were calculated once for the 52-item pilot test, and corrected item-total correlations were calculated several times during
inventory reduction. Both values were considered during initial inventory reduction; however, after the first item set was omitted and the reliability analysis was repeated on retained items, item-total correlations represented the sole measure of item discrimination.

Pilot data point-biserial correlations (r_{pb}) from the Scantron report ranged from -.01 to +.57 (M = +.29, SD = .14). Based on criteria described previously, 12 items showed good discrimination (r_{pb} > +.40) and 28 items showed fair discrimination (r_{pb} between +.20 and +.30). The other 12 items displayed poor discrimination (r_{pb} values < +.20) and were flagged for consideration of revision or omission. In addition, a minimum critical point-biserial value, representing two standard deviations above zero, was calculated to be +.13. Nine of the 12 flagged items fell at or below this level of discrimination. All this information informed initial inventory reduction.

Corrected item-total correlations calculated in SPSS ranged from -.11 to +.52 (M = +.23, SD = .14). Sixteen items correlated to the total score above the +.30 cutoff suggested by Nunnally & Bernstein (1994), and 15 items correlated between +.20 and +.30. The remaining 21 items, which had correlations below an arbitrary value of +.20, were flagged for review.

*Item correlation.* Item correlation was explored by reviewing the inter-item correlation matrix, the average corrected inter-item correlation, and Cronbach’s alpha if an item was deleted. In general, item intercorrelation was modest and did not impact decisions about item retention, although the effect on alpha if an item was deleted was considered during inventory reduction.

The 52 x 52 item correlation matrix revealed inter-item correlations ranging from -.22 to +.51 (M = +.07, SD = .09). Only 14 item pairs correlated in the +.3 to +.7 target range recommended by Ferketich (1991); those items were compared to see if they measured the same construct, but this information did not impact decisions about item retention. No item pairs
correlated > .7, which would indicate redundancy; in fact, the item pair with the highest correlation, questions 15 and 32 (Q15 and Q32), correlated at .51.

Average corrected inter-item correlations were calculated by importing data into Microsoft Excel. The values were modest and showed limited variance: The mean value was .084 with a range from <.001 to +.165. As described, items were ranked from highest to lowest average corrected inter-item correlation, and those rankings were displayed in Item Analysis Table (Appendix K). Most decisions about item retention, however, were made on other bases.

The value of Cronbach’s alpha for the entire scale if any item were deleted was also considered. As previously described, values were converted for ease of interpretation before entering them into the Item Analysis Table (Appendix K). Deletion of any of 40 items would decrease the alpha by as much as .13, deletion of five items would have no effect, and deletion of any of seven items would increase the alpha by up to .05. Those last seven items were flagged for review.

Response frequency. The final values considered during inventory reduction came from the Scantron response analysis data. These data were particularly useful later in the process, to inform revision of retained items. However, data for all 52 items were reviewed during initial item analysis to identify items with unselected distractors or distractors with negative point-biserial values.

The number of response options for the 52 items on the pilot inventory varied: 20 items had five possible answers, 31 items had four possible answers, and one item had just three response options. The total inventory therefore included 227 response options: 52 correct responses and 175 distractors. During pilot testing, none of the 175 distractors was unselected. The lack of unselected distractors likely reflects the derivation of item distractors from identified
student misconceptions. Twenty-three distractors had positive point-biserial values. During scale reduction, response data occasionally informed choices between two similar items, but the data were of principal interest during final revision of retained items.

**Inventory Reduction and Response Analysis**

Following pilot data analysis, the 52-item pilot version of the GNCI was reduced, applying the iterative process described in Chapter 3. The first items to be eliminated were those with values outside the target ranges for multiple criteria: difficulty, discrimination and correlation. Based on p value, $r_{pb}$ value, item-total correlation, alpha if an item was deleted, and ranked average corrected inter-item correlation, five items were eliminated during the first round. Three of those items were thought to be crucial to the content domain and were flagged for later revision. Next, following the recommendation of Nunnally & Bernstein (1994), item discrimination was applied as the primary criterion for item selection. Eight items with $r_{pb}$ values <.2 were omitted, checking to ensure the remaining 39 items covered the entire content domain. At this point, reliability analysis was run on the 39-item scale. A new item analysis table was created to display the psychometric features of the remaining items and facilitate further inventory reduction. Across several iterations, items were omitted on the basis of their psychometric characteristics with a constant eye on content domain. Each time a set of items was omitted, analysis was repeated on the remaining items, since removing even a single item influences item-total correlations values as well as the psychometric characteristics of the entire inventory. After five rounds of item analysis, the 52-item pilot version had been reduced to 26 items, and five items omitted because of unacceptable psychometric features had been identified as crucial to the content domain. Of the 26 retained items, 15 items were unchanged from their original form and 11 items were modified in nonsubstantive ways such as rewriting a response or
changing the genetic condition in the stem. Of the five rejected items, four were substantially revised, and one item (Q8) was replaced with a question from the Test of Genetics Concepts (Sadler, 2003; permission granted). In total, 31 items comprise GNCI beta version, one item more than the target range of 25 - 30 items.

Response analysis for 26 items retained in the beta version was examined in detail during final inventory review. The 26 items had a total of 119 possible responses: 26 were correct and 93 were distractors. Although no distractor was unselected, 11 distractors were selected by <3% of students. Because distractors that are rarely or never selected do not add to the value of a scale, those distractors were reviewed, in each case going back to the SSR data. None of the distractors was removed, pending further testing, although one was replaced. Review of point-biserial data identified positive $r_{pb}$ correlations for seven distractors. The values were low, ranging from .01 to .09 with an average of .04. Those distractors were also critically reviewed, however none were eliminated.

Response analysis concluded development of the GNCI beta version. All 31 items in the GNCI beta version (Appendix L) items are research based, having been developed from misconceptions identified in a sample of nursing students. Although 26 of those items have been tested, five items have yet to be validated with the target population. Therefore, the GNCI in its current beta version represents a preliminary step in inventory development.

Redefinition of the Inventory Content Domain

The content domain for the GNCI was modified throughout inventory development, and its final definition was necessarily deferred until inventory reduction was complete. The content domain was built on 85 concepts initially extracted from the essential competencies. The list was reduced to 65 concepts to constrain the burden on the expert panel, and by the end of Step One,
an initial content domain of 21 concepts (Table 2) had been defined. While 21 concepts were more than could reasonably be tested in a 25-to-30 item inventory, some concepts were expected to fall out during further inventory development. The 21-concept domain was unchanged following SSR surveys in Step Two. After cognitive interviews and inventory drafting in Step Three, however, concepts were removed, due either to inability to write robust items or concern about inventory length. At the end of Step Three, the content domain included 16 concepts, with six broad concepts subdivided into 20 subconcepts to support item analysis (Table 4). That content domain was represented by 52 items comprising the GNCI pilot version. At the end of inventory reduction in Step Four, the GNCI had been reduced to 31 items. Those items were mapped once again to the content domain, unrepresented concepts were eliminated, and the remaining concepts were reorganized into four topical groups. The product of this study, the 31-item GNCI beta version, reflects a content domain including 18 concepts in four topical categories (Table 5).

Estimation of GNCI Psychometric Features

The psychometric features of the GNCI beta version are unknown, since 5 of the 31 items are untested in their current form among the target population. An estimate was made, however, by running a reliability analysis on the 26 items that were tested in identical or similar form during Step Four (Appendix M). That analysis estimates, to the degree possible, how the inventory might perform in further testing.

Psychometric analysis of the 26 retained items indicated acceptable item and scale difficulty for a concept inventory. Item difficulty spanned a broad range, from 25% to 83% ($M = 53, SD = .17$), indicating capacity of the scale to measure varying levels of knowledge. The 53% scale difficulty is less than most concept inventories but within the target range of 40 to 55%.
### Table 5
Step Four: Content Domain for GNCI 31-item Beta Version
18 concepts in 4 topical categories

Overall, item discrimination was acceptable, with corrected item-total correlations ranging from +.19 to +.52 ($M = +.33$, $SD = .10$). One item, Q49 on the pilot inventory (Appendix J), had a poor $r_{pb}$ value of .17, indicating it does not discriminate between students with high and low inventory scores. The rationale for retaining this item is discussed in Chapter 5. All other items showed acceptable $r_{pb}$ values between .25 and .57, and the mean value of .37 lies between the ranges identified as “fair” and “good” according to Nunnally & Bernstein (1994).

Scale reliability was reflected by Cronbach's alpha for the 26 items of .804. Omission of any one of 25 items would leave the Cronbach's alpha unchanged or reduce it by as much as .015.
Omission of one item (Q12) would increase Cronbachs alpha by .02; this item was retained on rational grounds, as will be discussed in Chapter 5.

Measures of internal consistency revealed modest correlation between inventory items. This was not unexpected, given the broad content domain. Item-total correlation ranged from +.10 to +.52 ($M = .31, SD = .10$), and the average item-total correlation of .31 falls just above Nunnally & Bernstein’s (1994) recommended lower limit of .30. Average corrected inter-item correlation was low as anticipated, with a mean value of .11 ($SD = .03$).

The Genomic Nursing Concept Inventory, in its beta version (Appendix L), represents an empirically based scale to measure genomic literacy among baccalaureate nursing students. The content domain is based on recognized practice competencies and validated by genetic nurse experts. Inventory items reflect the most common genetic and genomic misconceptions held by a sample of nursing students, and preliminary psychometric testing indicates the inventory may be capable of distinguishing between students who understand important concepts and those who do not. The GNCI is ready for further testing and validation and offers to provide a ruler by which to measure genomic literacy among nurses. Such a tool has great promise in supporting genomic nursing education.
CHAPTER FIVE
DISCUSSION

This chapter provides a discussion of the research methodology, as well as results and limitations of the current study. Next steps for further inventory development and potential applications of the Genomic Nursing Concept Inventory (GNCI © Ward, 2011) in nursing education will also be considered.

Discussion of the Research Process

The four steps employed to develop the GNCI were adapted from processes used in the development of other concept inventories in several disciplines. The methodology and findings of each step will be summarized and evaluated in turn.

*Step One: Extraction and Validation of Key Concepts*

Development of the GNCI began with identifying an initial content domain for the inventory. This was a critical step, since the validity of an inventory ultimately rests on the degree to which it can measure the attribute of interest. Here, that attribute was genomic literacy, defined as the knowledge set required to “deliver competent genetic- and genomic-focused nursing care” (Consensus Panel, 2009, p.1). The challenge was to distill a large number of concepts embedded in a broadly-endorsed set of competencies (the *Essentials*) into a smaller number of concepts representing what nurses need most to know about genetics and genomics. Twenty-one concepts were identified and defined the initial content domain for the GNCI.

*Expert Sample and Survey*

An online survey invited genetic nurse experts in the United States to rank concepts embedded in the essential competencies according to salience to nursing practice. With this particular online survey, cost issues did not limit the sample size, and all identifiable experts who
met inclusion criteria were invited to participate. The 37% response rate corresponds to rates reported in one meta-analysis of 33% for surveys of all sizes and 41% for surveys with samples of fewer than 1000 individuals (Hamilton, 2009). The response rate may have been enhanced by the population’s anticipated interest in genomic literacy, and may have been restricted by timing. The survey was conducted in August, when the half of the group with faculty positions may have had limited availability.

Data indicating expertise of the expert panel were compelling, both in years of experience and range of expertise. Nursing education, practice, and research were represented, and 38% of participants had been working in genetic nursing for more than 10 years. Participation by 14 members of the Essentials Consensus Panel lent particular credibility to the data.

Survey Methodology and Design

Online survey methodology was well suited to the expert survey, being convenient, cost efficient and versatile. Sending invitations via email allowed recruitment of a targeted population without incurring printing or postage costs. The Survey Assistant™ platform supported both Likert-style questions for ranking concepts and text boxes to gather qualitative data. Displaying concepts related to each topical category on a single page allowed respondents to balance the relative importance of specific concepts, and the use of forced responses limited missing data. During survey design, respondent burden was a concern due to the large number of concepts to be ranked. Two respondents did make textual comments about the length of the survey, although only five of 104 respondents failed to complete the entire survey.

Concept Prioritization

Although the goal of the online survey was to prioritize the concept list, the overall rankings were also of interest. The mean ranking among all 65 concepts, based on a four-point
scale, was 3.3, which lies between important to know and critical to know. According to expert consensus, only 10 concepts fell below the important to know designation. It is perhaps not surprising that genetic nurses assign high priority to many genetic concepts, but 55 concepts identified as critical or important far exceed the capacity of a concept inventory. Additionally, when most concepts are rated as important and variability is limited, differentiating the relative importance of concepts may be unreliable.

A concept inventory is not meant to measure comprehensive achievement, but rather understanding of fundamental concepts within a domain of knowledge. For genomic nursing, the domain is broad, and expert consensus identified an abundance of concepts of high-ranking importance. Because the goal was to develop a 25-to-30 item inventory with one or two items for each concept, the concept list simply needed to be reduced. Unavoidably, a number of concepts with unmistakable salience to genomic nursing were excluded from the inventory. At the end of Step One, 21 concepts were carried forward.

On a larger scale, it is unlikely that nursing programs will be able to incorporate the 55 important or critical genetic/genomic concepts into curricula. This is particularly true in the current climate of multiple competing demands, numerous sets of competencies (all deemed “essential”), and recent discourse about overfull curricula to which content is continually added without anything being removed (Ironside, 2004; Benner et al., 2010). Such concerns are not unique to nursing education. Other health-related disciplines face a similar challenge of integrating ever more information into finite curricula (McInerney, 2008). Biology educators worry about content overload, noting that each textbook edition contains more pages with no indication students are learning more (D’Avanzo, 2008; Howitt et al., 2008; Michael et al., 2009). Physics faculty have called for reducing the amount of content covered in introductory
courses (McDermott, 1984). Nursing’s commitment to integrate new knowledge while avoiding content overload makes it crucial to identify what nurses most need to know about genetics and genomics.

The content domain, delineated according to consensus of more than 100 genetic nurse experts, was broad. Concepts ranged from basic science, such as the organization of genomic structures and patterns of inheritance, to clinical applications in cancer genetics and pharmacogenomics. However, the GNCI content domain reflects just a subset of the body of propositional knowledge identified as important for health care professionals. For example, principles related to proteomics, the genetics of race, and evolution have been identified as key to the education of health professionals (McInerney & Childs, 2004; McInerney, 2008). Those same concepts, however, were assigned relatively lower priority by the genetic nurse experts and were consequently omitted from the inventory domain. That does not mean genetic nurses consider proteomics and the genetics of race to be unimportant; in fact, both concepts were ranked well within the important category, with mean ratings of 3.24 and 3.29 respectively. Evolution, on the other hand, was ranked considerably lower, with a mean priority rating of 2.79. The capacity of a concept inventory is limited, however, and only 21 of 65 concepts were advanced past Step One. Concepts related to proteomics, race, and evolution were simply outranked in terms of salience to nursing practice.

Although the entire set of concepts the nurse experts found to be important is not represented in the GNCI, the list surely has utility in nursing education. Drawn from the Essentials and validated by expert consensus, the concept list provides a valuable compendium of knowledge to inform curriculum and course development, as well as other educational assessments.
Textual Comments

Two issues identified in the experts’ textual comments warrant consideration. These issues involve (1) delimiting the practice domain for baccalaureate nurses, and (2) responding to the rapid evolution of genetic knowledge.

Several genetic experts expressed uncertainty about the degree to which baccalaureate nurses should be expected to interpret genetic information, particularly the data in a family history. At least one expert suggested that while all nurses should be able to identify a client at increased risk and initiate a referral, quantifying the risk and developing an appropriate plan of care may more appropriately be expected of a genetic specialist or advanced practice nurse. The knowledge domains and skill sets required to achieve these two levels of competency are not easily distinguished, however, and even identifying a client at risk may require skills exceeding the expectations for all registered nurses.

The Essentials clearly expect all professional nurses to be able to elicit a family health history and document that history as a correctly-formatted genetic pedigree. Some degree of interpretation of a genetic history is also required. All nurses are expected to critically analyze history and physical assessment findings for genetic/genomic risk factors, to interpret selective [italics added] genetic/genomic information, and to identify clients who may benefit from genetic/genomic information or services (Consensus Panel, 2009).

The degree of interpretation expected by baccalaureate nurses, which was questioned by some of the experts, is perhaps unclear in the Essentials. Some degree of interpretation is necessary to identify clients who should appropriately be referred. Identifying those clients is not simple, however, particularly as rapidly-evolving knowledge is translated into ever-changing clinical guidelines. Consider making a genetic referral for cancer risk assessment: How would a
nurse know if having a single first-degree relative, or perhaps two second degree relatives, with early-onset cancer increases cancer risk sufficiently to warrant a genetic referral? What age of onset denotes “early” onset? Is it the same for all forms of cancer? While coursework should prepare baccalaureate-educated nurses to access current and credible clinical guidelines, the preparation of nurses in non-baccalaureate programs to identify and implement evidence-based practice is less certain. The Essentials, however, were written for “all registered nurses regardless of academic preparation, practice setting, role, or specialty” (Consensus Panel, 2009, p. 8).

Issues in documenting and interpreting a family health history exemplify the inherent difficulty in writing a single set of competencies for professionals with varying levels of academic preparation. The experts’ suggestion that documenting genetic history and making an appropriate referral lies within the realm of baccalaureate nurses, while interpreting the history to plan care represents advanced or specialty practice appears rational and realistic. However, expecting nurses without specific preparation in information literacy to identify and interpret current and credible evidence in order to make an appropriate referral is implausible.

The other theme that emerged from expert comments is that genomic literacy cannot be static. The hallmark of the fast pace of genetic discovery is “what is true today may not be true tomorrow.” Some genetic misconceptions are in fact historical. One example is the time-honored concept that one gene encodes one protein. That idea, once a central dogma of genetics, has long been known to be inaccurate but remains a common misconception (Albuquerque, de Almeida, & El-Hani, 2008). The classical molecular gene model portrayed a gene as an uninterrupted DNA segment with a single function and is insufficient to explain new knowledge that genes may be split, nested, or overlapping, and regulated by distant promoters. The metaphor of a gene
as “a bead on a necklace” no longer fits, and the very nature of a gene has been dubbed “a concept in trouble” (Keller, 2000).

Predicting future clinical applications of ongoing genomic discovery is an uncertain enterprise, and so genomic literacy for all health professionals must be impermanent. Recognizing the rapid and escalating pace of genomic discovery, the Consensus Panel (2006) acknowledged the competencies were based on the “state of evidence available at the time they were developed” (p. 1). Since 2006, the bounds of genomic literacy have been strained by discovery of phenomena such as copy number variation, the “missing” heritability of complex disease, and epigenetic effects on gene regulation. For example, as epigenetic findings are translated into clinical applications, nurses may need to have a better understanding of gene regulation. Several experts in the current study suggested epigenetics as an important concept to consider during inventory development, although overall the experts ranked the concept 62nd of 65. One expert, noting that epigenetics is “very hot right now,” contemplated whether knowledge of epigenetic effects would become important in genomic nursing. Only time will tell, of course.

In the meantime, framing genomic literacy in conceptual understanding of fundamental concepts provides the best chance to prepare a nursing workforce to implement genomic applications that cannot easily be predicted. Nurses who understand basic concepts of genetic structure, organization, and function will have a basis for understanding gene-based testing and interventions. This is true even as genomic discovery continues to mold basic conceptual understanding over time. All evidence indicates nurses lack basic genetic and genomic understanding today. Ensuring, to the degree possible, that the GNCl measures understanding of basic concepts most likely to support genomic nursing as it evolves, is a practical and useful aim.
Like the *Essentials* on which it is framed—and like the very definition of a gene itself—the GNCI represents a work in progress.

**Step Two: Exploration of Student Understanding and Inventory Drafting**

The purpose of Student Supplied Response (SSR) surveys in Step Two was to identify the level of understanding among baccalaureate students of the key concepts identified in Step One, and to identify the most common misconceptions. Open-ended questions about each of the 21 initial key concepts were administered to students, who provided textual responses. Analysis of those responses informed item writing for the draft inventory.

**Methodology**

The approach to learning what nursing students knew about the concepts identified in Step One was adapted from the method used by Bailey (2008) in the development of the Star Properties Concept Inventory. The process was efficient and generated nearly 6,000 textual responses to 50 open-ended questions. Ten SSR questions failed to evoke enough useful data to draft an inventory item. An example of an SSR question that missed its target was *what role(s) do proteins play in our bodies?* This question was designed to explore understanding that proteins mediate virtually all physiologic functions, which is fundamental to understanding proteomics. Unfortunately, nearly half of students described the importance of protein as a nutrient.

Data analysis was facilitated by using 6,000 index cards which were convenient to sort and rearrange during the process of thematic analysis. Coding was streamlined by the abundance of misconceptions and the sufficiency of a fairly coarse level of analysis to identify the most common misconceptions for each concept. Responses that were unique or difficult to interpret
were so coded and omitted from further consideration, without concern about missing key misconceptions.

SSR Survey Findings

Although previous studies to identify genetic understanding and misconceptions among nursing students were not identified, such research has been conducted among high school and undergraduate populations (Cho et al., 1985; Lewis, Leach, & Wood-Robinson, 2000; Marbach-Ad, 2001; Marbach-Ad & Stavy, 2000; Shaw et al., 2008). Those studies identified limited understanding and common misconceptions about the composition, structure, and function of genetic structures, and relationships between genetic concepts (Marbach-Ad).

Similar findings emerged from the SSR surveys in this study. In particular, poor understanding of the organization of genetic structures and the flow of genetic information was common. For example, only 27% of nursing students correctly identified that every cell contains a complete set of genes, 29% correctly identified a mutation as an alteration in DNA sequence, and 54% differentiated the homozygous and heterozygous states. Four SSR questions addressed the central dogma of genetics, which is that information flows from DNA to RNA to proteins. On average, only 27% of students demonstrated understanding of that concept. The nursing students also shared several misconceptions noted among high school and university students both in the United States and other countries. These misconceptions included that (a) different cells in the same individual have different genes, (b) DNA is composed of protein, (c) a gene is a trait or is responsible for a trait, and (d) genes determine traits, while DNA is responsible for inheritance (Cho et al., 1985; Lewis et al., 2000; Marbach-Ad, 2001; Shaw et al., 2008).

In some instances, knowledge deficit was so great that common misconceptions could not easily be identified. For example, the concept of an allele is foundational to genetic literacy and
has been reported in other student groups to be widely misunderstood (Pashley, 1994). Nursing students were no better informed: only 7% of nursing students correctly described an allele as a version of a gene, and 33% wrote they did not know what an allele was. Small numbers of students held one misconception or another, while the overwhelming proportion of students simply didn’t know. Because the concept of alleles was felt to be important, an item was drafted, tested during cognitive interviews, and included on the pilot test (Q4, Appendix J). Although the item was easily navigated during cognitive testing, its psychometric features were dismal: It was difficult \((p = .21)\) with a modest \(r_{pb}\) of .16 and poor correlation values. No promising solution to improve the item was identified, because the primary problem was thought to be its difficulty. The item was omitted from the beta version of the inventory, leaving a nontrivial gap in the content domain.

The allele question represents an intriguing dilemma. The goal of this study was to develop a robust inventory capable of discriminating between students who do and do not understand basic and important concepts. The concept of an allele is believed by the investigator to be both basic and important. Pashley (1994) reported that “once students had resolved any difficulty with the relationship between a gene and an allele, “their performance in genetics showed a significant improvement” (p. 1). The online survey did not query experts about the concept of alleles in particular; rather, the concept was included in a broader concept, *Gene basics: structure, function, size, locus, dominant-recessive alleles, homo- and heterozygosity*. The experts ranked this concept 7th most important out of 65 concepts. Further investigation would be necessary to determine the relative importance of the specific concept of alleles. Even stipulating, however, that the concept is of high importance, writing a robust item about a poorly-understood concept is fraught with difficulty. Robust items have moderate difficulty and good
discriminatory power; and difficult items such as the allele question tend to have poor discrimination. Therefore, irrespective of the concept’s importance, retention of the allele question threatened inventory reliability. The allele question might have been preserved on rational grounds, as were two other items with substandard psychometrics. In this case, however, and at this stage in inventory development, the item was omitted.

*Inventory Drafting*

As SSR data were compiled and analyzed, item writing for the GNCI began, utilizing the most common misconceptions as item distractors. The goal was to use simple language and, for the distractors, to use the same language students used during the SSR surveys. For some questions, the process was simple: The SSR question was restated and four or five possible answers listed, with one correct answer and three or four distractors reflecting students’ most common misconceptions. Questions about genetic structure and function, for example, were straightforward, particularly when most students wrote a response and the responses revealed abundant misconceptions on which to base distractors.

When possible, questions were framed in nursing applications, in order to reinforce the salience of genetics and genomics to nursing practice. Those items were more difficult to write, requiring more complex stems that threatened to undermine the item-writing guideline of avoiding the “window dressing” of excessive verbiage (Haladyna, Downing, & Rodriguez, 2002). An example is provided in the following item, which is stated in its initial form:

Jake and Allen are brothers who both inherited neurofibromatosis (NF) from their father. Jake’s symptoms are much more severe than Allen’s. What is the most likely explanation for their variation in symptoms?

a. Jake inherited a dominant form of the NF gene and Allen inherited a recessive form.

b. The DNA sequence of Jake’s NF gene is different than Allen’s.

c. Environmental factors were harmful in Jake and protective in Allen.

d. Jake and Allen inherited the same form of the NF gene, but the gene was expressed differently in Jake compared to Allen.
This question was developed to measure understanding of the concept of variable expression, which describes that in certain disorders, the same gene mutation can cause very different signs and symptoms. The question could have been written simply: *Variable expression means* . . . . However, an aim for the GNCI is that the very act of completing the inventory raise consciousness about the salience of genomics to nursing. During inventory development, the goal to set items in a nursing framework had to be balanced with limiting item length. Incidentally, the above item violated two other rules of item writing: The correct answer (d) was significantly longer than the distractors, and a valid argument could be made that an intended distractor (c) is also correct. This item was revised several times across cognitive interviews but was ultimately omitted from the inventory.

Three items were difficult to write without using a negative stem, i.e., *Which patient is at least risk for hereditary breast cancer?* Negatively-stated questions are prone to misunderstanding and should generally be avoided (Haladyna et al., 2002). Cognitive interviews readily revealed when students with adequate knowledge answered incorrectly solely because they missed the negative modifier.

Ten SSR questions failed to elicit a distinct set of misconceptions, which also complicated item writing. One such question asked students to identify diseases or health conditions important to include in a genetic risk assessment. This concept was assigned high priority by the expert survey; its intent was to explore whether students understood that genetic risk assessment should include not only rare traditional ‘genetic’ conditions, but also common chronic ‘genomic’ conditions such as diabetes and heart disease. Responses indicated remarkably good understanding of this concept: students named complex genomic conditions four times more often than traditional genetic diseases. These data may have been skewed, because the SSR
question was posed following presentation of family health history content in a concurrent class. Repeating this SSR question in a naïve student population might produce different results. Nevertheless, the question failed to elicit misconceptions sufficient to inform the creation of an inventory item. In a paradoxical example, widespread knowledge deficit made it difficult to write an item based on student understanding. The concept of privacy issues related to genetic testing was highly ranked by the expert panel, and three different SSR questions were posed to explore student understanding of privacy issues. The SSR questions failed, however, to uncover misconceptions. Students knew almost nothing about legal protections of genetic information, although most of their perceptions were reasonable. Because the concept was so highly ranked, an item (pilot version Q36) was written and pretested, however the question was very difficult (p = .15) with among the poorest discrimination values of all inventory items (r_{pb} = .12). Because a promising solution for improving the item was not identified, the item—and the concept—were omitted from the GNCI beta version. Similar to the issue of the allele concept, limited knowledge of genetic privacy dictated inventory construction. A robust inventory cannot be built of concepts lacking a minimal level of understanding in the target population. However, whether or not these concepts are represented on this particular inventory, data from the SSR surveys offers unique and critical insight into knowledge that must be integrated into nursing education.

*Step Three: Pretesting and Inventory Refinement*

The 52-item draft inventory was pretested with 15 first-year nursing students during individual cognitive think-aloud interviews to troubleshoot and improve inventory items. A matrix, as suggested by Conrad & Blair (1996) was used to objectify the coding of interview data. An iterative approach was employed, with items being revised as problems were identified and then retested in their new form as additional students were interviewed.
Cognitive Interview Methodology

In general, the cognitive interviews provided a useful and efficient means of pretesting the inventory, uncovering 57 coded problems in addition to other suggestions and insights. For example, two gaps in content domain were brought to light during the interviews. The interviews also generated insights into the usefulness of this methodology.

Wide variance was evident in the amount of verbalization that could be elicited from different students. Some students articulated their thoughts in detail with very little probing, while others offered little information about their thought processes despite frequent probes. The variable amount of data generated by the interviews was mirrored in interview duration, which ranged from 48 to 96 minutes to discuss 52 to 60 items.

Students were provided multiple opportunities to understand their role during the interviews as well as a significant monetary incentive. Upon recruitment, students were informed that the interview would entail reading test questions aloud and verbalizing their thoughts as they formulated answers. The process was described in greater detail in the informed consent statement signed by each participant. Before each interview began the purpose and process of the interview were again iterated. Specifically, students were asked to read each item stem, describe what they thought the question was asking, attempt to answer the question before uncovering the response options, and then verbalize their thoughts as they selected the best response. In retrospect, however, it may have been beneficial to invest time in teaching the students how to perform the think-aloud procedure at the beginning of each interview, in hopes of maximizing the data collected. Such a procedure is in fact recommended by Willis (2004).

A convenience sample of 15 volunteers who were just completing their second semester of nursing school participated in the interviews. Detailed demographic data was not collected,
although both male and female students participated, and two students had a primary language other than English. A sample size of 10 to 15 is, according to Willis (2004) probably adequate for a round of cognitive testing; however, sequential iterations of a scale are often tested in up to three or four rounds. In this study items were revised as problems were identified, and any revised item was therefore tested fewer than 15 times. Items that underwent revision late in the round of interviews were likely to be tested in their new format very few times. Therefore, the sample size of 15 may have been modest for this study. Any limitation associated with the sample size is mitigated by the qualitative nature of the interview data, which did not result in statistical estimations (Willis). The goal of the interviews was simply to improve the items.

**Cognitive Interview Findings**

The purpose of the GNCI is to measure knowledge. In order to minimize measurement error, it was therefore critical to identify and solve any problems students encountered with inventory questions other than problems related to knowledge deficit. The think-aloud process was critically important to identify such problems, most of which were unanticipated during item writing. Over the course of 15 cognitive interviews, students completed draft inventories that contained between 55 and 60 items.

Only 15 of the 55 to 60 items had no problems uncovered during the cognitive interviews. Four of those were very short questions with little anticipated occasion for misunderstanding, e.g., *When a gene is expressed, it __________*; and *An allele is __________*.

Several application questions requiring longer stems to establish a nursing framework were navigated easily by students. This was encouraging, as one of the goals of item development was to highlight the salience of genomics to nursing practice. That goal was tempered by the desire to avoid unnecessarily long item stems.
Types of problems identified during cognitive interviewing were described previously in Chapter Four, and solutions for most problems were readily apparent. Two problems, however, warrant specific consideration; these are negatively-worded questions and semantic issues. Unless otherwise stated, item numbers in the following discussion refer to the 60-item draft version of the GNCI (Appendix I).

Negatively-phrased questions increase the chance of measurement error, and effort was made to avoid such questions on the GNCI. However, despite those efforts, three such questions were included in the draft inventory. Two of the questions (Q1 and Q33) posed no problem for students during the interviews. In both cases the stem was very short. However, the majority of students answered Q27 incorrectly because they missed the word LEAST in the item stem, which was lengthy. Q27 addressed a high-priority concept and was thought to be important to the content domain. Several solutions, including bolding and underlining and shortening the stem, were tested but students still misread the question. The item was eventually rewritten using positive language, despite concern about reducing the item’s discriminatory power. On pilot testing the item (now Q48) showed poor discrimination ($p = .48, r_{pb} = .07$) and was omitted from the beta version.

Semantic issues also posed difficulty in writing robust inventory items. A form of linguistic shorthand has emerged in discussions about genes and disease. As a result, people may associate a gene per se, rather than a gene mutation, with a particular disease. This semantic issue has likely contributed to a common misconception that people with a genetic disease have a particular gene not found in other individuals (Collins, 2007). $BRCA$ genes provide a fitting example. Both $BRCA1$ and $BRCA2$ are tumor suppressor genes with roles in cancer prevention. Every individual inherits two copies of each gene, one from each parent. Cancer risk increases
when a \textit{BRCA} gene is altered in a way that impairs its ability to suppress tumors. The SSR surveys indicated 42\% of students to believe a woman with a positive BRCA mutation test has a gene not found in women whose BRCA test was negative. During the cognitive interviews, it quickly became apparent that very precise language was necessary in item stems and responses to distinguish, for example, between a \textit{BRCA1} gene and a \textit{BRCA1} mutation.

\textit{Step Four: Pilot Testing, Psychometric Analysis, and Final Inventory Revision}

In the last step of the research process, the 52-item GNCI pilot version was tested with 238 nursing students to provide data for psychometric analysis. Based on those data, the inventory was reduced to a 31-item beta version, which represents the product of this research. Of the 31 items on the beta version, 26 items were retained from the pilot version in identical form or with minor revisions, and 5 items were revised significantly or replaced. Because those 5 items are untested in the target population, the psychometric features of the GNCI beta version are unknown pending future testing. Currently, the best possible estimate of GNCI performance is based on reliability analysis of the 26 items retained from the pilot test. Pilot testing methodology, data analysis, and final inventory reduction are discussed below. References to inventory items reflect the item number on the GNCI pilot version (Appendix J).

\textit{Pilot Testing Methodology}

The GNCI was pilot tested in two classes of nursing students within regularly-scheduled classes as previously described. Incentives were provided to encourage students to complete every inventory item. Due to concern about whether students could complete the 52-item inventory in the available time, students were asked to complete the paper inventory before filling in their Scantron; however, all students completed both the paper inventory and the Scantron within 60 minutes. In some instances, Scantron bubbles were not filled, but in each case
that data was retrieved from the paper survey, resulting in no missing data among 12,400 responses.

Pilot Testing Sample

Adequate sample size was important to limit the effects of variance in scale scores. In this case, a convenience sample of cohorts of nursing students who had not participated in the SSR surveys or cognitive interviews was utilized, providing a sample of 238. This sample meets Nunnally and Bernstein’s (1994) recommendation of a sample size of at least 200 for pilot testing. It is also important to design an inventory for the population in which it is to be used. The sample in this study may not reflect the population of baccalaureate nursing students in the United States, and further validation testing is planned in multiple colleges of nursing across the country.

Of interest, only 20 of 238 students (8%) had ever taken a specific genetics course. Among the prerequisite requirements for the WSU College of Nursing are eight semester hours of anatomy and physiology which in turn require a single course in biology. This leaves relatively little opportunity to learn about genetic concepts prior to nursing school, although genetics is included in the recommended science curriculum in public schools in this country. Interestingly, WSU also requires eight semester hours of chemistry, to include inorganic, organic, and biochemistry. There is little in the literature to indicate a rationale for requiring nurses to study chemistry, although Benner et al., in the 2010 Carnegie Report, posit that “nurses must use knowledge from biochemistry to thoroughly understand acid-base balances, electrolytes, and solutions, biochemical cascades, coagulation, and fibrinolysis” (p. 27). It may be, in this day of crowded curricula, that critical review of nursing prerequisites will inform strategies to make the best use of the years students prepare to enter nursing school. A human
genetics course might be equally as useful as a chemistry laboratory in preparing tomorrow’s nurses.

Pilot Testing Findings

Following pilot testing, data analysis and inventory reduction, the psychometric features of the beta version of the GNCI were estimated by analyzing the set of 26 pretested and retained items. The difficulty, discrimination and reliability of the GNCI are discussed below. Item numbers refer to the pilot version (Appendix J).

Scale difficulty. Based on the 26 retained items, the difficulty of the GNCI is estimated to be 53%. Although this level of difficulty is lower than optimal for a traditional achievement test, it fell within the target range of 40 – 55%. Concept inventories (CIs) are designed to measure difficult concepts (Nelson et al., 2007) and typically feature low difficulty index values (reflecting higher difficulty). Many CIs have a level of difficulty of 40% or lower when administered as a pretest. Scale difficulty can be manipulated by removing items that are answered correctly by many students or by changing distractors to make them more plausible. Q52, in particular, may be too easily answered for this type of scale but was left on the inventory pending further validation. During further development, close attention will be warranted to ensure the GNCI has sufficient difficulty to be useful in both pre- and post-testing. Retained items did show sufficiently wide variability in difficulty (range = .26 to .83, \( M = .53 \)), which is a desirable feature in a scale designed to measure across a broad range of knowledge.

Discrimination. The ability of the GNCI to distinguish between students who do and do not understand genetic and genomic concepts relies on the discrimination index of individual scale items. As previously discussed, corrected item-total correlation was used to measure item
discrimination throughout inventory reduction. The goal was to achieve values of .3 or greater, although content domain coverage was not sacrificed to increase scale discrimination.

Corrected item-total correlations for the 26 retained items ranged from .13 to .52, with an average value of .33. The least discriminating item (Q12) was retained on rational grounds. This item was also the most difficult of the retained items (p = .26); difficult items are typically not very discriminatory (Nunnally & Bernstein, 1994). However, the item reflects the central dogma of genetics and tests the basis for microarray analysis, a technique increasingly used as a clinical measure of gene expression. Therefore, the item was retained for the next round of testing.

Tests of discriminatory reliability await further inventory development, however small but significant group differences were noted on pilot testing. Senior nursing students achieved scores 12% higher than junior students, and scores of students who had taken a previous genetics course were 10% higher than students who had no course. The lack of a correlation between score and age lends no support to a postulation that genetic knowledge may be greater among younger college students as a result of 1996 changes in the National Science Education Standards.

*Internal consistency*. Throughout inventory reduction, measures of internal consistency persistently revealed limited inter-item correlation. The average inter-item correlation for the GNCI beta version is estimated to be .17. This low level of internal consistency likely reflects the broad content domain of the GNCI. Only 14 item pairs correlated >.30; these items were compared to see if they measured the same construct, but in general this information did not impact decisions about item retention. No item pairs correlated >.70 which would indicate redundancy; in fact, the item pair with the highest correlation (Q15 and Q32) correlated at .541.
Reliability. The initial Cronbach’s alpha for the 52-item pilot test was .791. The alpha was recalculated several times during inventory reduction. Although reducing the number of items on a scale limits alpha, omitting items with poor performance was hoped to mitigate or reverse that effect. Although reliability for the final 31-item iteration is unknown, the best estimate on the basis described previously predicts an alpha of .804. This value exceeds the minimal recommendation by Nunnally & Bernstein of .70 for a new instrument, although it is noted that other genetic concept inventories have shown very high alphas. The 31-item Genetic Literacy Assessment Instrument, for example, has an alpha of .99 (Bowling et al., 2008).

Limitations of the Study

Several limitations to the current study are identified and will inform further inventory validation and development. The most critical of these regards the content domain. While the size and the expertise of the expert panel are indisputable, the method used to tap their expertise may not be the most robust. Clayton (1997) argues that the group average method may fail to capture participants’ “most thoughtful input” and certainly fails to provide “benefit of hearing other responses that might encourage a refinement of their contributions” (p. 373). In addition, limited variance between the priority ratings in the expert survey likely threatens the reliability of key concept identification, despite having over 100 participants. The remedy to this limitation lies in further inventory development. Development of a robust concept inventory is indeed a “long, arduous process” (Nelson et al., 2007), and there is no magical sequence of steps. Validation of the content domain using the Delphi method with a subset of genetic nurse experts is a logical next step and is discussed below.

A second limitation lies in sampling forSteps Two, Three and Four. The convenience sample of students at a single college of nursing, while readily available, may not accurately
reflect the population of baccalaureate nursing students for whom this inventory is designed. The solution to this limitation lies in testing the GNCI beta version at many colleges of nursing across the country, which is planned for the next step of inventory development. The option to randomly select students from among a large population is under consideration.

A third limitation lies in the sample size for the cognitive interviews. As described, 15 interviews are probably sufficient for a single round of cognitive interviews. In this study, however, because changes were made to inventory items as problems were identified, some items were pretested with fewer students, limiting the opportunity to find and correct problems with those items. It is possible that some items eventually omitted due to poor performance on the pilot test actually had fixable problems that simply had not been diagnosed. This limitation could possibly have been mitigated by providing more detailed instruction or more frequent probing to those students who verbalized little during the interviews.

Synthesizing Data across Research Steps: An Exemplar

An advantage of employing several methodologies during inventory development was the ability to capture a variety of data. The interviews, for example, were particularly useful to reveal student thought processes that would be difficult to discern without a qualitative methodology and in greater detail than was achievable with the SSR surveys. Although the process was linear, data interpretation was iterative, as data from a subsequent step informed or prompted reconsideration of previous findings. Throughout the process, concepts were apt to be deconstructed or recast and items were repeatedly revised to improve their ability to tap underlying knowledge. An exemplar is provided to illustrate the circular process. Items in this discussion are referenced by their number on the beta version (Appendix L).
Because concept inventories are designed to measure conceptual understanding, items should be written so that students are unlikely to select the correct answer using memorized information. One item (Q50) depicted a pedigree and asked students to predict recurrence risk for an autosomal dominant (AD) condition. Thirteen of the 15 students who answered this question during cognitive interviewing correctly predicted the risk to be 50%, but only five of those students arrived at that answer by applying a correct rationale. Two students guessed, and six others ultimately chose their answer based on memory. Those six students associated a 50% recurrence risk with AD conditions and selected that answer despite drawing a genotype or a Punnett square which, if correctly interpreted, would give different answers. Those six students, reflecting 40% of the students interviewed, demonstrated knowledge of the concept despite lack of conceptual understanding. The difference is germane: Concept inventories are designed to test conceptual understanding, not rote knowledge. Cognitive interviews revealed that Q50 failed in that regard. On pilot testing, Q50 demonstrated fair psychometric properties (p = .64, r_{pb} = .31). However, on rational grounds the item is likely unable to differentiate between students who do and do not understand key concepts associated with inheritance.

Part of the problem may be that Q50 is a synthesis question in which multiple concepts are embedded. A basic tenet of concept inventory design is that each item should test a single concept; however, concepts are not necessarily indivisible. Understanding the concept of AD inheritance, for example, requires knowledge about three other concepts: autosomes, dominance, and homo- and heterozygosity. In the words of Michael (2007), the concept *autosomal dominance* “unpacks” into the three aforementioned concepts. Each supporting concept was addressed individually in other inventory items. An item about dominance (Q18) and one about autosomal disorders (Q20) were robust on pilot testing. However, two items about zygosity (Q23
and Q49) performed poorly. Q23 assessed understanding of the term *heterozygous* and was very difficult although undiscriminating (p = .21, r_{pb} = -.01). Very difficult questions are often not very discriminating (Kaplan & Saccuzzo, 1997) but the psychometric features of Q23 were particularly unfortunate. Interestingly, the difficulty of Q23 indicated many students did not understand the term *heterozygous*, a lexical problem that eluded detection during cognitive interviewing. The other zygosity question, Q49, assessed knowledge that people with an AD condition are usually heterozygous for the condition. Q49 presumed understanding of the terms *homo-* and *heterozygous*, and, although it performed better than Q23, it was not robust (p = .30, r_{pb} = .17). In fact, Q23 had only three distractors, and its difficulty was similar to that expected if students had simply guessed. Of interest, Q23 and Q49 were negatively correlated with each other at -.086, suggesting that knowledge of zygosity *per se* was not associated with knowledge that AD disorders are usually heterozygous.

Because the concept of zygosity is important to the content domain, the performance of Q23 and Q49 was concerning. For the GNCI beta version, both zygosity questions and the synthesis question (Q50) were retained, although the zygosity questions were significantly revised. Wage et al. (2005) describe similar difficulty interpreting linked questions and synthesis questions when developing the Signals and Systems Concept Inventory.

The above example illustrates how data from all four steps of the inventory development process were assimilated to make decisions about item retention and revision. It is hoped that further testing will clarify the difficulty with these items, and it is anticipated that Q50, which incorporates multiple concepts, will be eliminated. Eliminating Q50 seems particularly fitting upon looking back to the expert survey data. Although *characteristics and mechanisms of*
common inheritance patterns for single gene disorders was ranked fourth of 65 concepts, predicting recurrence risk for single-gene disorders was ranked 27th.

Next Steps and Potential Applications

“Constructing a concept inventory is a multi-year process involving a team of professionals with expertise in both teaching the subject and educational assessment” (Richardson, 2004). With that in mind, the output of this research, an empirically-derived, 31-item inventory, represents only a first step in developing an assessment capable of measuring what nurses need most to know about genetics and genomics. Further validation and testing are planned and are briefly described.

Both the content domain and the construct validity of the GNCI beta version need to be validated with genetic nurse experts. Validation with nursing faculty who are ISONG members using the Delphi method is anticipated. Selection of appropriate participants is crucial to Delphi methodology (Nelson et al., 2007), and because the content domain in genetic and genomic nursing is broad, it will be important to sample the range of expertise among nursing experts. Further validation is important to ensure the concepts included in the GNCI content domain are critical to nursing and that each inventory item tests the concept it was designed to measure. The method will be adapted from that used by Nelson et al. to create the Thermal and Transport Concept Inventory and that used by Bowling et al. (2008) to create the Genetic Literacy Assessment Instrument.

Cognitive interviews will be repeated with the 31 items on the GNCI beta version. Because items were revised throughout the cognitive interviews, only half of retained items were pretested with 15 students. One item, Q8, was imported from another concept inventory (Sadler, 2003) and is untested among nursing students. These interviews may be conducted individually.
or as focus groups, following the method of Bowling (2007). She administered her concept inventory to groups of students using a personal response system (electronic clicker). After students answered each item, the question and the response options were discussed and students were probed with questions.

The GNCI will also be field tested among students in baccalaureate nursing programs across the country as a paper-and-pencil test using Scantron technology. Nurse educators will be recruited to test the inventory with their students. According to Nunnally (1978), the minimum acceptable sample for a full-scale field test is five to ten times as many subjects as test items, which for the GNCI would be 150 to 300 students. A much larger sample may be realistic, however, and would be useful to support exploratory factor analysis to examine the relationship between inventory items, constructs, and the entire content domain.

As a later step, tests of discriminant validity will be necessary to measure the degree to which the GNCI can distinguish between nursing students who have achieved genomic literacy and those who have not. This would entail a test of contrasted-groups construct validity testing. The same methodology could be applied to nursing graduates working in a variety of settings where genomic literacy is essential. It can be hypothesized that students or working nurses with high genomic literacy would score higher on the GNCI than those with low literacy. A confirmation of this hypothesis would add evidence for the construct validity of the GNCI.

The Genomic Nursing Concept Inventory holds great promise to support genomic nursing education by providing a reliable and valid ruler by which to measure genomic literacy. Like other concept inventories, it may be used as a pre- and post-test for a course or for a curriculum, to measure individual, course, or program outcomes. The GNCI has immediate utility for baccalaureate nursing programs, and it is reasonable to validate it with students in
other nursing programs. The *Essentials* were written for all professional nurses, regardless of academic preparation, which means the GNCI might logically be utilized with associate degree and diploma nursing students, graduate nursing students, practicing nurses and even nursing faculty. The genome era of health care began eight years ago; nurses can wait no longer to achieve genomic literacy. The hope is that this tool will not only support that effort, but make the outcome measurable.
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MEMORANDUM

TO: Melvin Haberman and Linda Ward,

FROM: Malathi Jandhyala, Office of Research Assurances (3005)

DATE: 7/8/2010

SUBJECT: Certification of Exemption, IRB Number 11490

Based on the Exemption Determination Application submitted for the study titled "Development of the Genomic Nursing Concept Inventory," and assigned IRB # 11490, the WSU Office of Research Assurances has determined that the study satisfies the criteria for Exempt Research at 45 CFR 46.101(b)(2).

This study may be conducted according to the protocol described in the Application without further review by the IRB.

It is important to note that certification of exemption is NOT approval by the IRB. You may not include the statement that the WSU IRB has reviewed and approved the study for human subject participation. Remove all statements of IRB Approval and IRB contact information from study materials that will be disseminated to participants.

This certification is valid only for the study protocol as it was submitted to the ORA. Studies certified as Exempt are not subject to continuing review (this Certification does not expire). If any changes are made to the study protocol, you must submit the changes to the ORA for determination that the study remains Exempt before implementing the changes (The Request for Amendment form is available online at http://www.irb.wsu.edu/documents/forms/rtf/Amendment_Request.rtf).

Exempt certification does NOT relieve the investigator from the responsibility of providing continuing attention to protection of human subjects participating in the study and adherence to ethical standards for research involving human participants.

In accordance with WSU Business Policies and Procedures Manual (BPPM), this Certification of Exemption, a copy of the Exemption Determination Application identified by this certification
and all materials related to data collection, analysis or reporting must be retained by the Principal Investigator for THREE (3) years following completion of the project (BPPM 90.01). Washington State University is covered under Human Subjects Assurance Number FWA00002946 which is on file with the Office for Human Research Protections (OHRP).

Review Type: New
Review Category: Exempt
Date Received: 7/7/2010
Exemption Category: 45 CFR 46.101 (b)(2)
OGRD No.: N/A
Funding Agency: N/A
### Appendix B
Step One: Initial Concept List for Expert Survey
65 concepts in 14 topical categories

<table>
<thead>
<tr>
<th>Initial Category</th>
<th>Initial Label</th>
<th>Concept description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Family history</td>
<td>FAMHXA</td>
<td>The role of a 3-generation family health history in health assessment and risk assessment</td>
</tr>
<tr>
<td></td>
<td>FAMHXB</td>
<td>Essentials of interpreting a genetic pedigree for single gene and multifactorial conditions (e.g., degree of relatedness, matrilinear/patrilinear inheritance)</td>
</tr>
<tr>
<td>Genomic healthcare</td>
<td>GENCARA</td>
<td>The genomic (genetic + environmental) basis for virtually all diseases and health conditions</td>
</tr>
<tr>
<td></td>
<td>GENCARB</td>
<td>Concept of personalized care based on an individual’s genomic profile as a hope of genomic healthcare</td>
</tr>
<tr>
<td></td>
<td>GENCARC</td>
<td>Mechanisms by which targeted interventions such as gene-based therapies, protein replacement and chaperone therapy treat genetic disease</td>
</tr>
<tr>
<td></td>
<td>GENCARD</td>
<td>Essentials of gene replacement therapy (indications, mechanisms, safety issues)</td>
</tr>
<tr>
<td></td>
<td>GENCARE</td>
<td>Genetic basis of cancer</td>
</tr>
<tr>
<td></td>
<td>GENCARF</td>
<td>Role of pharmacogenomics in personalized pharmacotherapy</td>
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<td></td>
<td>GENCARG</td>
<td>Role of lifestyle changes in modulating genetic risk for common diseases (e.g., diabetes and heart disease); issues of implementation</td>
</tr>
<tr>
<td></td>
<td>GENCARH</td>
<td>Risk of harm associated with genetic information and the need for safeguards against those harms</td>
</tr>
<tr>
<td></td>
<td>GENCARI</td>
<td>The role of epigenetic modifications in health and illness (e.g., as mechanism for environmental effects on health)</td>
</tr>
<tr>
<td>Genotype-phenotype</td>
<td>GENPHEA</td>
<td>Reduced penetrance (i.e., some individuals who inherit the altered gene will not express the disorder)</td>
</tr>
<tr>
<td>association</td>
<td>GENPHEB</td>
<td>Variable expression (i.e., individuals with the same genotype have variable degrees of severity)</td>
</tr>
<tr>
<td>Human genome basics</td>
<td>HUMGENA</td>
<td>Human genome basics: size, organization, coding/noncoding DNA and the limitations of current understanding, relationship between genome, chromosome, gene, and nucleotide</td>
</tr>
<tr>
<td></td>
<td>HUMGENB</td>
<td>DNA basics: structure, functions, mechanism of replication, transcription and translation, trinucleotides and codons</td>
</tr>
<tr>
<td></td>
<td>HUMGENC</td>
<td>DNA and human variation: degree of human genetic homogeneity, scope of genetic variation (from polymorphisms to aneuploidies), genotype/phenotype</td>
</tr>
<tr>
<td>HUMGEND</td>
<td>Mechanism and effects of evolution (random events subject to environmental selection; source of biologic variation)</td>
<td></td>
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<td>-----------------------</td>
<td>---------------------------------------------------------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td>HUMGENE</td>
<td>Basics of comparative genomics (shared genes among species)</td>
<td></td>
</tr>
<tr>
<td><strong>Inheritance patterns</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>INHA</td>
<td>Single gene inheritance patterns: autosomal dominant, autosomal recessive, X-linked, mitochondrial</td>
<td></td>
</tr>
<tr>
<td>INHB</td>
<td>Predicting recurrence risk for single-gene disorders</td>
<td></td>
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<tr>
<td>INHC</td>
<td>Characteristics and mechanisms of multifactorial inheritance</td>
<td></td>
</tr>
<tr>
<td>INHD</td>
<td>Issues in predicting recurrence risk for multifactorial disorders (modest effects, relative risk, environmental effects)</td>
<td></td>
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<tr>
<td><strong>Mutations</strong></td>
<td></td>
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<tr>
<td>MUTA</td>
<td>Specific genetic alterations (polymorphisms, mutations, repeats, inversions/deletions/translocations, copy number variation, aneuploidies)</td>
<td></td>
</tr>
<tr>
<td>MUTB</td>
<td>Germline versus somatic mutations</td>
<td></td>
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<tr>
<td>MUTC</td>
<td>Frameshift mutations</td>
<td></td>
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<tr>
<td>MUTD</td>
<td>Mechanism by which genetic alterations are expressed (disruption of protein production)</td>
<td></td>
</tr>
<tr>
<td>MUTE</td>
<td>That a genetic condition may be caused by any of multiple mutations; implications for genetic testing (e.g., cystic fibrosis, hereditary breast/ovarian cancer)</td>
<td></td>
</tr>
<tr>
<td><strong>Nature of a gene</strong></td>
<td></td>
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<tr>
<td>NATA</td>
<td>Gene basics: structure, function, size, locus, dominant/recessive alleles, homo- and heterozygosity</td>
<td></td>
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<tr>
<td>NATB</td>
<td>Introns and exons, alternative mRNA splicing to form &gt;1 protein/gene</td>
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<tr>
<td>NATC</td>
<td>Gene regulation: expression, silencing, role of promoters, tissue-specific and developmental effects</td>
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<tr>
<td>NATD</td>
<td>Gene imprinting</td>
<td></td>
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<tr>
<td>NATE</td>
<td>Epigenetic mechanisms of gene regulation (DNA methylation, histone modification)</td>
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<tr>
<td><strong>Pharmacogenomics</strong></td>
<td></td>
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<tr>
<td>PGXA</td>
<td>Genetic basis for human variation in drug response (roles of proteins in pharmacokinetics and pharmacodynamics)</td>
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<tr>
<td>PGXB</td>
<td>Pharmacogenomic effects on drug metabolism (e.g., CYP450 rapid/poor metabolizers)</td>
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<tr>
<td>PGXC</td>
<td>Pharmacogenetics effects on drug effectiveness d/t pharmacodynamic variation (e.g., altered drug receptors)</td>
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<tr>
<td>PGXD</td>
<td>Essentials of pharmacogenetic testing and personalized prescribing</td>
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<td><strong>Proteins</strong></td>
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<td>PROTA</td>
<td>Essential roles of proteins (enzymes, receptors, transporters, etc.) for physiologic function</td>
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<tr>
<td>PROTB</td>
<td>Central dogma and relationship between DNA sequence, amino acid sequence, protein folding and protein function</td>
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<tr>
<td>PROTC</td>
<td>Effects of DNA variations on protein production, structure, and function (e.g., nonsense mutation causing truncated protein)</td>
<td></td>
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<tr>
<td>Genetics and race</td>
<td>RACEA</td>
<td>Limitations and implications of race as a concept (e.g., lack of scientific basis, potential for marginalization, utility in tracking health disparities)</td>
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<tr>
<td></td>
<td>RACEB</td>
<td>Health-related implications of ancestral or geographic origins (effect on frequencies of certain genetic variations, e.g., sickle cell disease)</td>
</tr>
<tr>
<td>Genetics and risk assessment</td>
<td>RISKASMA</td>
<td>Identifying genetic ‘red flags’ (e.g., early age of onset, multiple cases of a condition in a family) from a genetic pedigree</td>
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<td></td>
<td>RISKASMB</td>
<td>Predicting recurrence risk for single gene disorders</td>
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<td></td>
<td>RISKASMC</td>
<td>Stratifying risk for common chronic disease based on family health history</td>
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<td></td>
<td>RISKASMD</td>
<td>Use of risk assessment to inform plan of care (lifestyle modifications, screening, pharmacoprevention)</td>
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<tr>
<td>Value of genetic tests</td>
<td>TCHARA</td>
<td>Clinical utility of (genetic) tests</td>
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<tr>
<td></td>
<td>TCHARB</td>
<td>Clinical validity of (genetic) tests</td>
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<td></td>
<td>TCHARC</td>
<td>Analytic validity considerations of (genetic) tests</td>
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<td>TCHARD</td>
<td>Sensitivity of (genetic) tests</td>
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<td>TCHARE</td>
<td>Specificity of (genetic) tests</td>
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<td>TCHARG</td>
<td>Positive and negative predictive value of (genetic) tests</td>
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<td></td>
<td>TCHARGF</td>
<td>Cost-benefit considerations of (genetic) tests</td>
</tr>
<tr>
<td>Issues in genetic testing</td>
<td>TESTA</td>
<td>Features of a genetic test (related to purpose of test, may examine DNA, RNA, gene products or metabolites)</td>
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<td></td>
<td>TESTB</td>
<td>Multiple purposes of genetic testing, both related and unrelated to health (e.g., ancestry, forensics)</td>
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<td></td>
<td>TESTC</td>
<td>Testing for germline versus somatic variations (i.e., blood sample or buccal swab vs tumor)</td>
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<td></td>
<td>TESTD</td>
<td>Issues around direct-to-consumer genetic testing (regulation, interpretation, potential benefits/harms)</td>
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<td>TESTE</td>
<td>Issues in DNA sequencing tests (e.g., targeted vs. whole genome; reflects gene structure but not gene expression)</td>
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<td>TESTF</td>
<td>Issues in RNA or protein testing (reflects gene expression, tissue specific, useful in tumor profiling)</td>
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<td>TESTG</td>
<td>Issues of privacy related to genetic testing</td>
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<td>TESTH</td>
<td>Issues of genetic exceptionalism (e.g., whether genetic tests have special ethical, legal and social implications that are different from other types of laboratory tests)</td>
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<tr>
<td>Types of genetic tests</td>
<td>TTYPESA</td>
<td>Applications and implications of genetic/genomic screening tests</td>
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<td></td>
<td>TTYPESB</td>
<td>Applications and implications of genetic/genomic diagnostic tests</td>
</tr>
<tr>
<td></td>
<td>TTYPESC</td>
<td>Applications and implications of predictive genetic/genomic tests</td>
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<tr>
<td></td>
<td>TTYPESD</td>
<td>Applications and implications of carrier status tests</td>
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<td></td>
<td>TTYPESE</td>
<td>Applications and implications of preimplantation genetic testing</td>
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Appendix C
Step One: Expert Survey of Genomic Nursing Concepts

Genomic Nursing Concepts: A survey designed to define Genetic and Genomic Concepts most essential for nurse to understand.

Consent and Demographics: Please read the survey description document and then answer the following Demographic questions:

1. Consent to participate: Please open and read the pdf below. Click “Yes” to acknowledge that you have read this document and that I may use the information you provide for inventory development and presentations or publications about the development process.

   ![Genomic Nursing Concepts Survey Description.pdf](Genomic Nursing Concepts Survey Description.pdf)

   - Yes – I have read the above statement. I agree to complete the survey, and I give permission for inclusion of my responses for inventory development and related presentations and publications.

2. For how long have you been involved in genetic/genomic nursing? Please check one:

   - Less than one year
   - One to two years
   - Two to five years
   - Five to ten years
   - Longer than ten years

3. How would you describe your role in genetic/genomic nursing? Please check all that apply:

   - Consensus panel member
   - Nurse educator or faculty member
   - Registered nurse in clinical practice
   - Advanced practice nurse in clinical practice
   - Clinical researcher
   - Laboratory researcher
   - Other (text box)

4. If you checked “Other” in response to question 2 above, please describe your role in genetic/genomic nursing below. (255 character limit)
Please rate the relevance of each of these “HUMAN GENOME” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Human genome basics: size, organization, coding/noncoding DNA and the limitations of current understanding, relationship between genome, chromosome, gene, and nucleotide

2. DNA basics: structure, functions, mechanism of replication, transcription and translation, trinucleotides and codons

3. DNA and human variation: degree of human genetic homogeneity, scope of genetic variation (from polymorphisms to aneuploidies), genotype/phenotype

4. Mechanism and effects of evolution (random events subject to environmental selection; source of biologic variation)

5. Basics of comparative genomics (shared genes among species)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

6. Additional concepts: Are there additional “HUMAN GENOME” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

7. Comments: Do you have any comments about any of the “HUMAN GENOME” concepts listed above?

   Text Box
Please rate the relevance of each of these “NATURE OF A GENE” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Gene basics: structure, function, size, locus, dominant/recessive alleles, homo- and heterozygosity

2. Introns and exons, alternative mRNA splicing to form >1 protein/gene

3. Gene regulation: expression, silencing, role of promoters, tissue-specific and developmental effects

4. Gene imprinting

5. Epigenetic mechanisms of gene regulation (DNA methylation, histone modification)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

6. Additional concepts: Are there additional “NATURE OF A GENE” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

7. Comments: Do you have any comments about any of the “NATURE OF A GENE” concepts listed above?

   Text Box
Please rate the relevance of each of these “MUTATIONS” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Specific genetic alterations (polymorphisms, mutations, repeats, inversions/deletions/translocations, copy number variation, aneuploidies)

2. Germline versus somatic mutations

3. Frameshift mutations

4. Mechanism by which genetic alterations are expressed (disruption of protein production)

5. That a genetic condition may be caused by any of multiple mutations; implications for genetic testing (e.g., cystic fibrosis, hereditary breast/ovarian cancer)

   o Critical to Know
   o Important to Know
   o Nice to Know
   o Not Important
   o No opinion

6. Additional concepts: Are there additional “MUTATIONS” concepts that you believe should be included as either “Critical to Know” or “Important to Know” that are not listed above?  

   Text Box

7. Comments: Do you have any comments about any of the “MUTATIONS” concepts listed above?  

   Text Box
Please rate the relevance of each of these “PROTEINS” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Essential roles of proteins (enzymes, receptors, transporters, etc.) for physiologic function

2. Central dogma and relationship between DNA sequence and protein function:
   DNA sequence → amino acid sequence → protein folding → physiologic function.

   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

3. Additional concepts: Are there additional “PROTEINS” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   [Text Box]

4. Comments: Do you have any comments about any of the “PROTEINS” concepts listed above?

   [Text Box]
Please rate the relevance of each of these “PHARMACOGENOMICS” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Genetic basis for human variation in drug response (roles of proteins in pharmacokinetics and pharmacodynamics)

2. Pharmacogenetics effects on drug metabolism (e.g., CYP450 rapid/poor metabolizers)

3. Pharmacogenomic effects on drug effectiveness d/t pharmacodynamics variation (e.g., altered drug receptors)

4. Essentials of pharmacognetic testing and personalized prescribing
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

5. Additional concepts: Are there additional “PHARMACOGENOMICS” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

6. Comments: Do you have any comments about any of the “PHARMACOGENOMICS” concepts listed above?

   Text Box
Please rate the relevance of each of these “FAMILY HISTORY” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. The role of a 3-generation family health history in health assessment and risk assessment

2. Essentials of interpreting a genetic pedigree for single gene and multifactorial conditions (e.g., degree of relatedness, matrilinear/patrilinear inheritance)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

3. Additional concepts: Are there additional “FAMILY HISTORY” concepts that you believe should be included as either “Critical to Know” or “Important to Know” that are not listed above?

4. Comments: Do you have any comments about any of the “FAMILY HISTORY” concepts listed above?
Please rate the relevance of each of these “RACE/ETHNICITY” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Limitations and implications of race as a concept (e.g., lack of scientific basis, potential for marginalization, utility in tracking health disparities).

2. Health-related implications of ancestral or geographic origins (effect on frequencies of certain genetic variations, e.g., sickle cell disease)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

3. Additional concepts: Are there additional “RACE/ETHNICITY” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

4. Comments: Do you have any comments about any of the “RACE/ETHNICITY” concepts listed above?

   Text Box
Please rate the relevance of each of these “INHERITANCE PATTERNS” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Characteristics and mechanisms of common inheritance patterns for single gene disorders: autosomal dominant, autosomal recessive, X-linked, mitochondrial

2. Predicting recurrence risk for single-gene disorders

3. Characteristics and mechanisms of multifactorial inheritance.

4. Issues in predicting recurrence risk for multifactorial disorders (modest effects, relative risk, environmental effects)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

5. Additional concepts: Are there additional “INHERITANCE PATTERNS” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

6. Comments: Do you have any comments about any of the “INHERITANCE PATTERNS” concepts listed above?

   Text Box
Please rate the relevance of each of these “GENOTYPE/PHENOTYPE ASSOCIATION” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Reduced penetrance (i.e., some individuals who inherit the altered gene will not express the disorder)

2. Variable expression (i.e., individuals with the same genotype have variable degrees of severity)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

3. Additional concepts: Are there additional “GENOTYPE/PHENOTYPE ASSOCIATION” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

4. Comments: Do you have any comments about any of the “GENOTYPE/PHENOTYPE ASSOCIATION” concepts listed above?

   Text Box
Please rate the relevance of each of these “GENETIC RISK ASSESSMENT” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Identifying genetic ‘red flags’ (e.g., early age of onset, multiple cases of a condition in a family) from a genetic pedigree

2. Predicting recurrence risk for single gene disorders

3. Stratifying risk for common chronic disease based on family health history

4. Use of risk assessment to inform plan of care (lifestyle modifications, screening, pharmacoprevention)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

5. Additional concepts: Are there additional “GENETIC RISK ASSESSMENT” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

6. Comments: Do you have any comments about any of the “GENETIC RISK ASSESSMENT” concepts listed above?

   Text Box
Please rate the relevance of each of these “GENETIC TESTING” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. Features of a genetic test (related to purpose of test, may examine DNA, RNA, gene products or metabolites)

2. Multiple purposes of genetic testing, both related and unrelated to health (e.g., ancestry, forensics)

3. Testing for germline versus somatic variations (i.e., blood sample or buccal swab vs tumor)

4. Issues in DNA sequencing tests (e.g., targeted vs. whole genome; reflects gene structure but not gene expression)

5. Issues in RNA or protein testing (reflects gene expression, tissue specific, useful in tumor profiling)

6. Issues of privacy related to genetic testing

7. Issues of genetic exceptionalism (e.g., whether genetic tests have special ethical, legal and social implications that are different from other types of laboratory tests)

8. Issues around direct-to-consumer genetic testing (regulation, interpretation, potential benefits/harms)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

9. Additional concepts: Are there additional “GENETIC TESTING” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

   Text Box

10. Comments: Do you have any comments about any of the “GENETIC TESTING” concepts listed above?

    Text Box
Please rate the relevance of each of these “GENOMIC HEALTHCARE” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

1. That virtually all diseases and health conditions have a genomic (genetic + environmental) basis

2. Personalized healthcare based on an individual’s genomic profile as a goal of genomic healthcare

3. Mechanisms by which targeted gene-based therapies (such as protein replacement and chaperone therapy) treat disease

4. Essentials of gene replacement therapy (indications, mechanisms, safety issues)

5. Genetic basis of cancer

6. Role of pharmacogenomics in personalized pharmacotherapy

7. Role of lifestyle changes in modulating genetic risk for common diseases (e.g., diabetes and heart disease); issues of implementation

8. Risk of harm associated with genetic information and the need for safeguards against those harms

9. The role of epigenetic modifications in health and illness (e.g., as mechanism for environmental effects on health)
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion

10. Additional concepts: Are there additional “GENOMIC HEALTHCARE” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

    Text Box

11. Comments: Do you have any comments about any of the “GENOMIC HEALTHCARE” concepts listed above?

    Text Box
Please rate the relevance of each of these “GENETIC TEST CHARACTERISTICS” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

Genetic Test Characteristics: The value of a genetic test is often assessed by examining specific characteristics of the test. How important is it for nurses to understand each of the following characteristics?

1. Clinical utility
2. Clinical validity
3. Analytic validity
4. Sensitivity
5. Specificity
6. Positive predictive value
7. Cost-benefit considerations
   - Critical to Know
   - Important to Know
   - Nice to Know
   - Not Important
   - No opinion
8. Additional concepts: Are there additional “GENETIC TEST CHARACTERISTICS” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?
9. Comments: Do you have any comments about any of the “GENETIC TEST CHARACTERISTICS” concepts listed above?
Please rate the relevance of each of these “TYPES OF GENETIC TESTS” concepts to nursing education and practice at the basic baccalaureate level. You may suggest “Additional Concepts” and/or add “Comments” at the end of this section.

Health-related genetic tests may be performed for a variety of purposes; each type of testing has specific applications and implications. How important is it for nurses to understand applications and implications of the following categories of genetic testing?

1. Screening
2. Diagnostic testing (e.g., Fragile X, tumor profiling)
3. Predictive testing
4. Carrier status testing
5. Preimplantation testing
   o Critical to Know
   o Important to Know
   o Nice to Know
   o Not Important
   o No opinion
6. Additional concepts: Are there additional “TYPES OF GENETIC TESTS” concepts that you believe should be included as either “Critical to Know” or ”Important to Know” that are not listed above?

Text Box

7. Comments: Do you have any comments about any of the “TYPES OF GENETIC TESTS” concepts listed above?

Text Box

End of Survey
Thank you for your participation!
Appendix D
Step One: Invitation to Participate in Expert Survey

Dear ____________,

You have been identified as a nurse with experience and expertise in genetics and genomics. I seek your help in defining genetic and genomic literacy for nursing by creating a prioritized list of concepts nurses must understand in order to achieve genetic and genomic competency. This information will be used as a part of my doctoral dissertation as I am developing a tool to assess genetic and genomic knowledge among baccalaureate nursing students.

Please find below your personalized License, User ID and Password and survey link so that you can access the Genetic Nursing Concepts online survey which lists fundamental concepts about genetics and genomics. These concepts have been derived from the essential competencies and reflect a nursing perspective. During the survey, you will be asked to categorize each concept according to its relevance to nursing practice. You will also be asked two questions about your professional role. Responses from this survey will be collated to identify the concepts most salient to genetic/genomic nursing, which will provide the knowledge domain for the inventory. The survey will take approximately 10-15 minutes.

Your personalized License, User ID and Password:

License:       gnc01
User ID:       101
Password:      password

Survey Link: You can access the Genetic Nursing Concepts online survey in three easy steps:

Step 1: Click this link: Genetic Nursing Concepts & enter your License, User ID & Password.
Step 2: Click on your personalized User ID (101) located to the right of the Calendar.
Step 3: Click on the Genetic Nursing Concepts link located on the right side of your screen & begin the survey!

For additional questions about this survey or about my dissertation project, please contact me, Linda Ward, linward@wsu.edu, 509 324-7450. For technical assistance, please contact Yvonne Smith, Research Assistant, Washington State University, College of Nursing, ytsmith@wsu.edu, 509.324.7328.

Thank you in advance for your assistance!

Linda D. Ward, MN, ARNP
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Clinical Assistant Professor
Washington State University, College of Nursing
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Spokane, WA 99202-1495
linward@wsu.edu
509 324-7450 (phone) * 509 324-7341 (fax)
Appendix E
Step One: Attachment to Invitation to Participate--Additional Information about GNCI Survey

Genetic/genomic literacy . . . . What do nurses need to know?

Dear Survey Respondent,

Survey overview and purpose: This survey is an initial step in the development of an educational assessment to measure genetic and genomic knowledge among baccalaureate nursing students. The essential nursing competencies for genetics and genomics provide the benchmark. A critical first step in the development of this tool is to define the knowledge most essential for nurses to understand. This foundational knowledge might be considered genomic literacy for nurses—necessary but not sufficient for the achievement of genomic competency.

You will be asked to prioritize a list of concepts extracted from the Essentials, based on their relevance to basic (not advanced) nursing practice. You will also have an opportunity to provide comments and suggest additional concepts that you believe are essential for nurses to understand. The information from this survey will be compiled to identify the knowledge domain for the concept inventory. Responses may be summarized and reported in publications and professional presentations. The survey will take approximately 15 minutes.

Privacy: The survey includes two questions about your experience and role in genetic nursing. All responses are confidential, identified by a research ID number only for purposes of tracking and survey follow up. Responses will be fully de-identified for analysis and stored within secured data centers with multiple layers of protection. This survey utilizes SurveyBooth™, which provides data confidentiality and security. All data will be analyzed and reported in aggregate. No identifying information about you will be reported in presentations or publications.

Consent to participate: By clicking “yes” to question 1 in the survey, you acknowledge that you have read this statement and that I may use the information you provide for tool development and presentations or publication about the development process.

Thank you again for your participation!

Linda D. Ward, PhD(c), ARNP
Clinical Assistant Professor
Washington State University College of Nursing
103 E Spokane Blvd * PO Box 1495
Spokane, WA 99202-1495
linward@wsu.edu
509 324-7450
Appendix F  
Step One: Follow-up Invitation to Survey Nonrespondents

Dear ________________,

In early August, you were sent an e-mail invitation to participate in a survey to identify the genetic and genomic concepts most important for nurses to understand. To date, we have not received your response. Data from this survey will be used to develop a tool to measure genetic and genomic knowledge among baccalaureate nursing students. Your assistance in identifying key concepts to include in the tool is critically important.

If you have already responded to this survey, thank you for your participation. **If you have not responded, will you please take the time to participate?** Your opinion is important to achieve consensus on what the nursing workforce needs to know about genetics and genomics.

Please find below your personalized **License, User ID and Password** and survey link so that you can access the Genetic Nursing Concepts online survey which lists fundamental concepts about genetics and genomics. These concepts have been derived from the essential competencies and reflect a nursing perspective. During the survey, you will be asked to categorize each concept according to its relevance to nursing practice. You will also be asked two questions about your professional role. Responses from this survey will be collated to identify the concepts most salient to genetic/genomic nursing, which will provide the knowledge domain for the inventory. The survey will take approximately **10-15 minutes**.

**Your personalized License, User ID and Password:**

License: gnc01  
User ID: 101  
Password: password

**Survey Link:** You can access the Genetic Nursing Concepts online survey in three easy steps:

Step 1: Click this link: Genetic Nursing Concepts & enter your License, User ID & Password.
Step 2: Click on your personalized User ID (101) located to the right of the Calendar.
Step 3: Click on the Genetic Nursing Concepts link located on the right side of your screen & begin the survey.

**For additional questions** about this survey or about my dissertation project, please contact me, Linda Ward, linward@wsu.edu, 509 324-7450. **For technical assistance**, please contact Yvonne Smith, Research Assistant, Washington State University, College of Nursing, ytsmith@wsu.edu, 509.324.7328.

**Thank you in advance for your assistance!**

Linda D. Ward, MN, ARNP  
PhD in Nursing Candidate, Clinical Assistant Professor  
Washington State University College of Nursing  
103 E Spokane Blvd, PO Box 1495  
Spokane, WA 99202-1495  
linward@wsu.edu  509 324-7450 (phone)  509 324-7341 (fax)
Appendix G
Step Three: Consent Form for GNCI Cognitive Interview

WASHINGTON STATE UNIVERSITY
College of Nursing
Research Study Consent Form

Study Title: Development of the Genomic Nursing Concept Inventory

Researchers:
Principle investigator: Mel Haberman, PhD
Professor, WSU College of Nursing
509 324-7358

Co-investigator: Linda Ward, MN
PhD candidate and Clinical Assistant Professor
WSU College of Nursing
509 324-7450

You are being asked to take part in a research study carried out by Mel Haberman, PhD, and Linda Ward, MN. This form explains the research study and your part in it if you decide to join the study. Please read the form carefully, taking as much time as you need. Ask the researcher to explain anything you don’t understand. You can decide not to join the study. If you join the study, you can change your mind later or quit at any time. There will be no penalty or loss of services or benefits if you decide to not take part in the study or quit later.

What is this study about?

This research study is being done to develop a concept inventory, which is a special kind of test designed to measure knowledge of genetic and genomic concepts among nursing students. You are being asked to take part because you are a nursing student in your first year of a baccalaureate program. Taking part in the study will take about 60 to 90 minutes. You cannot take part in this study if you have completed more than two semesters of a baccalaureate nursing program.

What will I be asked to do if I am in this study?

If you take part in the study, you will be asked to participate in an interview called a cognitive think-aloud interview. You will be asked to complete a paper-and-pencil test with about 50 multiple-choice questions about genetics and genomics. The interview will be limited to the questions described above, and you will not be asked any personal questions. You will be asked to read each question aloud and describe your thought process as you consider each question and mark the best answer on the test form. You may decline to answer any question in the inventory. The interview will be audio-recorded. The interviewer will be taking notes throughout the interview and may ask questions about your thoughts as you consider a question. The interview is estimated to take 60 to 90 minutes.
Are there any benefits to me if I am in this study?

You may gain a better understanding of what nurses should know about genetics and genomics by participating in this study. In addition, your participation may contribute to the development of a useful tool to measure genetic and genomic knowledge.

Are there any risks to me if I am in this study?

The only risks from taking part in this study are the inconvenience of taking time to participate and possible mental strain associated with answering multiple knowledge-based questions.

Will my information be kept private?

The data for this study will be kept confidential to the extent allowed by federal and state law. Your responses will be used for statistical purposes only. Your paper test, the interviewer’s notes, and the voice recording will be coded with a numeric identifier. Only the interviewer will be able to link your name to your interview materials, and the log linking your name and numerical code will be stored separately from the interview materials. All interview materials will be stored in locked files. Only research collaborators will have access to your interview materials. No published results will identify you, and your name will not be associated with the findings. Under certain circumstances, information that identifies you may be released for internal and external reviews of this project. The results of this study may be published or presented at professional meetings, but the identities of all research participants will remain anonymous. The data for this study will be kept for three years.

Are there any costs or payments for being in this study?

There will be no costs to you for taking part in this study. You will receive $30 at the time you complete the interview. You will not receive money or any other form of compensation if you quit the interview before completing all questions.

Who can I talk to if I have questions?

If you have questions about this study or the information in this form, please contact the co-investigator, Linda Ward, or the primary investigator, Mel Haberman.

Ms. Ward  WSU College of Nursing
PO Box 1495
Spokane, WA 99210-1959
509 324-7450  linward@wsu.edu

Dr. Haberman  WSU College of Nursing
PO Box 1495
Spokane, WA 99210-1959
509 324-7358  haberman@wsu.edu.
If you have questions about your rights as a research participant, or would like to report a concern or complaint about this study, please contact the Washington State University Institutional Review Board at (509) 335-3668, or e-mail irb@wsu.edu, or regular mail at: Albright 205, PO Box 643005, Pullman, WA 99164-3005.

What are my rights as a research study volunteer?
Your participation in this research study is completely voluntary. You may choose not to be a part of this study. There will be no penalty to you if you choose not to take part. You may choose not to answer specific questions or to stop participating at any time.

What does my signature on this consent form mean?
Your signature on this form means that:
- You understand the information given to you in this form
- You have been able to ask the researcher questions and state any concerns
- The researcher has responded to your questions and concerns
- You believe you understand the research study and the potential benefits and risks that are involved.

Statement of Consent
I give my voluntary consent to take part in this study. I will be given a copy of this consent document for my records.

Signature of Participant  Printed Name of Participant  Date

Statement of Person Obtaining Informed Consent
I have carefully explained to the person taking part in the study what he or she can expect. I certify that when this person signs this form, to the best of my knowledge, he or she understands the purpose, procedures, potential benefits, and potential risks of participation. I also certify that he or she:
- Speaks the language used to explain this research
- Reads well enough to understand this form or, if not, this person is able to hear and understand when the form is read to him or her
- Does not have any problems that could make it hard to understand what it means to take part in this research.

Signature of Person Obtaining Consent  Date

Printed Name of Person Obtaining Consent  Role in the Research Study
### Appendix H

**Step One: Prioritized Concept List of 65 Items Rank Ordered by Mean Item Scores**

*N* = 99-104 Respondents

<table>
<thead>
<tr>
<th>Initial Label</th>
<th>N</th>
<th>Mean</th>
<th>SD</th>
<th>Concept description</th>
</tr>
</thead>
<tbody>
<tr>
<td>FAMHXA</td>
<td>103</td>
<td>3.9</td>
<td>.298</td>
<td>Use of a 3-generation family health history in health assessment and risk assessment</td>
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<tr>
<td>RISKASMA</td>
<td>102</td>
<td>3.83</td>
<td>.375</td>
<td>Identifying genetic ‘red flags’(e.g., early age of onset, multiple cases of a condition in a family)from a genetic pedigree</td>
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<tr>
<td>TESTG</td>
<td>101</td>
<td>3.82</td>
<td>.410</td>
<td>Issues of privacy related to genetic testing</td>
</tr>
<tr>
<td>INHA</td>
<td>102</td>
<td>3.78</td>
<td>.459</td>
<td>Characteristics and mechanisms of common inheritance patterns for single gene disorders: autosomal dominant, autosomal recessive, X-linked, mitochondrial</td>
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<tr>
<td>MUTE</td>
<td>101</td>
<td>3.78</td>
<td>.482</td>
<td>That a genetic condition may be caused by any of multiple mutations; implications for genetic testing (e.g., cystic fibrosis, hereditary breast/ovarian cancer)</td>
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<tr>
<td>FAMHXB</td>
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<td>.577</td>
<td>Interpreting a genetic pedigree for single gene and multifactorial conditions (e.g., degree of relatedness, matrilinear/patrilinear inheritance)</td>
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<tr>
<td>NATA</td>
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<td>3.65</td>
<td>.591</td>
<td>Gene basics: structure, function, size, locus, dominant/recessive alleles, homo- and heterozygosity</td>
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<tr>
<td>GENCARH</td>
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<td>3.61</td>
<td>.565</td>
<td>Risk of harm associated with genetic information and the need for safeguards against those harms</td>
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<td>TTYPESA</td>
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<td>PGXA</td>
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<td>.637</td>
<td>Genetic basis for human variation in drug response (roles of proteins in pharmacokinetics and pharmacodynamics)</td>
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<td>PGXB</td>
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<td>.640</td>
<td>Pharmacogenomic effects on drug metabolism (e.g., CYP450 rapid/poor metabolizers)</td>
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<td>GENCARG</td>
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<td>.576</td>
<td>Role of lifestyle changes in modulating genetic risk for common diseases (e.g., diabetes and heart disease); issues of implementation</td>
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<td>HUMGENA</td>
<td>104</td>
<td>3.52</td>
<td>.591</td>
<td>Human genome basics: size, organization, coding/noncoding DNA and the limits of current understanding, relationship between genome, chromosome, gene, and nucleotide</td>
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<td>MUTB</td>
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<td>3.51</td>
<td>.712</td>
<td>Germline versus somatic mutations</td>
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<td>GENCARA</td>
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<td>3.5</td>
<td>.626</td>
<td>The genomic (genetic + environmental) basis for virtually all diseases and health conditions</td>
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<td>COURSE NUMBER</td>
<td>COURSE ABBREVIATION</td>
<td>COURSE TITLE</td>
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<tr>
<td>RISKASMD</td>
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<td>3.5</td>
<td>Use of risk assessment to inform plan of care (lifestyle modifications, screening, pharmacoprevention)</td>
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<td>GENCARE</td>
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<td>3.48</td>
<td>Genetic basis of cancer</td>
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<td>HUMGENC</td>
<td>104</td>
<td>3.48</td>
<td>DNA and human variation: degree of human genetic homogeneity, scope of genetic variation (from polymorphisms to aneuploidies), genotype/phenotype</td>
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<td>DNA basics: structure, functions, mechanism of replication, transcription and translation, trinucleotides and codons</td>
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<td>TESTH</td>
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<td>Issues of genetic exceptionalism (e.g., whether genetic tests have special ethical, legal and social implications that are different from other types of laboratory tests)</td>
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<td>GENPHEA</td>
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<td>3.44</td>
<td>Reduced penetrance (i.e., some individuals who inherit the altered gene will not express the disorder)</td>
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<td>Variable expression (i.e., individuals with the same genotype have variable degrees of severity)</td>
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<td>MUTA</td>
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<td>Specific genetic alterations (polymorphisms, mutations, repeats, inversions/deletions/translocations, copy number variation, aneuploidies)</td>
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<td>INHB</td>
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<td>Predicting recurrence risk for single-gene disorders</td>
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<td>PROTA</td>
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<td>3.38</td>
<td>Essential roles of proteins (enzymes, receptors, transporters, etc.) for physiologic function</td>
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<td>TCHARA</td>
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<td>3.37</td>
<td>Clinical utility of (genetic) tests</td>
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<td>GENCARB</td>
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<td>3.36</td>
<td>Concept of personalized care based on an individual’s genomic profile as a hope of genomic healthcare</td>
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<td>Pharmacogenomics and personalized pharmacotherapy</td>
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<td>3.36</td>
<td>Characteristics and mechanisms of multifactorial inheritance</td>
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<tr>
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<td>3.36</td>
<td>Pharmacogenetics effects on drug effectiveness d/t pharmacodynamic variation (e.g., altered drug receptors)</td>
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<td>RACEB</td>
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<td>Health-related implications of ancestral or geographic origins (effect on frequencies of certain genetic variations, e.g., sickle cell disease)</td>
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<tr>
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<td>Clinical validity of (genetic) tests</td>
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<td>Issues around direct-to-consumer genetic testing (regulation, interpretation, potential benefits/harms)</td>
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<td>Mechanism by which genetic alterations are expressed (disruption of protein production)</td>
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<td>3.26</td>
<td>Limitations and implications of race as a concept (e.g., lack of scientific basis, potential for marginalization, utility in tracking health disparities)</td>
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<td>NATD</td>
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<td>.786</td>
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</table>

- **Essentials of pharmacogenetic testing and personalized prescribing**
- **Predicting recurrence risk for single gene disorders**
- **Specificity of (genetic) tests**
- **Sensitivity of (genetic) tests**
- **Stratifying risk for common chronic disease based on family health history**
- **Central dogma and relationship between DNA sequence, amino acid sequence, protein folding and protein function**
- **Effects of DNA variations on protein production, structure, and function (e.g., nonsense mutation causing truncated protein)**
- **The role of epigenetic modifications in health and illness (e.g., as mechanism for environmental effects on health)**
- **Features of a genetic test (related to purpose of test, may examine DNA, RNA, gene products or metabolites)**
- **Testing for germline versus somatic variations (i.e., blood sample or buccal swab vs. tumor)**
- **Positive and negative predictive value of (genetic) tests**
- **Gene regulation: expression, silencing, role of promoters, tissue-specific and developmental effects**
- **Issues in predicting recurrence risk for multifactorial disorders (modest effects, relative risk, environmental effects)**
- **Applications and implications of preimplantation genetic testing**
- **Multiple purposes of genetic testing, both related and unrelated to health (e.g., ancestry, forensics)**
- **Cost-benefit considerations of (genetic) tests**
- **Analytic validity considerations of (genetic) tests**
- **Mechanisms of targeted interventions such as gene-based therapies, protein replacement and chaperone therapy**
- **Frameshift mutations**
- **Issues in DNA sequencing tests (e.g., targeted vs. whole genome; reflects gene structure but not gene expression)**
- **Mechanism and effects of evolution (random events subject to environmental selection; source of biologic variation)**
- **Introns and exons, alternative mRNA splicing to form >1 protein/gene**
- **Gene imprinting**
<p>| | | | |</p>
<table>
<thead>
<tr>
<th></th>
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<tbody>
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<td>NATE</td>
<td>102</td>
<td>2.73</td>
<td>.914 Epigenetic mechanisms of gene regulation (DNA methylation, histone modification)</td>
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<td>.728 Essentials of gene replacement therapy</td>
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<td>2.59</td>
<td>.827 Issues in RNA or protein testing (reflects gene expression, tissue specific, useful in tumor profiling)</td>
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<tr>
<td>HUMGENE</td>
<td>104</td>
<td>2.34</td>
<td>.663 Basics of comparative genomics (shared genes among species)</td>
</tr>
</tbody>
</table>
1. Which of the following does NOT describe the amount of DNA contained in a single human cell?
   a. 23 pairs of chromosomes
   b. 1 million genes
   c. 3 billion base pairs
   d. 6 billion nucleotides

2. Imagine you are examining the DNA sequence of two unrelated people. What percentage of the sequence do you anticipate will be identical between these people?
   a. 100%
   b. about 99%
   c. about 50%
   d. 10 to 20%
   e. less than 1%

3. Genes are composed of
   a. protein
   b. amino acids
   c. DNA
   d. RNA

4. The primary function of a gene is to
   a. determine a particular trait for an individual
   b. allow cell division
   c. direct the formation of specific protein(s)
   d. direct a particular physiologic function
   e. produce DNA

5. A gene contains the code for
   a. a specific trait
   b. one or more proteins
   c. a particular physiologic function
   d. cell division

6. Which statement best describes the flow of genetic information?
   a. Chromosomes contain the code to make genes
   b. Genes contain the code to make DNA
   c. DNA contains the code to make proteins
   d. Proteins contain the code to make genes
7. DNA “sequence” refers to the order of
   a. nucleotides
   b. genes
   c. chromosomes
   d. proteins
   e. RNA

8. What is the relationship between nucleotides, base pairs, and genes?
   a. adjacent base pairs form a gene; nucleotides are located near the gene
   b. two nucleotides create a base pair, adjacent base pairs form a gene
   c. multiple base pairs form a nucleotide, and adjacent nucleotides form a gene
   d. many base pairs form a gene, and genes are organized into nucleotides

9. A group of genes represents a(n)
   a. nucleotide
   b. protein
   c. chromosome
   d. allele

10. Which statement best describes the physical relationship between genes and chromosomes?
    a. Chromosomes are organized into genes.
    b. Genes are organized into chromosomes.
    c. Both genes and chromosomes are contained on a strand of DNA.
    d. Genes and chromosomes are physically distinct.

11. When a gene is expressed, it
    a. copied to create a new gene (duplicated?)
    b. results in a visible trait or characteristic
    c. causes a protein to be formed
    d. initiates cell division

12. A gene that is expressed is
    a. copied to form DNA
    b. manifested as a physical trait
    c. replicated
    d. transcribed and translated into a protein product

13. When does gene expression occur?
    a. constantly, if the gene is dominant
    b. when a cell is preparing to divide
    c. when a chemical signal turns the gene ‘on’
    d. when the gene senses a need for its product
14. A laboratory test to determine if a gene is being expressed might examine
   a. the DNA sequence of the gene
   b. the order of bases within the mRNA
   c. the quantity of amino acids available for protein building
   d. the amount of mRNA transcribed from the gene

15. A genotype is
   a. a specific type of gene
   b. a set of genes that encode a particular trait
   c. a set of dominant genes
   d. an individual’s unique total collection of gene variants

16. An individual’s phenotype for a particular trait matches their genotype for that trait when
   a. the trait is influenced by environmental factors
   b. the trait is determined by one gene pair
   c. the trait is inherited in a dominant pattern
   d. the gene or genes that determine the trait are expressed
   e. the trait is readily visible

17. The human insulin gene has been identified and designated INS. Which cells in your body contain the insulin gene?
   a. cells in the liver
   b. cells in the blood
   c. cells that utilize glucose
   d. pancreatic beta cells
   e. all nucleated cells

18. An allele is
   a. part of a gene
   b. a trait
   c. the product of a gene
   d. a version or alternate form of a gene
   e. a pair or set of genes that determine a particular trait

19. What role does the insulin gene have in maintaining glucose homeostasis?
   a. directs production of enzymes involved in glucose regulation
   b. monitors blood glucose levels and signals pancreatic cells to release insulin
   c. signals pancreatic beta cells to release insulin
   d. encodes insulin

20. Cells carry out transcription and translation in order to
   a. divide
   b. produce new genes
   c. make proteins
   d. produce both DNA and proteins
21. The relationship between a gene and a protein is best described as
   a. genes are made of protein
   b. genes are made by proteins
   c. genes contain the code to form proteins
   d. proteins help genes to function

The next two questions are about Helen, a 54-year-old female who is about to begin treatment for a new diagnosis of atrial fibrillation.

22. Helen is known to be heterozygous for a gene named VKOR. What does that mean?
   a. She has a single copy of the VKOR gene.
   b. She has two copies of the VKOR gene; one is dominant and one is recessive
   c. She has two copies of the VKOR gene that are different in some way
   d. She inherited an altered VKOR gene from her mother

23. The VKOR gene is associated with response to the anticoagulant, warfarin. What should the nurse who is administering a standard dose of warfarin to Helen consider?
   a. She will most likely be anticoagulated too much
   b. She will most likely not be anticoagulated enough
   c. She is at high risk to develop an allergy to warfarin
   d. Her response to warfarin may be different from expected

24. Huntington disease (HD) has been mapped to a specific gene called the Huntingtin gene, or HTT. What is the best explanation for HD in affected individuals?
   a. People with Huntington disease have the HTT gene; unaffected people do not
   b. People with HD have an incorrect number of HTT genes
   c. People with HD have an altered form of the HTT gene
   d. In people with HD, the HTT gene is expressed; in unaffected people it is not expressed

25. A few genetic diseases are known to be 100% penetrant. This means
   a. Affected individuals have the altered gene present in all of their cells
   b. All offspring of affected individuals will be affected
   c. Every individual who inherits the altered gene will develop the disease
   d. Individuals who are affected will have all signs and symptoms of the disease

26. Jake and Allen are brothers who both have inherited neurofibromatosis (NF) from their father. While Jake is severely affected, Allen has very few symptoms of NF. What is the most likely explanation for the variation in symptoms between Jake and Allen?
   a. Jake inherited a dominant form of the NF gene and Allen inherited a recessive form
   b. The DNA sequence of the gene associated with NF is different in Jake compared to Allen
   c. Environmental factors were harmful in Jake and protective in Allen
   d. Jake and Allen inherited the same form of the NF gene, but the gene was expressed differently in Jake compared to Allen
27. At a community breast cancer screening, you are helping clients record their family health histories. Four women have a positive family history of breast cancer. Which of them is at LEAST risk to develop hereditary breast cancer?

a. Freda, whose mother was diagnosed with left breast cancer at age 55
b. Jane, whose paternal grandmother was diagnosed with bilateral breast cancer at ages 45 and 52
c. Liz, whose sister was diagnosed with right breast cancer at age 45
d. Monica, whose maternal aunt was diagnosed with cancer of the right breast at age 68

The next 5 questions concern Anna, a 35-year-old woman who has tested positive for a hereditary breast cancer mutation in a gene known as BRCA1.

28. Which of Anna’s cells contain the BRCA1 mutation?

a. her breast cells
b. only tumor cells
c. adipose cells
d. cells in her breasts and reproductive organs
e. all her cells that contain a nucleus

29. BRCA 1 is known to be inherited in an autosomal dominant pattern. Which statement best describes Anna’s risk of passing the BRCA 1 gene to her children?

a. All her children, regardless of gender, will inherit one copy of the BRCA1 gene from Anna
b. All her daughters will inherit the BRCA1 gene; her sons will not
c. Each daughter has a 50% risk to inherit the BRCA1 gene; her sons are not at risk
d. Each of her children, regardless of gender, has a 50% chance to inherit the BRCA1 gene
e. Although her children are at increased risk to inherit the gene, specific predictions cannot be made

30. Anna joins a support group of women with BRCA1 mutations. What can be said of the BRCA1 genes among these women?

a. The DNA sequence of the BRCA1 genes in these women is most likely identical
b. The women have identical mutations, although there may be other differences in DNA sequence within their BRCA1 genes
c. The DNA sequence within the women’s BRCA1 genes varies according to whether they have a dominant or recessive form of the gene
d. The women most likely have unique BRCA1 mutations
31. Anna is concerned about whether having her test results in her medical record poses any threat of discrimination. Which of the following statements about discrimination based on genetic testing is true?
   a. Federal law considers genetic testing to be no different from any other type of laboratory testing, although some states have specific laws about genetic testing.
   b. Federal law prohibits discrimination in health insurance and employment.
   c. Federal law prohibits discrimination in health insurance, employment, and life insurance.
   d. The Health Insurance Portability and Accountability Act (HIPAA) specifically protects against any discrimination based on genetic testing.

32. Anna’s neighbor, Delores, has breast cancer that is not hereditary. Which of Delores’s cells have cancer mutations?
   a. all her breast cells
   b. only tumor cells
   c. her adipose cells
   d. cells in her breasts and reproductive organs
   e. all her cells that contain a nucleus

33. Which of the following findings are NOT red flags in a genetic family history?
   a. early onset of heart disease
   b. bilateral breast cancer
   c. various forms of cancer in multiple family members
   d. breast cancer in a male relative
   e. single spontaneous miscarriage

34. The primary benefit of including common multifactorial OR COMPLEX conditions such as hypertension and diabetes in a genetic family history is to
   a. track diseases in families
   b. inform family planning decisions
   c. help establish a diagnosis
   d. predict risk for disease
   e. identify mutations

35. A disease or health condition is said to be inherited in a dominant pattern
   a. when a single copy of the altered gene is sufficient to cause disease
   b. when two copies of the altered gene are required to cause disease
   c. when the disease occurs in male offspring more often than female offspring
   d. when all offspring of an affected parent are also affected

36. A disease or health condition inherited in a dominant pattern
   a. affects all offspring of an affected parent
   b. is transmitted from a parent to offspring of the same sex
   c. occurs due to several mutations on the same chromosome
   d. occurs when both genes in a pair are altered
   e. requires only one copy of the disease-associated gene
37. An autosomal disorder
a. is automatically expressed when a single altered copy of a gene is inherited
b. results in production of antibodies to one’s own tissues
c. is inherited equally by male and female offspring
d. occurs due to several mutations on the same chromosome

38. James is diagnosed with cystic fibrosis (CF), an autosomal recessive disorder. Based on that information, what can you infer?
   a. both his parents must be CF carriers
   b. one parent must be affected with CF
   c. both parents must be affected with CF
   d. there is insufficient information to make any of the above inferences

The next several questions are about Joe and Sally, a young couple who are beginning their family. Joe has an autosomal dominant condition, which Sally is known NOT to have.

39. Joe’s genotype for the condition is most likely
   a. homozygous
   b. heterozygous
   c. equally likely to be homozygous or heterozygous
   d. dominant

Joe and Sally are expecting their first child, whose gender is unknown.
Please consider the following pedigree:

The dark square indicates Joe, who is diagnosed with an autosomal dominant condition. The open circle indicates Sally, who does not have that condition. The diamond indicates their unborn child.

40. Which statement best predicts the child’s risk to have inherited Joe’s condition?
   a. 100%
   b. 75%
   c. 50%
   d. 25%
   e. The baby’s risk varies according to whether it is a boy or a girl
41. After two years, Joe and Sally’s son, Michael, has been diagnosed with the same condition as Joe, and a second son is born. Which statement best predicts the new baby’s chance to carry the same diagnosis as his father and brother?

a. The new baby is certain to have the condition  
b. He has the same chance as his brother Michael had  
c. His chance is less than 100% but greater than the chance Michael had  
d. He has a lesser chance than Michael had

42. Years later, Joe and Sally have had two sons who are both affected with Joe’s condition. They are expecting a new baby, a girl. Which statement best predicts their daughter’s risk to also have Joe’s condition?

a. Her risk is the same as that of each of her brothers  
b. Her risk is greater than that of her brothers, since both brothers are known to be affected  
c. Her risk is less than that of her brothers, since both brothers are known to be affected  
d. Her risk is less because she is female

43. Jacob has Duchenne Muscular Dystrophy (DMD), an X-linked condition. Given that information, which of the following statements is most likely to be true about Jacob’s family?

a. His father also has DMD  
b. Either his mother or his father is a DMD carrier  
c. Both his mother and father are DMD carriers  
d. His mother is a DMD carrier  
e. All Jacob’s siblings share an equal risk to inherit DMD

44. When creating a genetic pedigree, the nurse should consider which of the following findings to be a ‘red flag’ indicating a possible need for a genetic referral?

a. a previous miscarriage  
b. breast cancer in her mother at age 64  
c. coronary bypass surgery in her father at age 52  
d. a sister who had twins
45. **What is the difference between genetics and genomics in healthcare?**
   a. Genomics is the application of genetic information to improve health outcomes
   b. Genomics considers effects of multiple genes in addition to environmental effects
   c. Genetics is broader in scope than genomics
   d. Genomics is concerned with molecular activities, and genetics is concerned with clinical outcomes

46. **A ‘genetic’ disease is**
   a. apparent at birth
   b. fatal
   c. caused by the deletion of genetic material
   d. caused by one or more genes [unique?] which are present in affected individuals and not present in people without the disorder
   e. caused by genetic material that is present in all individuals but altered in affected individuals

47. **Most genetic diseases occur because of**
   a. an alteration in DNA sequence
   b. missing or extra DNA
   c. an incorrect number of chromosomes
   d. the presence of a gene that is not found in healthy individuals

48. **Which statement best describes the role of genetics in cancer?**
   a. cancer is caused by mutations in genes with roles in cell growth and cell division
   b. most forms of cancer have no genetic basis
   c. most cancer is directly caused by inherited genetic variations
   d. most cancer occurs due to genetic predisposition along with environmental triggers

49. **In which way is pharmacogenomics expected to make the biggest change in healthcare?**
   a. increase in genetic testing
   b. personalized prescribing
   c. better prediction of disease risk
   d. increased focus on disease prevention

50. **Patients respond variably to standard doses of medications. Which statement best describes the genetic influence on human drug response?**
   a. Genes interact variably with drugs, according to the gene’s DNA sequence
   b. Genes cause the immune system to react variably to drugs
   c. Genes change cells to make them more or less responsive to drugs
   d. Genes direct the formation of proteins which interact variably with drugs
51. A drug receptor is best described as a(n)
   a. Protein
   b. Enzyme
   c. Structure or organelle
   d. Cell
   e. Antigen

The next two questions concern Laurie and Carrie, who are sisters in a family with a strong history of breast cancer.

52. Both Laurie and Carrie were tested for hereditary breast cancer. Laurie’s test was positive for BRCA1, the most common type of hereditary breast cancer. Carrie’s test was negative. Based on that information, which of the following statements is true?
   a. Laurie’s DNA includes a BRCA1 gene; Carrie’s DNA lacks that gene
   b. Both sisters have BRCA1 genes but Laurie’s is altered
   c. Laurie has a dominant form of the BRCA1 gene; Carrie has a recessive form
   d. Both sisters have identical BRCA1 genes, but the gene is expressed only in Laurie

53. What does Laurie’s positive BRCA1 test mean for her health?
   a. She probably has breast cancer now
   b. She needs a biopsy to determine if she has breast cancer
   c. She will develop breast cancer in the future
   d. She has a greater-than-average risk to develop breast cancer

54. In the United States, every newborn is screened for various genetic diseases. A baby with a positive newborn screen
   a. has a genetic disease
   b. may have genetic disease and requires more testing
   c. may develop a genetic disease
   d. is a carrier for a genetic disease

55. The primary purpose of a screening test is to
   a. diagnose a specific condition
   b. identify individuals who are at increased risk to have a specific condition
   c. diagnose a condition before the onset of symptoms
   d. identify carriers in a population

56. Which of the following statements is true about the breast cancer gene?
   a. It is found only in females.
   b. It is normally found in all humans, and its alteration increases risk for cancer
   c. It is found in all humans but only increases cancer risk in females
57. Carrier testing might be done to see if an asymptomatic individual
a. carries a recessive gene that could be passed to offspring
b. carries either a dominant or recessive gene that could be passed to offspring
c. carries a pathogen that could be transmitted to others
d. carries a gene or genes that could cause disease in the future

58. A mutation most commonly entails
a. an alteration in DNA sequence
b. an extra or missing gene
c. an extra or missing chromosome
d. an alteration in gene shape
e. a change in gene function  this seems to be also true. Maybe a change in gene expression

59. The effect of a mutation on health is more likely to be
a. beneficial
b. harmful
c. either – mutation effects are random

60. The most common way that mutations lead to disease is by
a. causing increased DNA replication
b. directing the formation of altered proteins or unexpected amounts of proteins
c. disrupting the function of the cell containing the mutation
d. evading or weakening the body’s immune response
e. silencing the gene containing the mutation
Appendix J
GNCI Pilot Version

Genomic Nursing Concept Inventory Ward © 2011
Pilot Version

Name: __________________________________ Date of Birth: ______________________

Gender (circle one)  Male  Female

Previous genetics education (please circle one)

  I  have / have not  taken a specific course in genetics

____________________________________________________________________________________

Please answer these questions in the order presented, circling each answer on the paper survey.
If you are not sure about an answer, please make your best guess. Try not to leave any questions blank.
When you are finished with the inventory, please fill in your Scantron.

1. Genes are composed of
   a. protein
   b. amino acids
   c. DNA
   d. RNA

2. The primary function of a gene is to
   a. determine a particular trait
   b. allow cell division
   c. direct the formation of specific protein(s)
   d. regulate a particular physiologic function
   e. replicate DNA

3. DNA “sequence” refers to the order of
   a. nucleotides
   b. genes
   c. chromosomes
   d. proteins
   e. RNA

4. An allele is
   a. part of a gene
   b. a trait
   c. the product of a gene
   d. a version or alternate form of a gene
   e. the set of genes that determines a particular trait
5. Imagine you are examining the DNA of two unrelated people. What percentage of their DNA sequence do you anticipate will be the same?
   a. 100%
   b. about 99%
   c. about 50%
   d. 10 to 20%
   e. less than 1%

6. Which of the following statements INCORRECTLY describes the amount of DNA in a single human cell?
   a. 23 pairs of chromosomes
   b. 1 million genes
   c. 3 billion base pairs
   d. 6 billion nucleotides

7. A group of genes represents a(n)
   a. nucleotide
   b. protein
   c. chromosome
   d. allele

8. What is the relationship between nucleotides, base pairs, and genes?
   a. adjacent base pairs form a gene; nucleotides are located apart from the gene
   b. many base pairs form a gene, and genes are organized into nucleotides
   c. multiple base pairs form a nucleotide, and adjacent nucleotides form a gene
   d. two nucleotides form a base pair, and adjacent base pairs form a gene

9. Cells carry out transcription and translation in order to
   a. prepare for cell division
   b. produce sperm or ova
   c. make proteins
   d. copy DNA and produce proteins

10. What situation or event causes a gene to be expressed?
    a. dominance—dominant genes are expressed and recessive genes are not
    b. the gene senses a need for its product
    c. a chemical signal turns the gene ‘on’
    d. the cell is preparing to divide

11. When a gene is expressed, it
    a. is copied to create a duplicate gene
    b. results in a specific trait or characteristic
    c. causes protein(s) to be formed
    d. initiates cell division
12. A laboratory test to determine if a gene is being expressed might examine
   a. the DNA sequence of the gene
   b. the order of bases within the mRNA
   c. the quantity of amino acids available for protein building
   d. the amount of mRNA transcribed from the gene

13. “Genotype” refers to
   a. a specific type of gene
   b. a set of dominant genes
   c. the traits or characteristics determined by one’s genes
   d. an individual’s total collection of gene variants

14. Which statement describes an individual’s phenotype?
   a. Jessica has one gene for blue eyes and one gene for brown eyes
   b. Kurt has a mutation in a gene that encodes the enzyme lactase
   c. Dave’s blood type is A positive
   d. Sarah is a carrier for muscular dystrophy
   e. Sharon has the gene for freckles

15. The human insulin gene has been identified and designated \textit{INS}. Which cells in your body contain the insulin gene?
   a. liver cells
   b. cells in the blood
   c. cells that utilize glucose
   d. pancreatic beta cells
   e. all nucleated cells

16. What role does the insulin gene have in regulating glucose levels?
   a. allows glucose to enter cells
   b. encodes enzymes involved in glucose regulation
   c. signals the pancreas to release insulin
   d. allows insulin to be produced

17. Genetic diseases are most often caused by
   a. a variation in the order of base pairs
   b. the abnormal duplication or absence of one or more genes
   c. an incorrect number of chromosomes
   d. the presence of a gene that is not found in healthy individuals

18. A dominant genetic disease or health condition
   a. affects all offspring of an affected parent
   b. occurs when a single copy of the disease gene is inherited
   c. requires two copies of the disease gene to be present
   d. is transmitted to offspring of the same gender
   e. occurs due to mutations in several genes on the same chromosome
19. A few genetic diseases are known to be 100% penetrant. This means
a. affected individuals have the disease gene present in all of their cells
b. all offspring of affected individuals will be affected
c. every individual who inherits the disease gene will develop the disease
d. individuals who are affected will have all signs and symptoms of the disease

20. An autosomal disorder
a. is automatically expressed when a single altered gene is inherited
b. results in production of antibodies to one’s own tissues
c. is inherited equally by male and female offspring
d. occurs due to several mutations on the same chromosome
e. occurs due to an incorrect number of chromosomes

21. A mutation most commonly entails
a. an alteration in DNA sequence
b. an extra or missing gene
c. an extra or missing chromosome
d. an alteration in gene shape
e. a change in gene function

22. The most common way for a mutation to contribute to disease is by
a. causing increased DNA replication
b. directing the formation of altered proteins or unexpected amounts of proteins
c. interfering with the body’s immune response
d. silencing the gene containing the mutation

The next two questions are about Helen, who is about to begin treatment with warfarin, an anticoagulant medication. Helen is known to be heterozygous for a mutation in a gene named VKOR. VKOR is associated with response to warfarin.

23. What does it mean that Helen is heterozygous for VKOR?
   a. She has a single copy of VKOR.
   b. She has two copies of VKOR; one is dominant and one is recessive.
   c. She has two non-identical copies of VKOR.
   d. She inherited an altered VKOR gene from her mother

24. What should the nurse who gives Helen a standard dose of warfarin expect?
   a. Helen will most likely be anticoagulated too much.
   b. She will most likely not be anticoagulated enough.
   c. Her response to warfarin may be different from expected.
   d. She is at high risk to have an allergic reaction to warfarin.

* * * End of situation about Helen * * *
25. Jake and Allen are brothers who both inherited neurofibromatosis (NF) from their father. Jake’s symptoms are much more severe than Allen’s. What is the most likely explanation for their variation in symptoms?
   a. Jake inherited a dominant form of the NF gene and Allen inherited a recessive form
   b. The DNA sequence of Jake’s NF gene is different than Allen’s
   c. Environmental factors were harmful in Jake and protective in Allen
   d. Jake and Allen inherited the same form of the NF gene, but the gene was expressed differently in Jake compared to Allen

26. In the United States, every newborn is screened for various genetic diseases. A baby with a positive newborn screen
   a. has a genetic disease
   b. may have a genetic disease and requires more testing
   c. may develop a genetic disease
   d. is a carrier for a genetic disease

The next two questions are about James, a 2-year-old boy diagnosed with cystic fibrosis (CF), an autosomal recessive disease.

27. Based on the above information, what (if anything) can you infer about James’ parents?
   a. At least one parent must be affected with CF.
   b. Both parents must be affected with CF.
   c. Each parent must have at least one copy of the disease gene.
   d. There is insufficient information to make any of the above inferences.

28. James’ parents are expecting a new baby. What chance does the baby have to be affected with CF?
   a. 25%
   b. 50%
   c. 75%
   d. it depends on the sex of the baby

** End of situation about James **

29. Jacob has Duchenne Muscular Dystrophy (DMD), an X-linked condition. Given that information, which of the following statements is most likely to be true about Jacob’s family?
   a. His father also has DMD
   b. His father is a DMD carrier
   c. His mother is a DMD carrier
   d. Either his mother or his father is a DMD carrier
   e. Both his mother and father are DMD carriers

30. The primary benefit of including common multifactorial conditions such as hypertension and diabetes in a genetic family history is to
   a. track diseases in families
   b. inform reproductive decisions
   c. help establish a diagnosis
   d. predict risk for disease
   e. identify mutations
The next several questions concern Anna, a 35-year-old woman who has tested positive for a hereditary breast cancer mutation in a gene known as BRCA1.

31. Which of the following statements is true about the BRCA1 gene?
   a. This gene is not found in everyone; people who do have it are at increased risk for cancer
   b. This gene is found only in females
   c. It is found in all humans but only increases cancer risk in females
   d. It is normally found in all humans, and its alteration increases risk for cancer

32. Which of Anna’s cells contain the BRCA1 mutation?
   a. her breast cells
   b. only tumor cells
   c. adipose cells
   d. cells in her breasts and reproductive organs
   e. all her cells that contain a nucleus

33. What does Anna’s positive BRCA1 test mean for her health?
   a. She probably has breast cancer now
   b. She needs a biopsy to determine if she has breast cancer
   c. She will develop breast cancer in the future
   d. She has a greater-than-average risk to develop breast cancer

34. The BRCA1 mutation is known to be inherited in an autosomal dominant pattern. Which statement best describes Anna’s risk of passing the gene mutation to her children?
   a. All her children, regardless of gender, will inherit the mutation from Anna.
   b. Each child has a 75% chance to inherit the mutation.
   c. Each daughter has a 50% chance to inherit the mutation; her sons are not at risk.
   d. Each of her children, regardless of gender, has a 50% chance to inherit the mutation.
   e. Although her children are at increased risk to inherit the mutation, specific predictions cannot be made.

35. Anna joins a support group of women with BRCA1 mutations. Which statement best predicts the DNA sequence of these women’s BRCA1 genes?
   a. The sequence of their BRCA1 genes is most likely identical.
   b. The sequence of their BRCA1 genes may vary, although they likely have identical mutations.
   c. The sequence varies depending on whether they have a dominant or recessive form of the gene.
   d. The women are likely to have unique BRCA1 mutations.

36. Anna is concerned about whether having her test results in her health record poses any threat of discrimination. Which of the following statements is true?
   a. Federal law considers genetic testing to be no different from any other type of laboratory testing, although some states have specific laws about genetic testing.
   b. Federal law prohibits discrimination based on genetic testing in health insurance and employment.
   c. Federal law prohibits discrimination based on genetic testing in health insurance, employment, and life insurance.
   d. The federal Health Insurance Portability and Accountability Act (HIPAA) specifically protects against discrimination based on genetic testing.
37. Anna’s neighbor, Delores, has breast cancer that is not hereditary. Which of Delores’s cells have cancer mutations?
   a. all her breast cells
   b. only tumor cells
   c. her adipose cells
   d. cells in her breasts and reproductive organs
   e. all her cells that contain a nucleus

38. Anna’s sister, Noreen, also had BRCA1 testing, and her test result was normal. Based on that information, which of the following statements is true?
   a. Anna’s DNA includes a BRCA1 gene; Noreen’s DNA lacks that gene
   b. Both sisters have BRCA1 genes but Anna has an altered copy
   c. Anna has a dominant form of the BRCA1 gene; Noreen has a recessive form
   d. Both sisters have identical BRCA1 genes, but the gene is expressed only in Anna

   * * * End of situation about Anna * * *

39. When creating a genetic pedigree, the nurse should consider which of the following findings to indicate a possible need for a genetic referral?
   a. a previous miscarriage
   b. breast cancer in her mother at age 64
   c. a heart attack in her father at age 48
   d. a sister and grandmother who had twins

40. What is the difference between genetics and genomics in healthcare?
   a. Genomics is the application of genetic information to improve health outcomes.
   b. Genetics focuses on single-gene conditions, while genomics considers the effects of all genes as well as environmental effects.
   c. Genetics is broader in scope than genomics.
   d. Genomics focuses on molecular mechanisms; genetics is concerned with clinical outcomes.

41. The Browns are a healthy couple whose two children, Jill and Michael, are affected with the same rare genetic disorder. No one else in the family is known to be affected. The most likely inheritance pattern for this disorder is
   a. autosomal dominant
   b. autosomal recessive
   c. X-linked
   d. mitochondrial
   e. multifactorial

42. Patients respond variably to standard doses of medications. Which statement best describes how genes influence human drug response?
   a. Genes interact variably with drugs, according to the gene’s DNA sequence
   b. Genes cause the immune system to react variably to drugs
   c. Genes change cells to make them more or less responsive to drugs
   d. Genes direct the formation of proteins which interact variably with drugs
43. A drug receptor is best described as a(n)
   a. enzyme
   b. antigen
   c. protein
   d. structure or organelle
   e. cell

44. Carrier testing might be done to see if an asymptomatic individual
   a. carries a recessive gene that could be passed to offspring
   b. carries either a dominant or recessive gene that could be passed to offspring
   c. carries a pathogen that could be transmitted to others
   d. carries a gene or genes that could cause disease in the future

45. Which statement best describes the role of genetics in cancer?
   a. Most cancer is caused by inherited mutations
   b. Most forms of cancer have no genetic basis
   c. Most cancer is caused by mutations in genes with roles in cell growth and cell division
   d. Most cancer occurs due to inherited predisposition along with environmental triggers

46. What would a predictive genetic test most likely predict?
   a. risk to pass a genetic condition to offspring
   b. severity of symptoms of a genetic condition
   c. risk to develop a late-onset genetic condition
   d. risk to be a carrier for a genetic condition

47. The genetic contribution to complex disorders like hypertension and diabetes is thought to be
   a. genes that have not yet been identified
   b. genes with multiple alleles
   c. a collection of genes, each contributing a small risk
   d. relatively minor; environmental factors contribute most of the risk for complex diseases

48. At a community breast cancer screening, you see four women with a family history of breast cancer. Which of them is at greatest risk for hereditary breast cancer?
   a. Freda, whose mother was diagnosed with left breast cancer at age 65
   b. Jane, whose paternal grandmother was diagnosed with left breast cancer at age 45 and ovarian cancer at age 52
   c. Liz, whose sister was diagnosed with right breast cancer at age 72
   d. Monica, whose maternal aunt was diagnosed with right breast cancer at age 48

These last questions are about Joe and Sally, a young couple who are beginning their family. Joe has an autosomal dominant condition, which Sally is known NOT to have.

49. Joe’s genotype for the condition is most likely
   a. homozygous
   b. heterozygous
   c. equally likely to be homozygous or heterozygous
Joe and Sally are expecting their first child, whose gender is unknown. Please consider the following pedigree:

The dark square indicates Joe, who is diagnosed with an autosomal dominant condition.
The open circle indicates Sally, who does not have that condition
The diamond indicates their unborn child.

50. Which statement best predicts the child’s risk to have inherited Joe’s condition?
   a. 100%
   b. 75%
   c. 50%
   d. 25%
   e. The baby’s risk varies according to whether it is a boy or a girl

After two years, Joe and Sally’s son, Michael, has been diagnosed with the same condition as Joe, and a second son is born. Please consider this pedigree.

51. What risk does the new baby have to carry the same diagnosis as his father and brother?
   a. The new baby is certain to have the condition
   b. He has the same chance as his brother Michael had
   c. His chance is less than 100% but greater than the chance Michael had
   d. He has a lesser chance than Michael had

Years later, after both sons are known to be affected, Joe and Sally are expecting a new baby, a girl. Please consider this pedigree.

52. Which statement best predicts their daughter’s risk to be affected with the same condition?
   a. Her risk is the same as that of each of her brothers
   b. Her risk is greater than that of her brothers, since both brothers are known to be affected
   c. Her risk is less than that of her brothers, since both brothers are known to be affected
   d. Her risk is less because she is female

End of inventory. Please mark your Scantron. Thank you for your participation.
Appendix K
Pilot Test Item Analysis, 52 items

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Appendix L
Genomic Nursing Concept Inventory Ward © 2011
Beta Version
February, 2011

Name: ____________________________ Date of Birth: ____________________________

Gender (circle one)  Male     Female

Previous genetics education (please circle one)
   I have / have not taken a specific course in genetics

____________________________________________________________________________

Please mark your answer to each question on the paper survey and the Scantron form. If you are not sure about an answer, please make your best guess. Try not to leave any questions blank.

1. The primary function of a gene is to
   a. determine a particular trait
   b. regulate tissue development
   c. direct the formation of specific protein(s)
   d. control a particular physiologic function
   e. replicate DNA

2. DNA “sequence” refers to the order of
   a. nucleotides
   b. genes
   c. chromosomes
   d. proteins
   e. RNA

3. Imagine you are examining the DNA of two unrelated people. What percentage of their DNA sequence do you anticipate will be the same?
   a. 100%
   b. about 99%
   c. about 50%
   d. 10 to 20%
   e. less than 1%

4. The human insulin gene has been identified and designated INS. Which cells in your body contain the insulin gene?
   a. cells in the liver
   b. cells in the blood
   c. cells that utilize glucose
   d. pancreatic beta cells
   e. all nucleated cells
5. Which statement INCORRECTLY describes the amount of DNA in the human genome?
   a. 23 pairs of chromosomes
   b. 1 million genes
   c. 3 billion base pairs
   d. 6 billion nucleotides

6. Cells carry out transcription and translation to create
   a. DNA
   b. RNA
   c. proteins
   d. amino acids
   e. new cells

7. “Genotype” refers to
   a. a specific type of gene
   b. a set of dominant genes
   c. the traits or characteristics determined by one’s genes
   d. an individual’s total collection of gene variants

8. Rank the following genetic structures in terms of size starting with the largest and proceeding to the smallest: chromosome, gene, genome, nucleotide.
   a. genome, chromosome, gene, nucleotide
   b. genome, gene, chromosome, nucleotide
   c. chromosome, genome, gene, nucleotide
   d. chromosome, nucleotide, genome, gene
   e. chromosome, nucleotide, gene, genome

9. What role does the insulin gene have in regulating glucose levels?
   a. allows glucose to enter cells
   b. encodes enzymes involved in glucose regulation
   c. signals the pancreas to release insulin
   d. allows insulin to be produced

10. A dominant genetic disease or health condition
    a. affects all offspring of an affected parent
    b. occurs when a single copy of the disease gene is inherited
    c. requires two copies of the disease gene to be present
    d. is transmitted to offspring of the same gender
    e. occurs due to mutations in several genes on the same chromosome

11. A laboratory test of gene expression might examine
    a. the DNA sequence of the gene
    b. the order of bases within the mRNA
    c. the quantity of amino acids available for protein building
    d. the amount of mRNA transcribed from the gene
The next two questions are about Emma, who is about to begin treatment with warfarin, an anticoagulant medication. Emma had a genetic test showing she has a mutation in a gene that is associated with response to warfarin.

12. What should the nurse giving Emma a standard dose of warfarin expect?
   a. Emma will most likely be anticoagulated too much.
   b. She will most likely not be anticoagulated enough.
   c. Her response to warfarin may be different from expected.
   d. She is at high risk to have an allergic reaction to warfarin.

13. The genetic test shows that Emma is heterozygous for the gene mutation. What does that mean?
   a. She has a single copy of the gene.
   b. She has two copies of the gene; one is dominant and one is recessive.
   c. She has two copies of the gene, both of which are altered.
   d. She has two non-identical copies of the gene.
   e. She inherited an altered gene from her mother.

*** End of situation about Emma ***

14. In the United States, every newborn is screened for various genetic diseases. A baby with a positive newborn screen
   a. has a genetic disease
   b. may have genetic disease and requires more testing
   c. may develop a genetic disease
   d. is a carrier for a genetic disease

The next two questions are about Sophia, a 2-year-old girl with an unusual autosomal recessive condition called Kindler syndrome.

15. Based on the information above, what (if anything) can you infer about Sophia’s parents?
   a. At least one parent must be affected with Kindler syndrome.
   b. Both parents must be affected.
   c. Each parent must have at least one copy of the disease gene.
   d. There is insufficient information to make any of the above inferences.

16. Sophia’s parent are expecting a new baby. What is the chance that the new baby will also have Kindler syndrome?
   a. 25%
   b. 50%
   c. 75%
   d. it depends on the sex of the baby

*** End of situation about Sophia ***

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17. Jacob has Fabry disease, an X-linked condition. Given that information, which of the following statements is most likely to be true about Fabry disease in Jacob’s family?
   a. His father also has Fabry disease
   b. His father is a carrier
   c. His mother is a carrier
   d. Either his mother or his father is a carrier
   e. Both his mother and father are carriers

The next few questions concern Anna, a 35-year-old woman who has tested positive for a hereditary breast cancer mutation in a gene known as *BRCA1*.

18. Which of Anna’s cells contain the *BRCA1* mutation?
   a. her breast cells
   b. only tumor cells
   c. adipose cells
   d. cells in her breasts and reproductive organs
   e. all her cells that contain a nucleus

19. Anna joins a support group of women with *BRCA1* mutations. Which statement best describes the DNA sequence of these women’s *BRCA1* genes?
   a. Their *BRCA1* sequence is most likely identical
   b. Although their *BRCA1* sequence may vary, they likely have identical mutations
   c. Their sequence varies according to whether they have a dominant or recessive form of the gene
   d. The women are likely to have unique *BRCA1* mutations

20. Anna’s sister, Nicky, also had *BRCA1* testing, and her test result was normal. Based on that information, which of the following statements is most likely to be true?
   a. Anna’s DNA contains a *BRCA1* gene; Nicky’s DNA lacks that gene
   b. Both sisters have *BRCA1* genes but Anna has an altered copy
   c. Anna has a dominant form of the *BRCA1* gene; Nicky has a recessive form
   d. Both sisters have identical *BRCA1* genes, but the gene is expressed only in Anna

** *** End of situation about Anna *** **

21. The most common way for a mutation to contribute to disease is by
   a. increasing the rate of DNA replication
   b. directing the formation of altered proteins or unexpected amounts of proteins
   c. interfering with the body’s immune response
   d. silencing the gene containing the mutation

22. Carrier testing might be done to see if an asymptomatic individual
   a. carries a recessive gene that could be passed to offspring
   b. carries either a dominant or recessive gene that could be passed to offspring
   c. carries a pathogen that could be transmitted to others
   d. carries a gene or genes that could cause disease in the future
23. When creating a genetic pedigree, the nurse should consider which of the following findings to indicate a possible need for a genetic referral?
   a. a previous miscarriage
   b. breast cancer in her mother at age 64
   c. a heart attack in her father at age 43
   d. a sister and a grandmother who had twins

24. An autosomal disorder
   a. is automatically expressed when a single altered gene is inherited
   b. results in production of antibodies to one’s own tissues
   c. is inherited equally by male and female offspring
   d. occurs due to several mutations on the same chromosome
   e. occurs due to an incorrect number of chromosomes

25. The genetic contribution to common complex disorders like hypertension and diabetes is thought to be
   a. an incorrect number of chromosomes
   b. genes that have not yet been identified
   c. genes with multiple alleles
   d. a collection of genes, each contributing a small risk
   e. relatively minor; environmental factors contribute most of the risk for complex diseases

26. The primary benefit of including common conditions such as hypertension and diabetes in a genetic family history is to
   a. identify mutations associated with common conditions
   b. track diseases in populations
   c. inform reproductive decisions
   d. help establish diagnoses
   e. predict disease risk and plan care

27. A drug receptor is best described as a(n)
   a. enzyme
   b. antigen
   c. protein
   d. structure or organelle
   e. cell

28. Patients respond variably to standard doses of medications. Which statement best describes how genes influence human drug response?
   a. Genes interact variably with drugs, according to the gene’s DNA sequence
   b. Genes cause the immune system to react variably to drugs
   c. Genes change cells to make them more or less responsive to drugs
   d. Genes direct the formation of proteins which interact variably with drugs
The last questions are about Joe and Sally, a young couple who are beginning their family. Joe has an autosomal dominant condition, which Sally is known NOT to have.

29. Which statement is most likely true about Joe’s genotype for the condition?
   a. He most likely has two altered copies of the disease gene
   b. He most likely has one normal copy and one altered copy of the disease gene
   c. (a) and (b) are equally likely

Joe and Sally are expecting their first child, whose gender is unknown. Please consider the following pedigree:

![Pedigree Diagram]

The dark square indicates Joe, who is diagnosed with an autosomal dominant condition.
The open circle indicates Sally, who does not have that condition
The diamond indicates their unborn child.

30. Which statement best predicts the child’s risk to have inherited Joe’s condition?
   a. 100%
   b. 75%
   c. 50%
   d. 25%
   e. The baby’s risk varies according to whether it is a boy or a girl

Years later, Joe and Sally have had two sons, both affected with Joe’s condition. They are expecting a new baby, a girl. Please consider this pedigree.

![Pedigree Diagram]

31. Which statement best predicts their daughter’s risk to be affected with the same condition?
   a. Her risk is the same as that of each of her brothers
   b. Her risk is greater than that of her brothers, since both brothers are known to be affected
   c. Her risk is less than that of her brothers, since both brothers are known to be affected
   d. Her risk is less because she is female

* * * End of inventory * * *
## Appendix M
Analysis of 26 Items Retained on 31-item GNCI Beta Version

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