Continuing Education

Genes, Genetics and Genomics, Oh My!

Essential Genetic and Genomic Competencies for All Registered Nurses

At the completion of the article and the post test, the reader should be able to:

- State why professional nurses should achieve genetic/genomic competence.
- Differentiate two aspects of the genetic/genomic nursing competencies.
- Recognize strategies for integrating genetic/genomic competencies into nursing practice.
- Identify genetic/genomic resources for continued self learning and patient teaching.

The competencies related to genetics and genomics for all registered nurses were established by an independent panel of nursing leaders in 2005. The purpose of this article is to help nurses in clinical practice recognize the competencies they need in genetics and genomics and how these can positively impact their practice.

Patricia Newcomb, PhD, RN
CPNP is an Assistant Professor of Nursing and the Science Director of the Genomics Translational Research Lab at the University of Texas at Arlington. Her current research interests include investigating how genomic factors interact with environment in childhood asthma. She also practices as a Pediatric Nurse Practitioner in primary care in the Fort Worth area.

Barbara M. Raudonis, PhD, RN is an Associate Professor of Nursing at the University of Texas at Arlington. Her current research interests include associating gene variants with fatigue in women undergoing chemotherapy for breast cancer and the genomic factors involved in aging and end of life experiences.

The authors report no relevant financial relationships or conflicts of interest. They do not intend to discuss any off-label use of any product.

More essential competencies? Are we joking? No, but don't panic. The document, Essential Genetic and Genomic Competencies for All Registered Nurses, is your yellow brick road to competency. Here's why.

Over the past two decades a wave of change in approaches to preventing, assessing, diagnosing, and treating health problems has emerged, and many nurses remain unaware that they are on the crest of that wave. The wave is the first generation of interventions resulting from translating knowledge generated by the Human Genome Project into healthcare practice. For fifty years nursing leaders have called for more genetics content in undergraduate nursing programs to prepare nurses for a healthcare world in which genomics will be a major influence, but incorporating the
relevant essentials of a growing science like genetics into crowded nursing curricula has been a struggle (Brantl & Esslinger, 1962). As a result, few practicing nurses today have sufficient knowledge about genetics or genomics, and many fail to appreciate the importance of genomics in patient care.

Since the completion of the Human Genome Project discoveries regarding the genetic contribution to disease have grown rapidly. Genetics has moved from the classical Mendelian study of single genes and how specific traits are passed from one generation to the next, to genomics, which studies all the genes in an individual together, including how genes interact with each other and with the environment. It has become clear that all diseases and the health status of well individuals have a genetic component, thus genetics is now a central science for all healthcare professionals. Now and in the future prevention and treatment will increasingly exploit what is known about individual genomic characteristics. “Personalized medicine” will use information about patient genotypes or gene expression to design preventive interventions and treatments uniquely suited to each individual patient. Nurses who administer or monitor medical treatment, provide counseling to patients regarding health conditions, make referrals, manage cases, or design health promotion or disease prevention interventions in the community will need a working knowledge of the genomic framework that underpins “personalized medicine.”

The public, already primed by the excitement of the Human Genome Project and direct-to-consumer advertising for genetic tests, will expect healthcare professionals to be equipped to answer their questions and provide informed and safe health care that incorporates the advances that contemporary genetic and genomic discoveries promise.

Genetic and Genomic Competencies

Competencies are measurable abilities that are required to adequately fulfill a role. Typically, competencies must be demonstrated in order to achieve course credit, certifications, or other forms of recognition. Increasingly nursing education focuses on the demonstration of competencies rather than just grades, and competencies are evaluated regularly in most clinical practice settings. For instance, clinical nurses might be required to demonstrate competency in responding to respiratory emergencies or relating to patients in a culturally sensitive manner, while academic nurses are expected to demonstrate teaching competencies (National League for Nursing, 2005). As genomics becomes increasingly integrated into healthcare, it is no surprise that nurses will be expected to achieve a new set of competencies: genetic/genomic competencies.

The American Nurses Association and the National Institutes of Health through the National Human Genome Research Institute and the Office of Rare Diseases responded to the need to organize and articulate genetic/genomic competencies for nurses. They supported an independent panel of nurse leaders, representing clinical, research, and academic settings, as they developed a set of relevant nursing competencies for all nurses regardless of level of education or practice settings. The panel developed the competencies based on 1) existing guidelines, recommendations, peer-reviewed reports of practice based genetic/genomic competencies, 2) commentary from nurse representatives to the 2005 meeting of the National Coalition for Health Professional Education in Genetics (NCHPEG), 3) attendee statements from a 2005 meeting of key stakeholders, and 4) public comment from the general nursing community. To date 49 organizations have endorsed the consensus document (Competency Focus, 2008). The entire document can be accessed from http://www.genome.gov/17517037.

The essential competencies are organized in two categories: professional responsibilities and the professional practice domain. The professional responsibilities are consistent with the Scope and Standards of Practice (2004) published by the American Nurses Association. Living in the genomic era, Registered Nurses are now expected to incorporate genetic and genomic knowledge and skills into their nursing practice across settings (Consensus Panel, 2006). Competencies in the Professional Practice Domain address the following: Nursing Assessment that includes genetic and genomic factors, applying/integrating genetic and genomic Knowledge in care processes; identification of patients who could benefit from genetic services, genetic referral activities, and provision of education, care, and support, that includes genetic and genomic-related content or

---

**Genetics and Genomics Vocabulary**

**Genetics** is the study of inheritance, or the way traits are transmitted down through generations. For instance, the knowledge that sickle cell disease only occurs in the offspring of two carriers of a particular autosomal recessive gene is a genetic discovery.

**Genomics** is the study of all the genes in a person, as well as gene interactions with each other, the environment, psychosocial factors, and cultural factors. The finding that fish consumption results in lower concentrations of lipoproteins in people with a certain rare variation of the LPA gene than in others is a genomics discovery.

**Genotype** refers to heritable information carried in the DNA molecule in each cell of a living being. DNA that has two different forms of a gene for eye color is a genotype. DNA that has two identical forms of a gene for eye color is a different genotype.

**Phenotype** is the observable manifestation of the genotype. Eye color is a phenotype.

**Personalized medicine** is the practice of tailoring prevention and disease management activities for individuals based on genomic characteristics.
activities. See Genetics/Genomics Competencies in Box 2 for a listing of the individual competencies.

Implementation of the essential nursing competencies for genetics and genomics is well underway. Strategies for implementation include: incorporating genetic and genomic content in the NCLEX exam, certification exams, continuing education programs for practicing nurses, and accreditation criteria. A newsletter, Competency Focus, is published periodically to update nurses regarding the status of the implementation process and to share resources and successful strategies. Competency Focus is currently available by request from Kathleen Calzone, Senior Nurse Specialist (Research) in the NCI/CCR genetics branch of NIH at calzonek@mail.nih.gov.

Clinical Applications

Assessment

Since Florence Nightingale nurses have concerned themselves with assessing risk. From assessing the risk of future disease for ambulatory patients to assessing the risk of sepsis in an intensive care unit, nurses assess risk to prevent harm. Genetics and genomics offer the opportunity to include additional risk factors or protective factors in risk assessments.

The most powerful genomics tool available to any clinician is the family history. Collecting a family history is a nursing intervention that promotes comprehensive risk assessment. Recognizing the “red flags” that occur in family histories makes a difference. For instance, the school nurse who discovers that her high school quarterback’s father died of a heart attack at age 40 may save her patient’s life by conveying that important information to the child’s physician or arranging access to medical evaluation. Early cardiac death of a first degree relative is a recognized risk factor for serious heart disease and is easily determined through the family health history. Nurses play an important role in collecting family history information in both ambulatory and hospital practice.

In time, genetic testing is expected to provide risk information on which treatment will be decided. For instance, investigations of the genotypes of badly burned patients revealed that two gene variations were significantly associated with increased risk for severe sepsis (Barber et al., 2004). If further research supports the hypothesis that the presence of these genetic variants predisposes patients to severe sepsis, clinicians may consider genetic testing as an assessment mechanism for burn patients as a way of targeting preventive interventions for sepsis. Nurses will have roles in explaining the test to patients and families, collecting and preparing specimens, and delivering interventions based on the results.

Assessing client understanding of procedures and conditions is a primary responsibility of nurses. Care of persons with genetic disorders or complex illnesses with genetic components requires that the nurse understand the nature of the disorder, including its pattern of inheritance, how the disorder causes pathology, and appropriate treatments. Understanding the basics about disorders helps insure safe care, but also is necessary in order to communicate effectively with patients regarding their disease, treatments, and implications for healthy lifestyles.

Intervention

Teaching, advocacy, and direct patient care are traditional nursing interventions. In the post-Human Genome Project healthcare landscape, genetic testing will be done more frequently, genomic factors influencing disease origin and severity will be recognized, and genetic or genomic rationales for treatment will be increasingly prevalent. Explaining testing, disease processes, and rationales for treatment is an important part of nursing activity. Now genetic testing, the influence of genetics on health conditions, and the genetic or genomic basis for treatments must be included in the patient teaching repertoire.

Nursing interventions related to genetic or genomic issues include a variety of activities in addition to assessment and teaching, such as administering drugs based on genotype, recognizing the need for genetics services, and making referrals to community genetics services or resources. Patient advocacy will increasingly include activities directed toward protecting private information, including genetic information, and insuring that such information is not used to discriminate against classes of people.

Genetic and genomic competencies for nurses were designed to insure that all registered nurses will be able to assess, intervene, and advocate for patients when genetic/genomic issues are involved, just as they would when any other factor influencing health is at stake. The following vignette presents an example of how selected competencies might be useful in the clinical practice of the registered nurse.

Vignette

Charlotte is visiting her mother who is in the intensive care unit following surgery for pheochromocytoma (an adrenal tumor). As the nurse is checking her mother’s blood pressure, Charlotte confides that she heard the doctor say about 10 per cent of pheochromocytomas are heritable and now she fears she might develop pheochromocytoma, as well, and wonders what the chances are. The nurse replies that pheochromocytoma is very rare and less than 2/10 percent of people with hypertension actually have an adrenal tumor, but she would be happy to talk with Charlotte when she has a few minutes “downtime.” The nurse checks the chart and sees on the admission family history that some family members are thought to have had cancers, but there is no history of pheochromocytoma. The family history consists of check marks in boxes without explanatory information. The nurse recently attended a CE class at her institution in which she learned how to construct pedigrees. She decides to obtain a three-generation family history from Charlotte and record it in the chart.

After leaving Charlotte the nurse takes a few moments to check Online Mendelian Inheritance in Man by means of the Internet at the nurses’ station. She refreshes her information about heritable causes of pheochromocytoma. The nurse returns later and asks Charlotte if she could expand on the family history. Together they construct a pedigree using symbols to express the family medical history visually. Charlotte reports that her
maternal aunt had a spinal tumor that was removed several years ago. Her maternal uncle had problems with his vision since childhood, but because he lives in another country, she is unsure what they are. She thinks he had to have surgery on his eyes last year, but can’t remember what it was about. Charlotte’s mother telephones her brother once or twice a year and thinks he said he had glaucoma. Charlotte’s maternal grandfather died as a result of kidney cancer. The nurse recognizes these occurrences as “red flags” for Von Hippel-Lindau (VHL) syndrome and suggests that Charlotte consult with her physician or a genetics counselor or advanced practice nurse with expertise in genetics to explore the implications of her family history. Charlotte does not want to discuss genetic issues with her physician and requests a referral to a genetics counselor. The nurse is familiar with genetic/genomic resources in the community, and gives Charlotte the names and contact phone numbers of the two genetics counselors in the county.

The intervention took approximately 15 minutes, and Charlotte’s mother, who also followed up with the genetics counselor and her physician, received genetic testing that established that she carried a high risk mutation in the VHL gene. Charlotte and her brother elected to have genetic testing, as well. They discovered that Charlotte did not carry the mutation, but her brother did. Based on the genetic testing information the brother’s nurse practitioner recommended frequent and regular screening for neoplasms, including ophthalmologic testing, laboratory tests, and imaging, for most of his life. She suggested that because VHL syndrome affects multiple body systems coordination of care might best be performed by a clinical geneticist. He agreed and she arranged a consultation. When he became engaged a year later, she met with the brother and his fiance to discuss VHL and reinforced the information the genetics counselor and geneticist had given the family regarding inheritance of VHL and health implications of the syndrome.

The vignette illustrates nursing activities performed by both a staff nurse and an advanced practice nurse that were based on genetics/genomics competencies. The registered nurse caring for Charlotte’s mother in the ICU demonstrated that she is incorporating genetic/genomic knowledge and skills into her professional responsibilities and practice. Her attendance at a CE offering to learn how to construct pedigrees helped her acquire genetic skills which she applied at the bedside. She tailored her services to Charlotte and her mother based on their knowledge level and

Box 1.

**The Beginner’s Genetic and Genomic Related Websites.**

<table>
<thead>
<tr>
<th>Website Name</th>
<th>Description</th>
<th>Website URL</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene Tests</td>
<td>A publicly funded genetics information resource developed for healthcare providers and researchers</td>
<td><a href="http://www.genetests.org/">http://www.genetests.org/</a></td>
</tr>
<tr>
<td>Genetic Alliance</td>
<td>Genetics and genomics related policy information maintained by the Genetic Alliance, a coalition of advocacy organizations</td>
<td><a href="http://www.geneticalliance.org/">http://www.geneticalliance.org/</a></td>
</tr>
<tr>
<td>U.S. Surgeon General’s Family History Initiative</td>
<td>Computerized tool for creating family pedigrees developed and maintained by the US Department of Health and Human Services</td>
<td><a href="http://www.hhs.gov/familyhistory/">http://www.hhs.gov/familyhistory/</a></td>
</tr>
<tr>
<td>National Society of Genetic Counselors (NSGC)</td>
<td>Website for the professional organization representing genetic counselors and links to a list of genetic counselors</td>
<td><a href="http://www.nsgc.org">http://www.nsgc.org</a></td>
</tr>
<tr>
<td>International Society of Nurses in Genetics (ISONG)</td>
<td>Maintains documents produced by ISONG members and links to resources relevant to nursing practice</td>
<td><a href="http://isong.org">http://isong.org</a></td>
</tr>
</tbody>
</table>
preferences. She understood the relationship of genetics to health, prevention, screening, and diagnostics. She was able to elicit a three-generation family history and construct a pedigree. She critically analyzed the history for risk factors. She identified credible resources for her own learning and for her clients, and she facilitated an important referral. In the primary care setting the advanced nurse practitioner demonstrated similar competencies.

Genetic and Genomic Resources

Resources related to genetics and genomics are now easily accessible. Nursing professional journals are including increasing numbers of research reports that describe nursing studies incorporating genetics, as well as articles that address basic genetic/genomics information. Genetic/genomic related information is mushrooming on the World Wide Web, but nurses must be savvy consumers. Described below and in Box 1 are selected reputable resources that will provide an entry into this exciting area of clinical practice and science. The following are books and monographs of especial interest to nurses.

  Describes the scope and standards for nurses in genetics.

  Monograph describes the background and context of the competencies, lists the essential competencies, references and an extensive appendices of resources.

  This monograph is a compilation of the articles from the Genomic Medicine series from November 2002 through September 2003 in The New England Journal of Medicine. The series describes how genetics and genomics have advanced our knowledge and understanding of the biology of disease, making specific diagnoses and the development of new treatment options.

  The text highlights competencies, knowledge, skills, and attitudes recommended by NCHPEG that health care professionals need to care for individuals with particular genetic conditions. Stories addressing the related biological, personal, and psychosocial issues are also included in the chapters.

  This is a comprehensive overview that can read be sequentially or as a reference text. Information about basic human genetics, the Human Genome Project and research findings are included. The text focuses on genetic issues relevant to nursing practice.

Available from Sigma Theta Tau.

This is a compilation of the most recent and relevant articles on the topic previously published in the periodicals of the Honor Society over the past five years.

There is a voluminous amount of genetic/genomics content and resources on the World Wide Web. As in any web search, be aware of the source of the content. Information ranges from basic biology course content to commercial enterprises selling genetic tests directly to consumers. Nurses must educate our patients to be savvy as they also encounter commercial advertising. See Box 3 for a listing of useful and reputable websites.

Conclusion

Professors were once fond of telling nursing students that the information they encountered in their textbooks would be obsolete in five years. In the field of biomedicine and genomics, information is being generated so rapidly that it seems to be “old news” in about five months! Luckily nurses are not expected to be experts in genetics or biomedicine, but nursing practice is an applied science, and we expect to derive many future nursing interventions from the evolving field of genomics. Understanding our own practice will demand that we master the vocabulary and basic concepts of genomics. Excellence in nursing will include maintaining an awareness of how genomics and genetics information and technologies are influencing health-related disciplines, the public, and nursing activity. Achieving a comfort level with genomics and genetics in nursing starts with becoming familiar with what will be expected of nurses. Reading the genetics and genomics competencies for all nurses is a natural first step. Ruby slippers are optional.
Essential Nursing Competencies for Genetics and Genomics


IMPORTANT DEFINITIONS:
The essential nursing competencies document uses the following human health-centered definitions:
Genetics: The study of individual genes and their influence on rare, single-gene disorders.
Genomics: The study of all the genes in the human genome together, including their interactions with each other, the environment, psychosocial factors, and cultural factors.

COMPETENCIES RELEVANT TO PROFESSIONAL RESPONSIBILITIES:
- Recognize when one's own attitudes and values toward genetics and genomics may affect care of clients.
- Advocate for client access to desired genetic and genomic services and/or resources including support groups.
- Examine competency of one's practice regularly and identify areas of strength as well as areas in which professional development related to genetics/genomics would be helpful.
- Incorporate genetic/genomic technologies and information into nursing practice.
- Tailor genetic/genomic information and services to clients based on their culture, religion, knowledge level, literacy, and preferred language.
- Advocate for the rights of all clients to autonomous, informed genetic/genomic-related decision-making and voluntary action.

COMPETENCIES RELATED TO THE PROFESSIONAL PRACTICE DOMAIN

Nursing Assessment: Applying/Integrating Genetic/Genomic Knowledge
The registered nurse:
- Understands the relationship of genetics/genomics to health, prevention, screening, diagnostics, prognostics, treatment, and monitoring of treatment effectiveness.
- Elicits a minimum of a three-generation family health history
- Constructs a pedigree from family history information using standardized symbols and terminology
- Collects personal health and developmental histories that consider genetic, genomic, and environmental risks.
- Conducts comprehensive health and physical assessments incorporating knowledge of genetic/genomic and environmental influences.
- Assesses clients' knowledge, perceptions, and responses to genetic/genomic information.

Identification
The registered nurse identifies:
- clients who may benefit from specific genetic/genomic information and/or services based on assessment data.
- credible, accurate, appropriate, and current genetic/genomic information resources, services, and/or technologies specific to given clients.
- ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genetic/genomic information and technologies
- issues that undermine the rights of all clients for autonomous, informed, genetic/genomic-related decision-making and voluntary action.

Referral activities
The registered nurse:
- Facilitates referrals for specialized genetic/genomic services for clients as needed.

Provision of Education, Care, and Support
The registered nurse:
- Interprets selective genetic/genomic information or services for clients.
- provides clients with credible, accurate, appropriate and current genetic/genomic information, resources, services, and/or technologies that facilitate decision-making.
- Uses health promotion and disease prevention practices that consider genetic/genomic influences on personal and environmental risk factors AND incorporate knowledge of genetic/genomic risk factors
- Uses genetic/genomic-based interventions and information to improve client outcomes.
- Collaborates with healthcare providers in providing genetic/genomic care.
- Collaborates with third-party payers to facilitate reimbursement for genetic/genomic services
- Performs interventions appropriate to client genetic/genomic needs
- Evaluates effectiveness of genetic/genomic technology, information, interventions, and treatments on client outcomes.
References


Describes the scope and standards for nurses in genetics.


"Genes, Genetics, Genomics: Oh My!"
Essential Genetic and Genomic Competencies for All Registered Nurses

Purpose: The purpose of this article is to help nurses in clinical practice recognize the competencies they need in genetics and genomics and how these can positively impact their practice.

Objectives: At the completion of the article and the post-test, the reader should be able to:
1. State why professional nurses should achieve genetic/genomic competence.
2. Differentiate two aspects of the genetic/genomic nursing competencies.
4. Identify genetic/genomic resources for continued self-learning and patient teaching.

How to earn One Contact Hour:
1. Read the article.
2. Complete the posttest questions and program evaluation by circling the selected responses on the answer sheet.
3. Fill out the registration form.
4. Send registration form, answer sheet, and a check for $12.00

Registration Information:
Name: ________________________________
Address: ________________________________
City/State/ZIP: __________________________
State(s) of Licensure: _____________________
Telephone Number: ______________________
Email: ________________________________

Post Test Questions for Continuing Education Credit
Article: "Genes, Genetics, Genomics: Oh My!" Essential Genetic and Genomic Competencies for all Registered Nurses

Please circle your response for each question

1. All of the above It is important for registered nurses practicing at the bedside to achieve genetic/genomic competence because
A. Nurse leaders have been calling for genetics competence for more than 50 years
B. Genetics content will be included in the NCLEX examination
C. Treatment, monitoring, and prevention activities will increasingly rely on patients’ genetic/genomic characteristics
D. Genetic/genomic competencies will be added to clinical ladder criteria in most healthcare facilities

2. Two genetic/genomic competencies related to the professional responsibilities of all registered nurses include
A. Recognizing how one's attitudes toward genetics affect patients and advocating for client access to genetic services
B. Eliciting 3-generation family histories and constructing pedigrees
C. Identifying clients who may benefit from genetic/genomic information and facilitating referrals for specialized genetic services
D. Interpreting genetic/genomic information for clients and collaborating with colleagues to provide genetic/genomic care

3. The genetics and genomics competencies for nurses were developed by
A. The American Nurses Association
B. The American Academy of Colleges of Nursing
C. An NIH panel of genetics experts
D. An independent panel of nursing leaders

4. A registered nurse could integrate genetic/genomics competencies into which of the following cases?
A. An 11-year-old child presents for his annual physical examination
B. A patient is admitted to an intensive care unit with third degree burns to 45 percent of her body surface area.
C. An young adult with sickle-cell trait requests contraceptive services prior to her wedding
D. All of the above

5. An up-to-date and reliable resource for use by health care professionals to learn how genetic testing is used in making clinical diagnoses is:
A. The Genetic Alliance website
B. The Gene Tests website
C. The Genetics Home Reference website
D. The ISONG website

6. Health care providers can design unique preventive interventions and treatments for individual patients by using the patient's own
A. Genotype or gene expression
B. Phenotype or RNA levels
C. Karyotype analysis
D. Chromosomes

7. Content related to the genetic/genomic competencies is going to be included
A. In the Nurse Practice Acts of each state
B. In the NCLEX examinations
C. In the NIH grant proposal criteria
D. In the Medicare reimbursement criteria

8. Family history is a clinician's most powerful genomics tool available for
A. Analyzing genotypes
B. Risk assessment
C. Analyzing pedigrees
D. Assessment of causes of mutations

9. Patient advocacy will include protecting private information including
A. Diagnoses
B. Genetic information
C. Lab results
D. All of the above

10. In the post-Human Genome Project era nursing interventions will now include
A. Genetic testing related to data from a family history
B. Recognition of genomic factors influencing development of disease
C. Explanation regarding the genetic or genomic basis for treatments
D. All of the above

Program Evaluation
Strongly Disagree          Strongly Agree
Objective 1 was met.                  1 2 3 4 5
Objective 2 was met.                  1 2 3 4 5
Objective 3 was met.                  1 2 3 4 5
Objective 4 was met.                  1 2 3 4 5
The article was effective as a learning resource/tool.                  1 2 3 4 5
The objectives were relevant to the overall purpose.                  1 2 3 4 5
The activity met your expectations.                  1 2 3 4 5
List two ways that you will integrate what you learned in this activity into your practice and/or work environment: __________________________

The following were disclosed to me as part of this educational activity:
Requirements for successful completion
Yes  No
Conflicts of interest
Yes  No
Non-Endorsement of Products
Yes  No
Off-label use
Yes  No
Did you as the participant, notice any bias that was not previously disclosed in this presentation? Yes  No
If Yes, please describe______________________________

State the number of minutes it took you to read the article, complete the test and evaluation __________________ min.