Essential Nursing Competencies for Genetics and Genomics: Implications for Critical Care

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The implications of genetics and genomics for critical care nurses are becoming more evident, not only in the care provided but also in the numerous medications administered. Genetic causes are being discovered for an increasing number of chronic illnesses and diseases, such as Huntington disease. Because of the scientific and pharmacological advances, leading nursing organizations, such as the American Nurses Association, have established competencies in genetic knowledge for nurses. Such competencies help ensure quality care. Recent advances in the pharmacogenomics of therapy for human immunodeficiency virus disease, cancer, cardiovascular disease, and malignant hyperthermia have indicated a genetic linkage; therefore treatments are targeted toward the genetic aspect of the abnormality. Critical care nurses need knowledge of these genetic conditions and of medications affected by genetic factors. (*Critical Care Nurse.* 2011;31[5]:e1-e7)

Since the 1990s, and certainly since the completion of the Human Genome Project in 2003, the need for nurses to understand genetic information has become increasingly evident.[1] Knowledge of genetics, genomics, and pharmacogenomics (ie, the study of heredity, gene function, and how medications target genes) continues to increase,[2] and these 3 areas are increasingly important in critical care in the treatment of abnormalities such as sickle cell disease. Genetics addresses the association between sickle cell disease and a gene abnormality, specifically a mutation in the *HBB* gene on chromosome 11. Genomics is concerned with how this mutated gene interacts with the other genes in a person's body and is associated with the severity of expression of sickle cell disease. Pharmacogenomics comes into play when pain medication must be changed because a patient with sickle cell disease is unresponsive to a prescribed narcotic.

As diagnostic testing is improved and as medications increasingly target specific genetic alterations, critical care nurses are in a unique position to share knowledge of genetics and genomics with patients and patients' families. When this knowledge is applied to a patient's condition, nurses function as educators and advocates, helping patients understand the science behind the treatment plan. In this article, we provide a brief historical overview of genetics in nursing and address the knowledge nurses need at the bedside today.

CE Continuing Education

This article has been designated for CE credit. A closed-book, multiple-choice examination follows this article, which tests your knowledge of the following objectives:

1. Understand the implications of genetics and genomics for critical care nurses
2. Identify the essential genetic competencies for nurses
3. Differentiate genetic causes for specific chronic illnesses and diseases

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Background

Nurses began applying genetics to clinical practice formally in the 1970s, with a primary emphasis on prenatal screening and newborn metabolic disease.[3] In 1998, the first publication explaining the scope and standards for clinical genetics in nursing became available.[4] In 2006, the American Nurses Association

published specific genetic competencies for nurses that can be used to guide educators in teaching the importance of genetics to nursing students.[5,6] The essential nursing competencies and curricular guidelines for genetics and genomics (Table 1) do not replace existing standards of practice. Rather, the competencies and guidelines are intended to provide a genetics and genomics perspective.

Currently, genetics are a part of all aspects of nursing. Advances in science lead to advances at the bedside, and the relationship between genetics and nursing becomes more obvious each year. Nursing schools lay the foundation of genetic knowledge, but nurses in practice must stay current throughout the nurses' careers. Genetics education can be acquired through continuing education, reading, and conferences (Table 2).

Implications for Critical Care Nurses

The components of the competencies and guidelines[1,4-7] are easily incorporated in critical care. Nurses show genetic competency when they provide patients and patients' families education on medications specific to a genetic mutation or on the genetic basis of an illness (Table 1). Table 3 lists some of the more common conditions in critical care that have a genetic component.

Genetics

Long-QT syndrome can be manifested in the hospital as sudden cardiac arrest and even sudden death. Nurses who realize that this syndrome is sometimes recognized after a patient has died are in a unique position to provide education to patients' families. Discussing the possible genetic familial connection and encouraging family testing and counseling are examples of genetic competency. In addition to educating families who may have long-QT syndrome, nurses can discuss the use of β-blockers and the likelihood of possible syncope, including syncope prevention.[8]

Malignant hyperthermia is an autosomal-dominant condition that can be life-threatening. This condition is associated with mutations in the *RYR1* gene in about half of cases, and other mutations have been found through testing.[9] Examples of a nurse's genetic competency with a patient who has malignant hyperthermia include obtaining the histories of the patient and the patient's family, analyzing genograms, educating the family about the abnormality, and discussing genetic testing for family members. In a preoperative setting, the nurse would inform the anesthesia provider that the patient or a member of the patient's family has malignant hyperthermia so anesthetic accommodations can be made to avoid complications associated with this abnormality.[9]

Marfan syndrome is an autosomal-dominant condition caused by mutations of the *FBN1* gene. Indications include abnormal findings of aortic dilatation and mitral valve regurgitation on magnetic resonance images and of long limbs, scoliosis, and pectus excavatum on physical assessment.[10] Nurses can demonstrate their genetic competency by assessing a patient's knowledge of Marfan syndrome, explaining the inheritance pattern, describing the treatment plan, and discussing family testing for the mutations.

Huntington disease is an autosomal-dominant progressive neurological condition that has its onset in adulthood. The disease is caused by abnormalities in the *HTT* gene on chromosome 4.[2] Nurses are in a unique position with patients with Huntington disease because the patients are usually admitted to a critical care unit after the diagnosis has been made and after family members have been tested to see if others in the family have the *HTT* gene. A genetically competent nurse will realize the genetic nature of the disease and foster communication between the patient, the patient's family, and genetic experts if this relationship has not already been established. Nurses can maintain awareness that members of a patient's family who have tested positive for *HTT* may be the caregivers of the patient and may need

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Table 1 Essential genetic competencies for nurses[a]

Professional responsibility
1. Recognize when one's own attitudes and values related to genetic and genomic science may affect care provided to clients.
2. Advocate for clients' access to genetic/genomic services and or resources including support groups.
3. Examine competency of practice on a regular basis, identifying areas of strength, as well as areas in which professional development related to genetics and genomics would be beneficial.
4. Incorporate genetic and genomic technologies and information into registered nursing practice.
5. Demonstrate in practice the importance of tailoring genetic and genomic information and services to clients based on their cultural, religion, knowledge level, literacy, and preferred language.
6. Advocate for the rights of all clients for autonomous, informed genetic and genomic-related decision making and voluntary action.
Scope of practice
Applying integrated genetic and genomic information
1. Demonstrate an understanding of the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.
2. Be able to construct a minimum of three-generation family health history information.
3. Be able to construct a pedigree from collected family history information using standardized symbols and terminology.
4. Be able to collect personal, health and developmental histories that consider genetic, environmental and genomic influences and risks.
5. Be able to conduct comprehensive health and physical assessments which incorporate knowledge about genetic, environmental, and genomic influences and risk factors.
6. Be able to critically analyze the history and physical assessment findings for genetic, environmental and genomic influences and risk factors.
7. Be able to assess the client's knowledge, perceptions and responses to genetic and genomic information.
8. Develop a plan of care that incorporates genetic and genomic assessment information.
Identification
1. Identify clients who may benefit from specific genetic and genomic information and or services based on assessment data.
2. Identify credible, accurate, appropriate, and current genetic and genomic information, resources, services and or technologies specific to given clients.
3. Identify ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genetic and genomic information and technologies.
4. Define issues that undermine the rights of all clients for autonomous, informed genetic and genomic-related decision making and voluntary action.
Referral activities
1. Facilitate referrals for specialized genetic and genomic services for clients as needed.
Provision of education, care and support
1. Provide client with interpretation of selective genetic and genomic information or services.
2. Provide client with credible, accurate, appropriate, and current genetic and genomic information, resources, services and or technologies that facilitate decision making.
3. Use health-promotion disease-prevention practices that *consider genetic and genomic influences on personal and environmental risk factors; *incorporate knowledge of genetic and genomic risk factors (eg, a client with a genetic predisposition for high cholesterol who can benefit from a change in lifestyle that will decrease the likelihood that the genetic risk will be expressed.)
4. Use genetic and genomic based interventions and information to improve clients' outcomes.
5. Collaborate with health care providers in providing genetic and genomic health care.
6. Collaborate with insurance providers/payers to facilitate reimbursement for genetic and genomic health care services.
7. Perform interventions/treatments appropriate to clients' genetic and genomic health care needs.
8. Evaluate impact and effectiveness of genetic and genomic technology, information, interventions, and treatments on clients' outcome.

[a] From Consensus Panel on Genetic/Genomic Nursing Competencies.[7]

a referral for counseling, because they are witnessing what their genetic makeup holds for the future.

Although colorectal cancer has many causes, one cause is familial adenomatous polyposis, which has an onset during adulthood. Familial adenomatous polyposis is autosomally dominant and is linked to mutations of the *APC* gene on chromosome 5.[10] Manifestations include numerous polyps, which result in colon resections and complications.[10] Nurses working with patients with familial adenomatous

Table 2 Educational Web sites for critical care nurses

National Genome Research Institute, National Institutes of Health, home page http://www.genome.gov
Descriptions of genetic tests and lists of testing locations http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests
Genetics Home Reference, home page—a guide to genetic disorders http://ghr.nlm.nih.gov/
Pharmacogenomics Knowledge Base, home page http://www.pharmgkb.org/
Warfarin dosing site to assist in calculating doses http://www.warfarindosing.org/Source/Home.aspx
Oncology Nurses Society, home page http://www.ons.org
Genetics/Genomics Competency Center for Education, home page http://www.g-2-c-2.org
Public Health Genomics, Centers for Disease Control and Prevention, home page http://www.cdc.gov/genomics/resources/diseases/index.htm
International Society of Nurses in Genetics, home page www.isong.org

Table 3 Conditions with genetic links common in critical care patients

Blood clotting disorders
Breast cancer
Familial hypercholesterolemia
Heart disease
Hemochromatosis
Huntington disease
Hypertrophic cardiomyopathy
Long-QT syndrome
Lung cancer
Malignant hyperthermia
Marfan syndrome
Sickle cell disease
Stroke
Types of colon cancers
Types of leukemia

polyposis show genetic competency by assessing the knowledge of the patients' family members of the disease and by discussing inheritance patterns, screening techniques, and disease progression.

Pharmacogenomics

Pharmacogenomics, the study of how medications target a specific individual's genotype, is advancing in ways that will affect all nurses.[11] For some conditions, medications are prescribed to target a patient's gene alterations, or alleles. For example, patients with breast cancer who test positive for the human epidermal growth factor receptor 2 will have a different medication regimen than will patients who test negative for the receptor. Although testing positive for the receptor is not necessarily a hereditary condition, overexpression of the receptor protein occurs at the cellular DNA level and warrants treatment with the medication herceptin in addition to traditional breast cancer medications.[12] A nurse's knowledge of the hereditary factors of the *BrCA2* gene and pharmacogenomics knowledge of herceptin will be imperative as the nurse and patients with breast cancer discuss referral and counseling needs and information about medications.

The pharmacogenomics of treatment of human immunodeficiency virus (HIV) disease is advancing. The Food and Drug Administration has approved genetic testing before patients receive a common HIV medication called abacavir. This medication should be given only to HIV-positive clients who do not have the HLA-B*5701 allele.[13] Patients with this allele are at a significantly higher risk for hypersensitivity reactions to abacavir than are patients without the allele. Hypersensitivity reactions can occur within 2 weeks of beginning treatment with abacavir. Signs and symptoms of the reactions include rash, fever, nausea, vomiting, and respiratory difficulty. If a patient is positive for HLA-B*5701, medications such as carbamazepine and allopurinol should also be avoided to decrease the likelihood of sensitivity reactions.[13] Nurses with pharmacogenomics competency use this information in administering medication, teaching patients, and planning discharge from the hospital.

Patients with chronic myeloid leukemia are routinely tested for the presence of the Philadelphia chromosome, which is formed by translocation of parts of chromosome 9 and chromosome 22.[10] The medication imatinib (Gleevec) specifically targets tumors positive for the Philadelphia chromosome by turning off the protein BCR-ABL.[14] A new medication, nilotinib, is also specific for chronic myeloid leukemia cells positive for the Philadelphia chromosome but can cause an increase in the QT interval.[15] Nurses who are aware of this possible side

effect show pharmacogenomics competency by obtaining thorough patient histories and doing genogram analysis to rule out preexisting long-QT syndrome. These nurses also educate patients with chronic myeloid leukemia who are taking nilotinib to avoid food and medications, such as grapefruit juice, ciprofloxin, fluconazole, and some HIV medications, that inhibit the CYP3A4 pathway because these foods and drugs can increase the side effects of nilotinib.[16]

Anticoagulants, specifically warfarin, are often used in critical care. A continuing struggle associated with warfarin treatment is achieving therapeutic levels as soon as possible without causing adverse effects.[17] Variants of the *CYP2C9* and *VKORC1* genes, found on chromosome 11, affect warfarin metabolism and patients' sensitivity to the medication.[18] The Food and Drug Administration now recommends that information about these alleles be considered in calculating the initial dose of warfarin in efforts to decrease side effects and achieve therapeutic levels quickly.[18] A link to the dosing site is provided in Table 2. Although patients are highly encouraged to have genotyping before beginning treatment with warfarin, the typing is not currently required by the Food and Drug Administration.[18] Debates on the cost-benefit analysis of testing before dosing continues, and nurses can expect to hear more about the pharmacogenetics of warfarin.

In another example, tests can be performed to determine if patients are fast or slow metabolizers of certain medications by determining which enzymes of the cytochrome P450 pathway the patients have. Alleles alter the activity of the enzymes of this pathway, resulting in slower or faster than expected metabolism of some drugs. Commonly used medications that may be affected include fluoxetine, haloperidol, metoprolol, and warfarin.[11] Poor metabolizers are at risk for toxic levels of medication and decreased excretion. In contrast, rapid metabolizers excrete the medication before the complete therapeutic effects are realized.[10] Such patients often state ineffectiveness of a drug, especially when the drug is the pain medication codeine or an antidepressant that is not helping the patient at the standard starting dose.

The metabolism of the β-blocker metoprolol, a commonly used medication in critical care, is a good example of alleles for the enzymes in the P450 pathway. β-Blockers are primarily metabolized by CYP2D6, an enzyme coded for by 1 of the 6 major genes of the cytochrome P450 pathway.[19] Many variants of the gene for CYP2D6 affect the rate at which metoprolol is metabolized. The most common allelic combination yields slow metabolism of metoprolol, and therefore the serum concentration of the medication quickly reaches levels that can cause adverse events.[19] Two other β-blockers, carvedilol and bisoprolol, also are metabolized through the P450 pathway. The necessity of genetic testing for patients before treatment with β-blockers is being debated, and studies are ongoing to determine the clinical relevance of this pharmacogenomics relationship.[19]

Nurses who are aware of the effect of variations in the cytochrome P450 pathway can advocate for their patients by requesting different dosing regimens or an alternative medication from the same class. Nurses should be mindful that slow metabolizers are at increased risk for adverse drug reactions and toxic effects. The time required for excretion of medications such as codeine differs between slow and fast metabolizers.[20] When a nurse sees a pattern of fast or slow metabolism of a medication they know is processed through the cytochrome P450 pathway, talking to the patient or the patient's physician about genetic testing would show genetic competence.

Summary

When the Human Genome Project began reporting results, it became apparent nursing would feel the impact of genetics and genomics research. The impact is not limited to a single area such as pharmacology or critical care nursing; rather, all fields of nursing feel the dramatic effects of these new areas of study. Nurses must remain proactive in examining the changes needed to ensure that nurses are competent in genetics and genomics. Strategies that critical care nurses can use to demonstrate genetics and genomics competencies in practice are summarized in Table 4. As science moves forward, nurses must be prepared to continue as career-long learners of genetic and genomic content in order to care fully for the patients of tomorrow. CCN

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Financial Disclosures
None reported.

Table 4 Strategies that critical care nurses can use to demonstrate genetic competence

Construct a 3-generational genogram that uses your patient's genetic condition
Provide an explanation of medications that are given on the basis of results of genetic tests
Discuss basic components of cellular genetics that contribute to genetic conditions
Offer patient education about the genetic component of a condition
Advocate for medication change and genetic testing when patients have indications of fast or slow drug metabolism
Offer patients referrals to genetic counselors or genetic clinics
Educate patients and their families about genetic testing done in the hospital

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CE Test

Test ID C1152: Essential Nursing Competencies for Genetics and Genomics: Implications for Critical Care

Learning objectives: 1. Understand the implications of genetics and genomics for critical care nurses 2. Identify the essential genetic competencies for nurses 3. Differentiate genetic causes for specific chronic illnesses and diseases

1. What best describes pharmacogenomics?
a. It addresses the association between a disease and a gene abnormality.
b. It is concerned with how a mutated gene interacts with other genes.
c. It is associated with the severity of disease expression.
d. It is concerned with how medication targets genes.

2. What was the primary emphasis when nurses formally began applying genetics to clinical practice?
a. Breast and lung cancers
b. Prenatal screening and newborn metabolic disease
c. Heart disease and stroke
d. Marfan and long-QT syndromes

3. What is correct about genetics and genomics in nursing?
a. Genetics is the study of gene function.
b. Genetic competencies replace existing standards of practice.
c. Genomics is the study of heredity.
d. Genetics is a part of all aspects of nursing.

4. Ensuring that anesthetic accommodations are made to avoid complications is an example of a nursing genetic competency for a patient with which disorder?
a. Long-QT syndrome
b. Malignant hyperthermia
c. Marfan syndrome
d. Huntington disease

5. Marfan syndrome is caused by mutations of which of the following genes?
a. *HBB*
b. *RYR1*
c. *FBN1*
d. *HTT*

6. In addition to traditional breast cancer medications, patients who test positive for the human epidermal growth factor receptor 2 should receive which of the following drugs?
a. Letrozole
b. Herceptin
c. Anastrozole
d. Exemestane

7. The Food and Drug Administration has approved genetic testing before patients receive which of the following human immunodeficiency virus medications?
a. Lamivudine
b. Zidovudine
c. Didanosine
d. Abacavir

8. Patients with the HLA-B*5701 allele are at increased risk for what adverse drug reaction compared with patients who do not have this allele?
a. Hypersensitivity reactions
b. Lactic acidosis
c. Severe hepatomegaly with steatosis
d. Immune reconstitution syndrome

9. Performing genogram analysis before nilotinib administration is done to rule out what preexisting condition?
a. Hyperkalemia
b. Diabetes mellitus
c. Long-QT syndrome
d. Hypercalcemia

10. Which alleles should be considered when calculating an initial warfarin dose?
a. *CYP2C9* and *CYP2D6*
b. *CYP2D6* and *VKORC1*
c. *CYP2C9* and *VKORC1*
d. *CYP2C9* and *RYR1*

11. Rapid metabolizers should be monitored for what risk?
a. Drug ineffectiveness
b. Increased adverse drug reactions
c. Drug toxicity
d. Decreased drug excretion

12. What is an example of applying integrated genetic and genomic information?
a. Developing a plan of care that incorporates genetic and genomic assessment data
b. Identifying patients who may benefit from specific genetic and genomic services based on assessment data
c. Facilitating referrals for specialized genetic and genomic services
d. Using genetic- and genomic-based interventions to improve patient outcomes

Test answers: Mark only one box for your answer to each question. You may photocopy this form.

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