Editor's Note: In this issue a new feature—Focus—begins. The inaugural series of articles to be published over a 2-year period is "Genomics for Health." Jean Jenkins, RN, PhD, FAAN, is the series editor. In this introductory article, Dr. Jenkins and colleagues describe the series and its importance for nurses globally.

Nurses and the Genomic Revolution

Jean Jenkins, Patricia A. Grady, Francis S. Collins

Purpose: *To increase nurses' genetics and genomics literacy through a series of articles focused on genomic research discoveries and their importance for nursing education, practice, policy, and research.*

Organizing Framework: *"Genomics for Health" is one of three themes, along with genomes to biology and genomes to society, emanating from applications of the Human Genome Project (HGP).*

Methods: *In this series of articles, nurse scientists who are experts in genetics and genomics sciences explain terminology, provide background information about the HGP, discuss clinical examples, and recommend changes in nursing practice, education, and research.*

Conclusions: *The HGP has already led to major changes in clinical practice, research, education, and policy, and even more dramatic changes are predicted for people throughout the world. Mastering this information is necessary for nurses globally because genomic information will ultimately pervade all of health care.*

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Since completion of the Human Genome Project in 2003 and subsequent ongoing efforts globally in genomic research, the practice of nursing and medicine internationally has been undergoing major change. The leading edge of genomic healthcare is already evident in patient care (Feetham & Williams, 2004; Guttmacher & Collins, 2002), with even more dramatic changes anticipated. The full potential will be gradually realized as knowledge continues to increase about the human genome, the physiologic consequences of genetic variations, and outcomes of gene interactions with the environment (Varmus, 2002). The applications of genomic discoveries in clinical care are expanding diagnosis, treatment, and prevention at an accelerating pace, and that knowledge will continue to improve the health of people throughout the world.

Nurses are pivotal in using information about the function, structure, and interactions of all the genes in the human body (i.e., genomics) with the goal of improved outcomes for everyone. This focus on health, rather than merely on disease, is creating important and profound changes in nursing education, practice, policy, and research. The vision of researchers and clinicians in genomic healthcare contains three themes: genomes to biology, genomes to health, and genomes to society. These themes constitute a framework for understanding how genomics will effect continued changes in healthcare (Collins, Green, Guttmacher, & Guyer, 2003; Guttmacher, Collins, & Drazen, 2004).

Over the next 2 years, a series of articles will be published in JNS about genomic research discoveries and their importance in nursing education, practice, policy, and research. The idea for this series, Genomics for Health, came from nurses who are leaders in genetics, because of their recognition of the significance of genetics and genomics research to nurses, as well as recognition of the important work already being done by nurses. These leaders, representing a collaborative effort across two U.S. federal agencies (National Institutes of Health and the Health Resources and Services Administration) agreed that this work should be brought together and disseminated to inform nurses globally. Authors of the series of articles are scholars and leaders in nursing and genetics or genomics, with diverse education,

Jean Jenkins, RN, PhD, FAAN, *Kappa*, Senior Clinical Advisor; **Francis S. Collins**, MD, PhD, Director; all at National Human Genome Research Institute; **Patricia A. Grady**, RN, PhD, FAAN, *Tau*, Director, National Institute of Nursing Research, National Institutes of Health, Bethesda, MD. Correspondence to Dr. Jenkins, National Human Genome Research Institute, National Institutes of Health, 31 Center Drive, Building 31, Room 4B09, Bethesda, MD 20892. E-mail: jean.jenkins@nih.gov

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experience, and cultural perspectives. Topics covered in the series will pertain to (a) areas that are influenced by genomic discoveries (e.g., emerging technologies, ethical issues, education; informatics); (b) all specialties (e.g., oncology, cardiovascular, psychiatric); and (c) the entire life continuum (e.g., preconception and prenatal, chronic dementing conditions). These articles will be designed to provide answers to questions of readers, to explain terminology, and to improve nurses' genetic literacy.

Mastering this information is necessary for all health care professionals to make the conceptual shift as genetic and genomic information ultimately pervades all health care. Key concepts, an introduction to terminology, a glossary of terms, and examples of why genomics is relevant for nurses now are introduced in this issue (see **Table**). We encourage you to take time to read all articles in the series. These articles will show how nurses are an essential part of this important effort to apply genomics to health.

The following cases illustrate how genomic information is increasingly permeating clinical, educational, and nursing research activities. We invite readers to reflect on their knowledge and practice, and to consider the potential ethical and societal implications in this new era of healthcare.

Example 1

Jan was recently diagnosed with non-Hodgkins lymphoma. She read about the use of a Lymphochip using microarray technology to provide a more sensitive molecular classification of lymphoid malignancies (Staudt, 2002; Dave et al., 2004). These studies showed that molecular markers such as gene expression and chromosomal rearrangements might identify people needing more aggressive cancer treatment, and might even indicate the possibility of targeted therapy (Kipps, 2002). This information is especially important to Jan because selection of her treatment is imminent.

Nurses have important responsibilities in assuring that Jan can give informed consent for the interventions. For example, nurses need to know enough to be able to describe the Lymphochip test process and results to Jan. On the basis of the test results, can you describe for her the rationale for the proposed targeted drug therapy, expected side effects, and long-term risks? Does Jan need to know about any issues of privacy or confidentiality related to this treatment?

Example 2

Todd is an undergraduate nursing student. He is working in a cardiovascular clinic and caring for a woman (BL) who at the age of 42 is recovering from a heart attack. BL reports that she is watching her diet, taking recommended medications, and exercising three times a week. BL brought with her a listing of family history information to discuss with the nurse practitioner (NP). She used a family history portrait tool available online to guide collection of information from her family about incidences of diseases in her family (http://www.hhs.gov/familyhistory). BL is concerned about a possible hereditary component to her health problems. She tells the NP and Todd that several family members have hypercholesterolemia and that several have died at young ages from heart attacks. Todd asks his faculty supervisor about the genetic factors related to health risks and interventions available to BL and her family.

As the educator in Todd's program, do you know what information and resources will assist Todd in interpreting family history, offering testing, and designing care options for BL (Cheek & Cesan, 2003)? Has your curriculum committee begun to integrate genomic concepts into all nursing courses?

Example 3

You are a nurse scholar interested in designing a program of research to integrate genomic principles into nursing studies (Williams, Tripp-Reimer, Schutte, & Barnette, 2004). Many opportunities for research are available to focus on the relationships among genetic factors and health outcomes. For instance, what is the best approach to prepare people to make decisions about genetic testing (Calzone et al., in press)? How can emerging technologies, such as clinical biomarkers, be used to answer research questions in clinical settings (Cashion, Driscoll, & Sabek, 2004)? Is a biobehavioral research model appropriate as a guide for inclusion of new genetics knowledge in nursing studies (Kang, 2003)?

Nurses can assume a wide range of responsibilities in advancing genetics and genomics from basic research to clinical application. Because of the importance of nurses in genomic healthcare, an effort is in process to identify the minimal nursing competencies related to genetic and genomic knowledge. A document to guide curriculum content and evaluation of nurse competency is under review with the goal of facilitating integration of genetics and genomics into curricula, the NCLEX exam, accreditation, and certification processes. Genomics is a principle-based science that can be integrated into curricula and practice. Nurses in the United Kingdom have recently completed this process and have published a document to be used as a framework for genetics education of nurses (Kirk et al., 2003).

Conclusions

Nurses can assimilate and integrate this burgeoning genetic and genomic science in concert with interdisciplinary colleagues. Genetics and genomics literacy for all nurses is important (Guttmacher, Jenkins, & Uhlmann, 2001) because sufficient specialists will not be available to respond to all questions about genetics and genomics. Patients and families expect healthcare providers to have this knowledge. Primary care providers will often be the point of contact for the questions and concerns of patients and their families. Nurses, with a long tradition of being educators, and with sensitivity to emotional and psychological issues and advocacy, are ideally suited to address the emerging needs of patients, families, and communities. Through education, research, and clinical applications, nurses can

Table. Glossary of Terms Related to Genomics

Allele. One of the variant forms of a gene at a particular locus, or location, on a chromosome. Different alleles produce variation in inherited characteristics such as hair color or blood type. In an individual, one form of the allele (the dominant one) may be expressed more than another form (the recessive one).

Autosome. Any chromosome other than a sex chromosome. Humans have 22 pairs of autosomes.

Base pair. Two bases which form a "rung of the DNA ladder." A DNA nucleotide is made of a molecule of sugar, a molecule of phosphoric acid, and a molecule called a base. The bases are the "letters" that spell out the genetic code. In DNA, the code letters are A, T, G, and C, which stand for the chemicals adenine, thymine, guanine, and cytosine, respectively. In base pairing, adenine always pairs with thymine, and guanine always pairs with cytosine.

Carrier. An individual who possesses one copy of a mutant allele that causes disease only when two copies are present. Although carriers do not have the disease, two carriers can produce a child who has the disease.

Chromosome. One of the threadlike "packages" of genes and other DNA in the nucleus of a cell. Different kinds of organisms have different numbers of chromosomes. Humans have 23 pairs of chromosomes, 46 in all: 44 autosomes and two sex chromosomes. Each parent contributes one chromosome to each pair, so children get half of their chromosomes from their mothers and half from their fathers.

Codon. Three bases in a DNA or RNA sequence which specify a single amino acid.

Deletion. A particular kind of mutation: loss of a piece of DNA from a chromosome. Deletion of a gene or part of a gene can lead to a disease or abnormality.

DNA. Deoxyribonucleic acid, the chemical inside the nucleus of a cell that carries the genetic instructions for making living organisms.

DNA sequencing. Determining the exact order of the base pairs in a segment of DNA.

Epigenetic. Nonmutational phenomena, such as methylation or histone modification, that may modify the expression of a gene.

Exon. The region of a gene that contains the code for producing the gene's protein. Each exon codes for a specific portion of the complete protein. In some species (including humans), a gene's exons are separated by long regions of DNA (called introns or sometimes "junk DNA") that have no apparent function.

Gene. The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein.

Genetic. Inherited; having to do with information that is passed from parents to offspring through genes in sperm and egg cells.

Gene mapping. Determining the relative positions of genes on a chromosome and the distance between them.

Genetic screening. Testing a population group to identify a subset of individuals at high risk for having or transmitting a specific genetic disorder.

Genetic susceptibility. An inherited increase in the risk of developing a disease.

Genetic testing. Analyzing DNA to look for a genetic alteration that might indicate an increased risk for developing a specific disease or disorder.

Genome. All the DNA contained in an organism, which includes both the chromosomes within the nucleus and the DNA in mitochondria.

Genomics. Study of the functions and interactions of all the genes in the genome, including their interactions with environmental factors.

Genotype. The genetic identity of an individual that does not show as outward characteristics.

Haplotype. A group of nearby alleles that are inherited together.

Heterozygous. Possessing two different forms of a particular gene, one inherited from each parent.

Homozygous. Possessing two identical forms of a particular gene, one inherited from each parent.

Human Genome Project. An international research project to map each human gene and to completely sequence human DNA.

Inherited. Transmitted through genes from parents to offspring.

Intron. A noncoding sequence of DNA that is initially copied into RNA but is cut out of the final RNA transcript.

Karyotype. The chromosomal complement of an individual, including the number of chromosomes and any abnormalities. The term is also used to refer to a photograph of an individual's chromosomes.

Linkage. The association of genes and markers that lie near each other on a chromosome. Linked genes and markers tend to be inherited together.

Mendelian inheritance. Manner in which genes and traits are passed from parents to children. Examples of Mendelian inheritance include autosomal dominant, autosomal recessive, and sex-linked genes.

Microarray technology. A new way of studying how large numbers of genes interact with each other and how a cell's regulatory networks control vast batteries of genes simultaneously. The method requires use of a robot to precisely apply tiny droplets containing functional DNA to glass slides. Researchers then attach fluorescent labels to DNA from the cell they are studying. The labeled probes are allowed to bind to complementary DNA strands on the slides. The slides are put into a scanning microscope that can measure the brightness of each fluorescent dot; brightness reveals how much of a specific DNA fragment is present, an indicator of how active it is.

Multifactorial. Caused by the interaction of multiple genetic and environmental factors.

Mutation. A permanent structural alteration in DNA. In most cases, DNA changes either have no effect or cause harm, but occasionally a mutation can improve an organism's chances of surviving and passing the beneficial change on to its descendants.

Pedigree. A simplified diagram of a family's genealogy that shows family members' relationships to each other and how a particular trait or disease has been inherited.

Penetrance. The likelihood that a person carrying a particular mutant gene will have an altered phenotype.

Pharmacogenomic. Generally refers to the inherited variability in drug metabolism and disposition; allows for optimal choice and dose of drugs.

Phenotype. The observable traits or characteristics of an organism, for example, hair color, weight, or the presence or absence of a disease. Phenotypic traits are not necessarily genetic.

Polymorphism. A common variation in the sequence of DNA among individuals.

(*Continued*).

Table. Continued.

Selection bias. An error in choosing the individuals or groups to participate in a study. Ideally, participants in a study should be very similar to one another and to the larger population from which they are drawn (for example, all people with the same disease or condition). If they have important differences, the results of the study might not be valid.

Single nucleotide polymorphism (SNP). Common but minute variations that occur in human DNA at a frequency of one every 1,000 bases. These variations can be used to track inheritance in families. SNP is pronounced "snip."

Variation. Most of any one person's DNA, about 99.9 percent, is exactly the same as any other person's DNA. (Identical twins are the exception, with 100 percent similarity). Differences in the sequence of DNA among individuals are called genetic variation.

Sources and additional information:
http://www.genome.gov10002096
http://www.genome.gov/10000202
http://www.cancer.gov/dictionary/
Guttmacher, Collins, & Drazen, 2004.

accelerate the pace of integrating genomics into options for care, thereby contributing significantly to reshaping and optimizing health care.

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