

# Nursing Leadership in Genomics for Health and Society

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***Purpose:** This article is part of the series regarding genomics and nursing practice, science, education, and policy. Issues in genetic testing, genetic information and the lessons learned through applications of genetic and genomic science are analyzed and discussed.*

***Framework:** Scientists, scholars, and members of the public have articulated a vision to guide genomics research and scholarship. The three overarching themes of this conceptual framework are genomes to biology, genomes to health, and genomes to society.*

***Conclusions:** Nurses can promote the use of genomic research technologies and information in the context of health, biology, and society, as well as in nursing research, practice, education, and policy.*

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Historically, research in human genetics was conducted by a small number of scientists and health professionals who specialized in studying and caring for individuals and families who were known or believed to have one of the rare, inherited, “genetic disorders.” In recent years, however, this situation has changed. As understanding of the human genome has increased, so too has the understanding that genes are important in virtually all human diseases, from relatively rare Mendelian disorders such as Huntington disease (HD), cystic fibrosis, and sickle cell disease, to common diseases such as cardiovascular disease, cancer, and diabetes. Evidence now indicates that genes not only can cause diseases, but they also affect disease susceptibility and resistance, prognosis and progression, and responses to illness and their treatments.

The scientific publication at the completion of the Human Genome Project (the mapping and sequencing of all human genes) in April 2003 and the analysis of these data is the underlying reason for the rapidly increasing understanding of the relationships among genes, the environment, health, and disease (Collins, Green, Guttmacher, & Guyer, 2003). The Human Genome Project was an international effort with scientists from 20 research centers in six countries: China, France, Germany, Japan, United Kingdom, and United States (International Human Genome Sequencing Consortium, 2001). The completion of the Human Genome Project denoted not the end, but the beginning of the genomic era and a new trajectory of discovery expected to increase exponentially through the coming decades. The publication occurred 50 years to the month after the landmark publication of Watson and Crick’s report of the double helical structure of DNA in April 1953 (Watson & Crick, 1953).

To put this achievement in context, some have said that the knowledge gained about genomics is to this century what the knowledge of infectious disease was to the last century (Guttmacher, 2002) and what Mendeleev’s publication of the periodic table of the elements was to the 19th century (Lander, 2001).

In 2003 Collins and colleagues articulated a vision for the future of genomics (Collins et al., 2003). This vision resulted from the deliberations of hundreds of researchers, clinicians, and others during a series of workshops over a 2-year period. It is a conceptual framework for genomics research and scholarship, with three overarching themes: (a) genomes to biology, (b) genomes to health, and (c) genomes to society, all grounded on the information from the Human Genome Project. The first theme, Genomes to Biology, is focused on research that will be foundational in future biology. Increased understanding to result from this focus includes the structure of genomes, human as well as other organisms; genetic variations in humans and other organisms; the functional elements of the genome; how genome-encoded functions are integrated to perform both cellular and organismal functions; and how genomes change and take on new

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functions. The second theme, Genomes to Health, is focused on the underlying mechanisms for human health and disease processes, including gene-gene, gene-environment, and their interactions. This knowledge will be used to develop new tools and technologies to diagnose, treat, and prevent human diseases. The third theme, Genomes to Society, is the foundation for research to improve the appropriate use and interpretation of genetic and genomic information and technologies. Issues such as privacy and fair use of genetic information, genetic nondiscrimination and access to desired genetic technologies and services will be integral to research and policy development related to this theme (Collins et al., 2003).

An era of utilizing genomic information in all aspects of basic and applied research and health care has begun. Roles and activities of nurses in this new era include: (a) active participation in genomic research, including the study of the biologic, behavioral, family, ethical, legal, and social implications; (b) the development and integration of genomic technologies in health care and other settings; (c) the interpretation and use of genomic information and efforts to protect against the misuse of information; and (d) assuring that genomic research, technologies, and information be viewed in the context of other biopsychosocial factors and cultural norms so that reinforcement of the concept of genetic determinism is not an unintended by-product of this recent emphasis on genomics.

This article is part of the series, Genomics for Health, focused on the implications of genomics for nursing practice, science, education, and policy. Issues in genetic testing technologies and the increasing availability of genetic information in health and nonhealth related settings will be presented along with the concept of genetic exceptionalism, whether genetic information is particularly sensitive information, and whether more needs to be done to assure that it is kept private. Some of the lessons learned as genetic research and knowledge have advanced will be discussed. Finally, the implications of these new technologies and information for leadership in nursing research, education, practice, and policy are described.

## Genomics and New Conceptualizations of Health and Illness

Why genomics and not genetics? The term genomics was first used in 1987 (McKusick & Ruddle, 1987). Previously, the term "genetics" had been used in the study of individual genes as the basis for relatively rare single-gene disorders. The term "genomics" refers to the study of all of the genes in the human genome together, including their interactions with each other and the environment. Some have used the term genetics and genomics interchangeably; however, Guttmacher and Collins (2002) said that genetics is "the study of single genes and their effects" and genomics is "the study not just of single genes, but of the functions and interactions of all the genes in the genome" (p. 1512).

The completion of the Human Genome Project (HGP) has made the study of genomics possible. The study of genomics today and in the future will take into consideration the study of the whole genome and its variations and internal interactions, and also the study of the genome's interactions with the environment and other psychosocial and cultural factors. Lack of such a broad view and comprehensive approach would result in failure to fully understand, appreciate, and interpret the genome, its functions, and its contribution to health and disease. Detailed discussion of some specific examples of these discoveries will be covered in subsequent articles in this series.

Some scholars and researchers have reported that the findings from genomic research and advances in genetic technologies and information obtained through genetic testing require a reframing of how health professionals and the lay public think of the continuum of health and illness, and even the concept of disease (Guttmacher & Collins, 2002). The application of genomics creates a central challenge for health care and public health to evolve from the model of intervention after disease or loss of function to more predictive models of interventions, before the onset of disease or loss of function (Feetham, 1999, 2000; Varmus, 2002). Childs (2003) contended that as the understanding of genomics increases, so too will the understandings of the mechanisms of disease that will contribute to more targeted and individualized care. The way in which diseases are named, categorized, described, and ultimately how they are treated, managed, or prevented will also change.

As knowledge of the mechanisms of disease increases, individuals and families will be faced with a reframing of their concepts and experiences with diagnosis, treatment, and prevention to understand the influence of genes, the environment, and behavior. The boundary between health and chronic illness might become blurred, because the degree of influence of genes on health and illness might vary from significant (25% to 50% risks), as observed in single-gene disorders such as sickle cell disease and Huntington Disease, to far lower risks (2% to 3% increase) as seen in most forms of diabetes, heart disease, and hypertension (Childs, 2003; Rolland & Williams, in press).

For individuals and their families choosing to obtain genetic information, the findings might result in the need to extend the concept of "illness time" phases to include preawareness or lack of knowledge about a genetic risk state, or in some cases, a nonsymptomatic phase (Rolland 1999; Rolland & Williams, in press; Street & Soldan, 1998). The preawareness risk state refers to the time before a person knows he or she has a genetic risk factor. The risk might become known through awareness of a family history of disease or by having a genetic test. The nonsymptomatic phase occurs when the person is aware of the genetic risk, but remains nonsymptomatic. This phase might extend from years to decades until the onset of the disease, such as in HD. The phase might extend throughout the remainder of a person's life if the genetic risk does not result in the expression of the disease, such as in iron overload, cardiomyopathy,

or even breast, ovarian, or colon cancer (Rolland, 1999; Rolland & Williams, in press). Knowledge of risk state might require interventions for individuals and families to respond to the increased awareness of risk, the newly gained genetic risk information, or the earliest (previously denied or unobserved) occurrence of symptoms. Family members might need to begin to deal with anticipatory loss, to accept increased surveillance, to adhere to changes in health behaviors, or to accept interventions that could potentially delay the onset or progression of the disease (Rolland, in press).

Of particular importance for nurses is understanding the meaning and interpretation as well as the limitations of genetic information. Genetic information is not deterministic, that is, having a genetic mutation does not allow prediction with certainty that a disease is present or will develop. Furthermore, the lack of a demonstrated genetic mutation does not mean that a person has no disease present or no risk to develop disease, a concept that is particularly difficult for many people to understand. Understanding one's genetic risk could result in behaviors to reduce risk, such as increased surveillance and changes in diet and exercise. On the contrary, a person could interpret the presence of a genetic predisposition to be deterministic, and adopt a more fatalistic attitude about the likelihood of their developing a disease no matter how much they change their behavior. Health care providers should assure that genetic information is interpreted and used in the context of what is known about the person, the family, their sociocultural perspectives, and their other risk factors. Only then will this information be likely to result in benefit and not harm to individuals and families.

### Genetic Testing

Genetic testing in the US started in the late 1950s as an experimental laboratory test, done as often on research animals as on humans (personal communication, Zellweger, 1976). At that time, cytogenetic testing was a "blunt instrument" that could detect the presence or absence of a whole chromosome as seen in Down syndrome (trisomy 21) and Turner syndrome (45, X0). In the 1970s, as a result of new staining technologies, cytogenetic testing could identify smaller and smaller deletions or additions in chromosomes as could be seen in Cri du Chat syndrome (5p deletion) or Prader Willi syndrome (15q deletion). In the 1980s new recombinant DNA technologies were often used for genetic tests called linkage studies, which allowed researchers and clinicians to begin to track disease-causing mutations along with known genetic markers. In the 1990s far more sophisticated testing technologies facilitated discovery of very specific genetic mutations: individual base pair deletions, additions, and substitutions associated with diseases such as breast, ovarian, and colon cancer, Alzheimer disease; and repeat sequences of base pairs as seen in HD, spinal muscular atrophy, and Fragile X syndrome. Today, genetic tests are available for more than 1000 genetic disorders. Tests

are used for preconception, prenatal, and newborn screening; predispositional and presymptomatic testing; diagnostic confirmation; prognostic information; and also in choosing optimal therapeutic alternatives, as in pharmacogenomic testing (Burke, 2002; Patenaude, Guttmacher, & Collins, 2002).

Iceland, the United Kingdom, and other countries are creating national bio banks of DNA and other data to advance the understanding of genotype and phenotype relationships and in some instances the contributions of the environment (<http://drosenthal.org/dbinst.html>). Data from these countries that have more homogeneous populations and national health programs that provide infrastructure to these "bio banks" can be useful information to nurses in other countries about the complex challenges including privacy, interpretation of genetic testing, and disease management (Bragadóttir, Björnsdóttir, Thorhallsdóttir, & Erlendsdóttir, 2004).

The significance of the family history as a source of genetic information cannot be overstated. Rich and colleagues reported that the family history is considered the most important tool for diagnosis and risk assessment in health care genetics, and it is a critical tool in the use of predictive genetic testing in primary care (Rich et al., 2004). For example, cholesterol screening should occur at a younger age for men and women with a family history of high cholesterol (U.S. Preventive Services Taskforce, 2002); yet health professionals and the lay public might not recognize the importance of early testing (Prendergast, Bunney, Roberson & Davis, 2004).

Pharmacogenomic testing will likely become common. Evidence has shown that some drugs have different levels of effectiveness in different people. In addition, adverse reactions to drugs have become one of the leading causes of morbidity and mortality in the developed world (Lazarou, Pomeranz, & Corey, 1998; Prows & Prows, 2004). Pharmacogenomic tests will become routine or commonplace in the delivery of health care as they become available to identify people who will more likely respond to a certain drug treatment, and perhaps more importantly, to identify those who are at increased risk for an adverse or toxic reaction to certain pharmaceuticals.

For example, the  $\beta_2$ -adrenergic receptor gene is the target of medications often used in the treatment of asthma. A study showed a relationship between genotype in the  $\beta_2$ -adrenergic receptor gene and the therapeutic response to Albuterol in children with asthma. The response range was 10% for children with two glycine amino acids (*GlyGly*) for position 16 in the gene, 25% for children with one arginine and one glycine amino acid (*ArgGly*) in the gene, and 60% in children with two arginine mutations (*ArgArg*) (Martinez, Graves, Baldini, Solomone, & Erickson, 1997). Thus, this study showed that the effectiveness of Albuterol appeared to be highest in children with the *ArgArg* genotype. However, a subsequent study by Palmer, Silverman, Weiss, and Drazen (2002) showed that the children with *ArgArg* genotype had a significant decrease in the response to the drug with repeated

use, but those with *GlyGly* had no decrease in effectiveness with repeated use.

Although application of genetic testing for response to Albuterol is not currently a common practice, findings such as these will stimulate the increased use of pharmacogenomic testing. The choice and dose of therapeutic agents for common diseases, such as deep-vein thrombosis, cancer, diabetes, depression, and heart disease, will increasingly depend on this type of pharmacogenomic testing.

### Genetic Exceptionalism

In the use of genetic science in practice, one concept for attention is genetic exceptionalism. Implicit in this concept is that genetic information is inherently unique and should receive special consideration and be handled separately and perhaps differently from other types of personal and clinical information (Gostin & Hodge, 1999; Green & Botkin, 2003; Ross, 2000, 2001; Suter, 2001).

Several factors underlie this perspective. Genetic information is a unique identifier that is specific to an individual, except for identical twins. Unlike blood pressure, hemoglobin, or kidney function tests, genes do not vary or change each time they are measured. Genetic information is heritable, shared through generations, and thus it is relevant to family members, including ancestors and descendants. Because of the individual and intergenerational nature of genetic information, the interpretation and dissemination of this information within a family can have psychological and social consequences for the whole family (Feetham, 1999). It can heighten or relieve anxiety and alter family relationships positively or negatively. Genetic information can be obtained to identify a risk state or allow prediction of future disease. It might be used to encourage people to change health behaviors to reduce personal risk. At the same time it has the potential to be used to stigmatize and discriminate against certain individuals, families, or groups (Ross, 2001; Suter, 2001).

Some have argued that most of the characteristics of genetic information are also true with other medical information such as HIV status (Ross 2001; Suter, 2001; Task Force on Genetic Information and Health Insurance, 1993). Although this interpretation might become more generally accepted, one cannot be sure under current conditions how genetic information will be used. Early in the genomic era, until such knowledge is more pervasive and applied to everyone, the issue of genetic exceptionalism is relevant and should be considered. Concerns about genetic exceptionalism, however, might prove to be transitory and an interim issue of concern.

## Lessons Learned

As a result of improved understanding of the human genome, nurses have learned some important lessons that are useful to consider for movement into the genomic era. Some of these lessons involve new understandings and complexities.

### Not So “Simple Genetic Disorders”

An increased understanding of the genomics and genetics of cystic fibrosis (CF) has yielded many interesting and unanticipated findings. The gene associated with CF (cystic fibrosis transmembrane conductance regulator [CFTR]) was first reported in 1989. It was identified on chromosome number 7 and was found to be about 250,000 DNA base pairs in length. At the time, a single mutation, a three base-pair deletion ( $\Delta F508$ ), was found to account for about half of the people in the United States with CF. Also at the time, scientists believed that another few mutations in the gene might be discovered and the genetics of this so-called “simple Mendelian disorder” would be known.

More than a decade later, over a thousand mutations have been described in the CF gene, and these mutations still account for only about 90% of “classical CF” in the United States. A genetic test for CF mutations has been developed and is beginning to be widely used for preconception, prenatal, and postnatal diagnostic testing. Use of the test in a much more diverse population has resulted in many unexpected findings. First, not all people with “classical CF” have been found to have mutations in CFTR. Second, people with the same CFTR mutations can have quite different courses of disease (variable expression), even when they are from the same family, thus making prognostic assertions based on genotype likely to be extremely flawed. Third, the frequency of CF mutations varies substantially from one population group to another, for example, Ashkenazi Jewish, 1 in 29; European Caucasian, 1 in 29; Hispanic American, 1 in 46; African American, 1 in 65; Asian American, 1 in 90 (American College of Obstetrics and Gynecology, 2001; www.geneclinics.edu). This range results in variable testing sensitivity, specificity, and predictive value of the genetic test. Fourth, not all people who have two mutations in their CFTR gene have “classical CF.” Relatively healthy people have been found to have two CF mutations, including men whose only health problem has been infertility because of congenital absence of the vas deferens.

As more genes are being discovered and genetic tests are provided to large and diverse populations, similar unexpected findings continue to occur. Thus, what was once believed to be a “simple genetic disorder” of autosomal recessive inheritance three or four decades ago is now understood to be more complex, requiring more sophisticated knowledge and expertise for interpretation. In addition to its complexity has come a greater understanding of the molecular biology, the biologic functioning, and possible targets for therapeutic interventions for this disease.

### Penetrance—A Moving Target

Another issue that has become increasingly challenging is the interpretation of the meaning of the presence or absence of disease-causing mutations, how to interpret the penetrance of mutations in various genetic disorders, and how this information should best be used.

The concept of penetrance—the chance that a person who has altered gene(s) will have or develop the disease—sometimes seems to be a moving target. For decades, people believed that in autosomal-dominant disorders (when only a single altered gene is necessary to get the disease) if you inherited an alteration, you would one day, ultimately, develop the disease. Early gene-discovery studies often take place in so-called “high-risk” families, in which multiple family members in multiple generations are affected with a certain disease. Enlisting the participation of members of these families can make the discovery of the gene and its mutations somewhat more straightforward. However, as a result of this “selection bias,” estimations of the penetrance of the mutation provided by the results of the initial studies are often higher than are those in subsequent studies in which lower-risk families and members of the general population are recruited. Unfortunately, the later publications often receive less media attention and the knowledge and interpretation about penetrance can remain faulty for many years.

This example occurred in the discovery and description of breast and ovarian cancer genes, BRCA1 and 2, in which early studies indicated a penetrance of 85% to 90%, but later studies showed a 27% to 55% penetrance (Satagopen et al., 2001; Struewing et al., 1997; Wacholder et al., 1998). The complexity of interpreting risk status and the changing information of penetrance was evident in a report of a meta-analysis of 22 BRCA1 or BRCA2 and ovarian cancer risk studies, unselected for family history (Antoniou et al., 2003). These studies showed the significance of family history in addition to the presence of a mutation when determining risk and age-specific penetrance (Antoniou et al., 2003; Chatterjee et al., 2001; Satagopan et al., 2001).

Healthcare providers should understand that risk information can change over time and that the reports of penetrance in the initial studies and announcements of a gene discovery might be different, and often lower, in subsequent studies (Feetham, 1999). Healthcare providers should continue to monitor the scientific literature to provide the most accurate and up-to-date information to patients and their families. Healthcare providers and families also need assistance to understand that not having a mutation cannot be interpreted as having no risk to develop a disease. For example, even if a woman does not test positive for BRCA1 or BRCA2 she still has the national risk estimate of about 1 in 8 women in the US (approximately 13.3%) who will develop breast cancer during her lifetime. To add to the complexity, an increasing list of genes likely will be found to have mutations associated with breast cancer.

### Too Much Information

Obtaining genetic information can result in learning unexpected and sometimes unwelcome information. For example, whenever a family genetic study is undertaken as a part of research or clinical care, one risk that the family must be informed about is the possibility that misattributed pa-

ternity might be uncovered. The discovery and subsequent disclosure (inadvertent or not) of such information is likely to disrupt family relationships and possibly change family dynamics.

Another possibility is unanticipated discovery of an individual's health risks. One example of such an occurrence was the outcome of early research related to the gene for apolipoprotein E (APOE). APOE is essential for regulating lipid metabolism, but it also is involved in many other physiologic processes. It facilitates cellular uptake of cholesterol and lipoproteins. The APOE gene, located on the long arm of chromosome 19 (19q13.2), is polymorphic with three common alleles, e2, e3, and e4 (Mann et al., 2004). The APOE e4 allele was shown through association studies to be a genetic risk factor for cardiovascular disease, because it affects high circulating cholesterol levels, especially LDL (Menzel, Kladetzky, & Assmann, 1983). As studies of APOE continued, other researchers reported the observation that the e4 allele was also associated with an increased risk for late-onset Alzheimer disease (Corder et al., 1993). So when some people agreed to participate in research to examine the association of APOE e4 with cardiovascular disease, they may have inadvertently learned of their increased risk for Alzheimer disease.

The APOE gene is but one of many that will likely be found to contribute to common diseases with complex genetic underpinnings. It has been associated with risk of serum lipid elevations, coronary artery disease and Alzheimer disease. Many analytic challenges in studying the effects of APOE have been identified (Jarvik, 1997), including the fact that allele frequencies and effects can change with age, the gene might have pleiotropic effects, multiple alleles might affect the dosage of the genes, and interactions among risk factors can occur. Other common polymorphisms likely will be found in other important alleles with complex patterns of inheritance. Both researchers and clinicians will need to be prepared to understand and use such changing information.

### “One Size Fits All” Genetic Health Policies Might Not Work

Another challenge has been associated with attempts to develop health policies related to genetics and genomics for an extremely diverse population, such as that in the US. In the case of hereditary hemochromatosis, an inherited form of iron overload, the mutations in HFE genes were first discovered in patients and their families who were quite severely affected with iron overload. Those early studies indicated that HFE mutations accounted for most cases of iron overload in the US. Once the HFE mutations were discovered, a response was the call for immediate genotypic screening of the entire U.S. population. The reasons given were that this mutation was very common (1 in 10 Caucasians had been found to have a single mutation and about 1 in 400 were homozygous for two mutations—which should have made it one of the most common genetic disorders in the US). Because the disorder was thought to be so common and to lead to very serious diseases (diabetes, heart

disease, cirrhosis of the liver, liver cancer, arthritis, and impotence), and the treatment and prevention were inexpensive and easy (phlebotomy), it could become the “poster disease” for genetic testing. However, many studies of this condition (Cogswell et al., 2003; Waalen, Felitti, Gelbart, Ho, & Beutler, 2002) have yielded contrary information.

Large follow-up studies (Beutler et al., 2002; McLaren et al., 2003) showed that in addition to the fact that the penetrance rates were far lower than predicted by the early studies conducted in iron-overload patients and their families, the prevalence of the HFE mutation, C282Y, was very different among various populations (Caucasian, 1 in 10; Hispanic American, 1 in 30; African American, 1 in 50; and Asian American, 1 in 1000). Thus, the idea of genotypic screening of an entire diverse population, in which the sensitivity and specificity of the test were diminished and large segments of the population were not likely to benefit, resulted in moderation of enthusiasm for screening for this disorder in the U.S. population. These results overall showed that people could be “labeled” with hemochromatosis mutations and yet never become ill. Such a label could potentially have distressing effects, including possible loss of health or life insurance benefits because of stigmatization and discrimination, altered family relations, or psychological stress and anxiety.

## Implications for Nursing

### Health Professionals' Knowledge and Education in Genetics and Genomics

Genomics is a central science to be integrated into all areas of health care and public health professional education. No longer is it true that, “I can teach about diabetes, cancer, or Alzheimer disease, or I can teach about genetics.” The time has come that teaching about diabetes, cancer, or Alzheimer disease requires teaching genetics and genomics. Much is known about integrating genomics into practice, as reported from many countries (Ando, 2000; Expert Panel, 2000; Hager, 1999; Jenkins, Prows, Dimond, Monsen, & Williams, 2001; Kolb, Aguilar, Dinenberg, & Kaye, 1995; Mizoguchi, Yokoyama, Wada, Morita, & Ando, 2000; Scanlon & Fibison, 1995; Skirton et al., 1998).

With increasing knowledge of the application of genomics to human health and disease, all healthcare and public health professionals will be expected to have the knowledge, skills, and abilities to integrate genomics into their research, education, and practice. This knowledge underlies use of genetic technologies in health care and public health systems and for the understanding and interpretation of the information into optimal health and social policies. In the US, more than 11 million health professionals and thousands of healthcare and public health professional students require this knowledge. Because genomics affects all areas of health care and public health, the presence of any significant gap in knowledge might affect equity and access to desired services and the quality of health care. The integration of genetic infor-

mation into every aspect of health professional education is necessary. Numerous roles will be expected of health care providers in relation to genetic conditions: assessing and educating patients and their families on the risks of certain conditions, giving specific treatments or drugs because of genetic profiles, screening for genetic risk, and understanding and managing genetic information (Kirk, 2004; Lea & Monsen, 2003; Prows et al., 2003).

Curriculum recommendations are now in the literature in many countries for the integration of genetics and genomics into health professional programs (Expert Panel, 2000; Horner, Abel, Taylor, & Sands, 2004; Hetteberg & Prows, 2004; Lea, 2002; Arimori et al., 2000; Kirk, 1999; Mizoguchi et al., 2000; Skirton, Barnes, Curtis, & Walford-Moore, 1997). These recommendations include theories to guide the diffusion of genetics in the curriculum, specific content to be incorporated, and checklists to assure that the critical content is integrated across multiple courses. Such integration of genetics and understanding the broader more interactive concept of genomics will enable healthcare and public health professionals to be effective in the next era in health care and society by moving beyond the concept of genetic information as pertaining only to single-gene disorders in children.

Interdisciplinary education is a natural for genomics. National leaders from many countries and disciplines have identified core competencies to guide practice and educational programs development (Department of Health 2003; Feetham & Williams, 2004; National Coalition for Health Professional Education in Genetics, 2000). Interdisciplinary programs on genomics would promote common goals and understanding of genetic information as well as enhance communication among disciplines on the use and application of the information with individuals and families. An interdisciplinary approach to genetic education does not substitute for the integration of genetic and genomic information in curricula but it promotes more effective use of the information integrated across disciplines. Evidence of the significance of education in genetics and genomics is that agencies in the Department of Health and Human Services (e.g., Centers for Disease Control and Prevention, Human Resources and Services Administration, National Institute of Health) have made a commitment to the education of health professionals in genetics by providing funding for the education of healthcare and public health professionals with many grants focused on interdisciplinary education. Later articles in this series will provide a synthesis of issues in the education of healthcare and public health professionals in genomics.

### Research

In their synthesis of clinical nursing research, Hinshaw, Feetham, and Shaver (1999) noted that, although genetics is a burgeoning research and healthcare focus involving people of all ages, nursing research has a conspicuous absence of genetics topics. Typically, nursing research has been focused

on responses of individuals and families in their states of health and illness and on symptom management in disease states. Nursing research can be extended to address questions emerging from the HGP and other genomic and genetic research, including how genes affect health, the identification of genetic risk states and interventions to reduce risk, how genes affect disease progression and treatment, and responses to disease or genetic risk states (Feetham, 2000; Skirton & Williams, 2001).

The scientific knowledge and methods from nursing research can be applied to address important biological, behavioral, family, social, and ethical questions. The National Institute of Nursing Research, through its Intramural Program Summer Genetics Institute and funding of Institutional Research Training (T32) in genetics to expand the research capacity of students and faculty, has a strong science policy statement of the significance of nursing research and genomics and contributes to building a cadre of nurse scientists to conduct programs of research in genomics and health. With genomics as a central science, nurse scientists are encouraged to identify nursing research opportunities across NIH such as from the National Human Genome Research Institute, the National Cancer Institute, and other NIH Institutes supporting genetics and genomics research, and to examine the relevance of their programs of research to the research priorities of other institutes and funding agencies.

### Nursing Leadership and Genomics

For decades, a small number of nursing leaders from many countries have provided significant directions to nursing and other disciplines in genetics (Arimori et al., 2004; Bottorff et al., 2004; Williams, 2001; Williams, Skirton, Reed, Maas, & Daack-Hirsch, 2001). Unfortunately, for much of this time, nurses paid scant attention to this burgeoning area of science. Recently, however, nursing leaders have begun to promote the integration of genetics into research, education, and health care.

Incorporated in 1989, the International Society of Nurses in Genetics (ISONG) has provided key leadership in nursing and genetics through the development of their scope of practice standards, in conjunction with the American Nurses Association (ANA). In addition, its members have been strategic and have taken seriously their charge to educate other nurses and health professionals. For example, ISONG members participated in the publication of special issues in professional journals such as the *Journal of Nurse-Midwifery*. As an international organization, the members and leaders are from many countries.

The American Academy of Nursing's Genetic Health Care Expert Panel focuses on policy discussions and genetic issues, including providing public testimony to the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS). ISONG, the ANA, and other professional nursing organizations are members of the interdisciplinary National Coalition for Health Professional Education in Genetics (NCHPEG) with the goal to advance

the education of all health care and public health professionals in genetics. In 2003, the World Health Organization (WHO) recognized the contributions of nurses as it actively sought nursing leaders in genetics from around the world to participate in its Genomics Resource Centre (<http://www.who.int/genomics/en/>). This initiative provides Web-based access to leaders and resources in genetics.

Nurses should take the initiative to be informed of the activities across the governmental agencies in their countries for opportunities for involvement and integration of those activities in their research, education, and policy. Examples in the US are the CDC's Evaluation of Genomic Applications in Practice and Prevention (EGAPP) project, the Secretary's Advisory Committee on Genetics Health and Society, the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, and the Surgeon General's and DHHS Family History Awareness Initiative ([www.hhs.gov/familyhistory](http://www.hhs.gov/familyhistory); Guttmacher, Collins, & Carmona, 2004).

Forty nursing leaders from nine countries, who contributed to the International Council of Nurses (ICN) monograph *Nursing Leadership: 21st Century Genetics for Global Health* (Feetham & Williams, 2004) identified reasons the engagement of nurses is important for health and public policy in genomics. The central reason is that nurses bring a personal health perspective to policy discussions, that is, recognition of the responses of individuals to health and illness, and the interdependence of individuals, families, and communities with broader social, political, and physical environments. A second reason is that nurses understand the association between personal and public health perspectives. These and other reasons are discussed in the ICN monograph to provide direction to the discipline for leadership in genomics for global health. The monograph includes actions of nursing leaders nationally and internationally to advance the roles of nurses in genetics, the education of health care and public health professionals, and advocacy to so that the potential benefits of the genomic era are achieved in all countries (Ando, 2000; Feetham & Williams, 2004; Genetic Nursing Committee of Japan, 2000; Kirk, 1999; 2004; Skirton, Barnes, Curtis, & Walford-Moore, 1997).

Knowledge of all areas related to genomics will position nurses to be full partners with other health care and public health professionals and policymakers to advance global health through more informed health promotion and more targeted prevention and treatment of complex conditions. Nursing leaders can bring issues to the appropriate forums, use results of research in health and social policy debates, and participate in developing optimal practices and policies that are in the best interests of individual patients, families, and groups (Olsen et al., 2003; Wakefield, 2004).

### Conclusions

The genomic era provides significant responsibilities and opportunities for nurses to provide leadership for nursing,

health care, public health, and health policy. All health care and public health professionals will have to increase their ability to anticipate and plan for the opportunities and potential consequences of the exponential knowledge from genomics for changes in health care. Through the history and tenets of the discipline, nurses are well positioned to provide the interdisciplinary leadership that is required to reach the potential of the genomic era for improved health and health care.

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