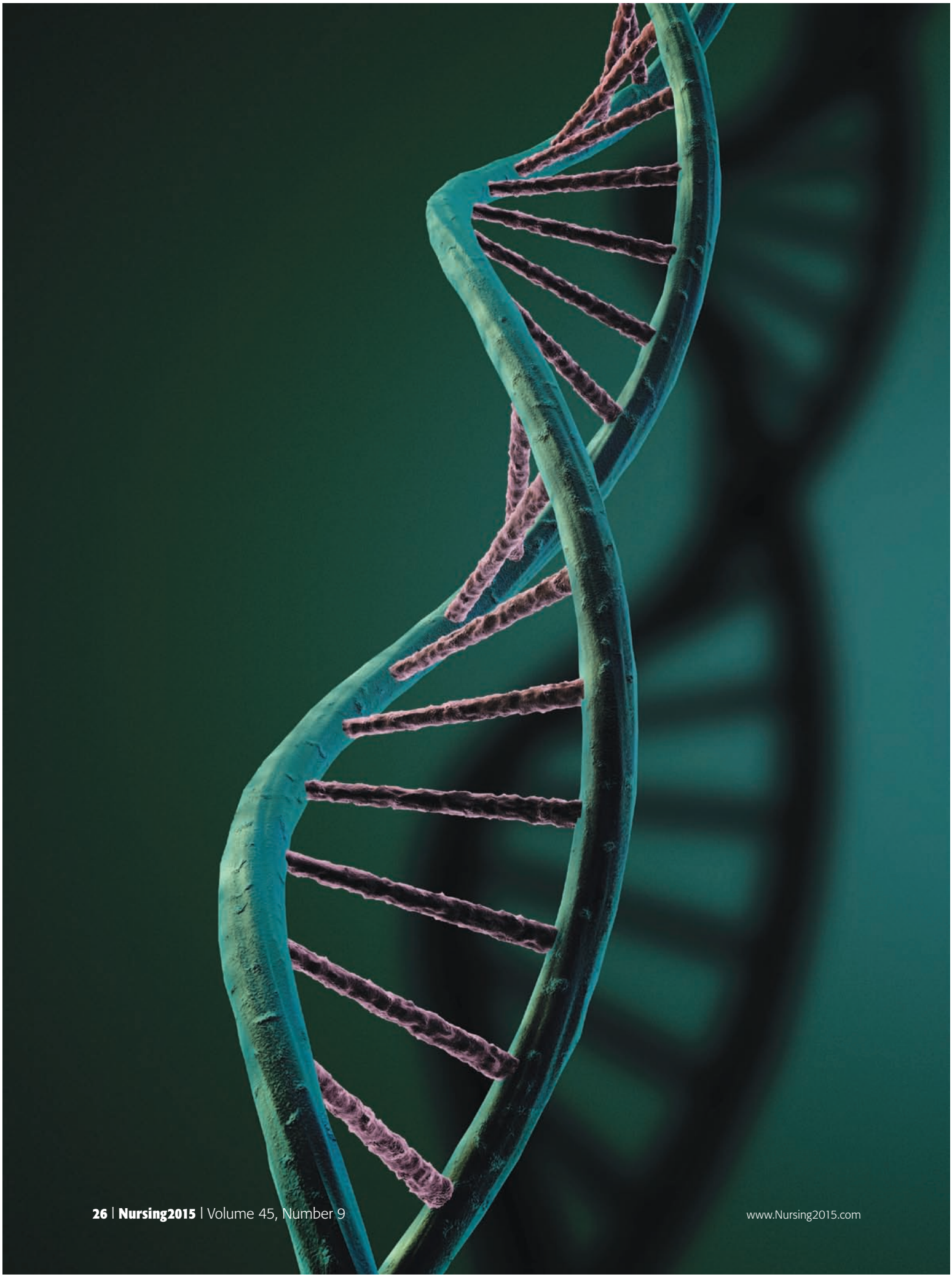

Mapping the human genome:

Implications for practice

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TWO PATIENTS on the medical-surgical unit present as follows. Mr. C, an Asian-American patient, was admitted for excessive bleeding after starting warfarin therapy initiated at the usual recommended dosage. Mrs. K, a postoperative patient of Greek descent, experienced respiratory depression after a single dose of codeine. As the nurse caring for these two patients, would you be prepared to explain the possible reason for their unusual reactions to these medications? If you were asked to construct a family history pedigree for a newly admitted patient, could you complete this request—and understand what it's for?

The Human Genome Project (HGP) holds the key to these questions. Completed in 2003, it's had a dramatic impact on healthcare and opens new possibilities in many areas of patient care. Regardless of their practice area, nurses are now responsible for developing the knowledge, skills, and attitudes required by the genomic era.[1,2] This article discusses the implications of the HGP and its relevance to everyday clinical situations.

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How to build a human

The HGP began in 1990 when the National Institutes of Health (NIH) and the Department of Energy collaborated with global partners to sequence the human genome. A genome is an organism's complete set of DNA, including its genes. The information an organism needs to maintain itself is contained in each genome. All human cells that have a nucleus contain a complete copy of the genome.[3]

The HGP enabled scientists to "read nature's complete genetic blueprint for building a human being" for the first time.[4] Under the leadership of Dr. Francis Collins, the publicly funded HGP laid the foundation for the evolution of genomic medicine.[5]

The traditional view that the value of genetics is limited to rare medical conditions is not only far outdated, but also dangerous.[6] Mapping the human genome created new opportunities to personalize the approach to genetic and genomic medicine for many patients. Advances in genomic technology have led to a reduction in cost for mapping an individual's genome; the projected reduced cost of $1,000, a target for the genomic sequencing industry, can result in new approaches to clinical management and research.[7,8]

Genetic or genomic?

Genetic research (the study of particular genes) and *genomic* research (the study of the entire genome) have different implications for healthcare. The initial discoveries from the HGP created new opportunities to treat monogenic genetic disorders—those involving a single gene. Monogenic disorders include cystic fibrosis and hereditary hemochromatosis.[6] Cystic fibrosis was once considered a terminal illness but new treatments are improving patient outcomes and quality of life.[9] Although single gene disorders are uncommon, they have a significant impact on an affected person's health.

Moving beyond single gene disorders, the HGP revealed connections between the entire genome and other nongenomic factors, such as environmental influences, that can affect the genome and health. *Epigenetics* refers to heritable changes that don't affect the DNA sequence but do influence gene expression. *Epigenomics* refers to the multitude of chemical compounds and proteins that attach to DNA and affect gene expression. Both are relatively new fields of study that are contributing to the understanding of gene-environmental interactions—environmental factors that influence gene activity without altering the DNA sequence.[2,10] (For more definitions related to genetic and genomic topics, see *Glossary of terms*.)

Environmental factors, including radiation and chemicals, can impact the genome and result in significant

Glossary of terms

Allele: one of two or more versions of a gene responsible for hereditary variation. Individuals inherit two alleles for each gene, one from each parent. If the alleles are different, the dominant allele will be expressed and the recessive allele's effect will be masked. In order to develop a recessive genetic disorder, an individual must inherit two copies of the mutated allele.

Epigenetics: the study of changes caused by the activation and deactivation of genes without any change in the organism's DNA sequence.

Epigenomics: the study of the epigenome, a multitude of chemical compounds and proteins that attach to DNA and direct various actions, such as controlling the production of proteins in particular cells.

Fluorescence in situ hybridization (FISH): a lab technique for detecting and locating a specific DNA sequence on a chromosome. The technique relies on exposing chromosomes to a small DNA sequence called a probe that has a fluorescent molecule attached to it. The probe sequence binds to its corresponding sequence on the chromosome.

Gene: a sequence of DNA occupying a specific location on a chromosome; considered the basic unit of heredity.

Genetics: the study of a particular gene.

Genomics: the study of an organism's entire genome.

Genotype: an individual's collection of genes. The term also can refer to the two alleles inherited for a particular gene.

Genome-wide association studies (GWAS): an approach used in genetics research to associate specific genetic variations with particular diseases. The method involves scanning the genomes from many different people and looking for genetic markers that can be used to predict the presence of a disease.

Genome: the entire set of genetic instructions found in a cell. In humans, the genome consists of 23 pairs of chromosomes, found in the nucleus, as well as a small chromosome in the cell's mitochondria. In their entirety, the 23 chromosomes contain approximately 3.1 billion bases of DNA sequence.

Monogenetic disorders or monogenic inheritance: disorders caused by a mutation in a single gene. The mutation may be present on one or both chromosomes (one chromosome inherited from each parent). Examples include sickle cell disease, cystic fibrosis, polycystic kidney disease, and Tay-Sachs disease.

Polymorphism: involves one of two or more variants of a particular DNA sequence.

Single nucleotide polymorphism (SNP): the most common type of polymorphism; involves variation at a single base pair.

health consequences, such as cancer and coronary artery disease (CAD).[11] The Genome-Wide Association Study (GWAS) is an approach used to connect specific genetic variations with particular diseases. The researcher scans the genomes of different individuals and looks for genetic markers that could predict the presence of disease. These markers could contribute to the understanding of genetic contributions to disease and result in better prevention and treatment approaches.[12,13] The GWAS is exploring the association between CAD and particular genes and investigating the genetic contribution to stroke.[14] The results of the HGP have improved understanding of these and other impacts and are leading the way to genomic medicine.[15,16]

Maintaining genomic competencies

In the new era of genomic healthcare, nurses have a professional responsibility to practice based on current knowledge, skills, and attitudes.[1,17,18] The American Nurses Association (ANA) and other professional organizations have published resources that define the expected competencies for nurses at all levels of education and practice. (See *Resources for keeping current*.)

Nurse leaders and nurse educators play key roles in guiding nurses to achieve the required competencies, yet reports reveal limited progress in this area.[17,19-21] Nurse leaders can support the efforts of practicing nurses to gain the knowledge, skills, and attitudes needed to integrate the genetic and genomic competencies into practice, but some barriers exist. (See *More education needed—at every level*.)

Family history and pedigree

A specific competency for all levels of nurses in professional practice is the ability to gather a complete family health history and construct a three-generation family pedigree using standardized symbols and terminology.[22,23] Gathering data for a family history is a valuable tool that can be integrated into clinical practice, but it's often underused.[19,24] The family history can provide a record of the illnesses and causes of death of biologically related family members, which might give clues as to the impact of genetic inheritance, environmental influences, and lifestyle factors on the health of family members.[21]

Key elements of a family history include

- the number of relatives affected by the patient's disorder
- the degree of relationship for affected relatives. Parents, siblings, and children are first-degree relatives. Second-degree relatives are half-siblings, aunts, uncles, grandparents, nieces, and nephews. Third-degree relatives are first cousins.[25]
- gender of affected relatives
- age at onset
- ancestry (ethnicity or region of origin)
- lineage (maternal or paternal relatives).[24]

A pedigree is a diagram of the family history for three generations representing grandparents, parents, siblings, half-siblings, children, aunts, uncles, and cousins.[26] The pedigree must incorporate standardized terminology and structure in order to ensure consistency and accuracy in communication among the members of the healthcare team.[27]

An online resource, *The Medical Family History: Tools for Your Practice,* is available from the National Coalition for Health Professional Education in Genetics (NCHPEG). This is an excellent tool for guiding the collection of a family history and constructing a pedigree. Case studies give nurses examples of interpreting different clinical case presentations and show how decisions are made regarding genetic disease and the risk of inheritance.[25]

Focusing on red flags

When taking a family history in the primary care setting, nurses can learn to ask focused questions and relate the patients' answers to the assessment data. One approach is to look for genetic red flags. A helpful mnemonic is *Family Genes*. *Family* refers to taking a family history and looking for multiple affected family members in multiple generations. *Genes* can be spelled out as a reminder to check for these points:

Resources for keeping current

The American Nurses Association (ANA) has defined competencies for all levels of nursing in the *Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators*, 2nd Edition.[22] The ANA and the International Society of Nurses in Genetics (ISONG) published the *Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees* to define a higher level of expertise for nurses prepared at the master's and doctoral levels.[23]

An interdisciplinary resource to promote collaboration among a diverse group of leaders is the National Coalition for Health Professional Education in Genetics (NCHPEG). The ANA joined with the American Medical Association and the National Human Genome Research Institute to establish NCHPEG with the goal of promoting education and access to information about advances in human genetics. Learn more at www.nchpeg.org.

The *National Human Genome Research Institute Talking Glossary of Genetic Terms* is a free resource, available for download to mobile devices or computer. Genetic and genomic terms are defined via text and audio versions. Learn more at www.genome.gov/glossary.

• G: groups of congenital anomalies that can relate to a genetic syndrome, such as three relatives from two generations with CAD.
• E: extreme or exceptional presentation of common conditions, such as early onset of disease; developing CAD at age 35, for example, should raise suspicions.
• N: neurodevelopmental delay or degeneration, such as early onset of neurologic deterioration in adults
• E: extreme or exceptional pathology, such as multiple primary cancers
• S: surprising lab results that don't seem to fit the clinical picture.[15,28]

Keeping these points in mind when assessing a patient, the nurse can identify genetic red flags, report observations to the patient's healthcare provider, and document assessment findings. The healthcare provider can then refer patients to a genetic counselor and a clinical geneticist, a provider certified in clinical genetics. A genetics counselor takes a nondirective approach to explain the facts and provide accurate information so the patient and family can make informed healthcare decisions.[29]

Implications for acute care

Documenting family history is attracting renewed interest in acute care settings as well as primary care settings. Nurses working in medical-surgical settings, ICUs, and EDs should consider genetic disorders that can result in dangerous complications for patients and families. For example, malignant hyperthermia (MH) is a potentially fatal, inherited disorder usually associated with the administration of certain general anesthetics and/or succinylcholine. MH can be detected with genetic testing. Assessing patients for a family history of MH before surgery is an important responsibility for nurses, especially in the perioperative setting.[30]

A comprehensive cardiovascular assessment can provide important information about the risk of a genetic disease. For example, early identification of hypertrophic cardiomyopathy can help prevent sudden cardiac death.[31] A history of dyspnea, fatigue, and systolic murmur should prompt additional testing. A history of sensorineural hearing loss could be related to the autosomal recessive form of long QT syndrome called Jervell and Lange-Nielsen syndrome, which can also cause sudden death.[32,33]

Marfan syndrome (MFS) is a single gene disorder of connective tissue that can cause significant morbidity and mortality. A patient with MFS is at increased risk for aortic root disease, leading to aneurysmal dilation, aortic regurgitation, and dissection. Assessment findings for a patient with MFS may include a tall and slender body, arachnodactyly, scoliosis or kyphosis, and pectus excavatum or pectus carinatum.[34,35]

Personalizing drug therapy

In the era of genomic and personalized medicine, nurses need to be aware of the impact that pharmacogenetics and pharmacogenomics has on patients in clinical settings.[36] *Pharmacogenetics* refers to altered drug responses that relate to single gene variations. Depending on the specific genetic variation, the patient may have a range of adverse drug reactions. *Pharmacogenomics* is a broader term that refers to differences in people's genomes that affect the

More education needed—at every level

Nurses have a professional responsibility to practice based on current knowledge, skills, and attitudes.[1,17,18] Nurse leaders and nurse educators play key roles in guiding nurses to achieve the required competencies in genomic healthcare, yet research reveals limited progress in this area.[17,19-21]

In 2010, a genomic survey was administered to nursing leaders attending the American Nurses Association (ANA) House of Delegates meeting to assess "attitudes, receptivity, confidence, practice and competency of nurses in genomics." The results revealed that nurse leaders perceived a benefit in terms of better recommendations for preventive services and better treatment decisions, but identified as disadvantages increased insurance discrimination and increased patient anxiety about risk.[17] These nurse leaders weren't current in their knowledge of the Genetic Information Nondiscrimination Act of 2008, which specifically addresses discrimination issues.[56] The nurse leaders identified the importance of a family history but couldn't correctly identify the core elements of a family history collection, even though family history and pedigree construction are competencies included in the ANA's *Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators*, 2nd ed.[22]

A survey of nurses in practice revealed that 71% valued genetics as important, but 81% had poor or fair understanding of the role of genetics in common diseases. They didn't routinely use family history in clinical practice, but were interested in learning more about genomic education.[19]

Seeking to improve genomic competency in practice was the focus of a yearlong research project conducted with nurse dyads of a nurse administrator and a nurse educator employed at Magnet hospitals.[54] Supported by the National Council of State Boards of Nursing and West Virginia University, the project aimed to improve integration of genomic competencies into 23 Magnet® hospital settings across the country. Nearly 8,000 nurses participated in the project. The results revealed that nurses had some knowledge and valued genomics, but the researchers identified a "genomic nursing competency deficit affecting all nurses regardless of academic preparation or role." They concluded that additional nursing education is needed to achieve the required competencies.[54]

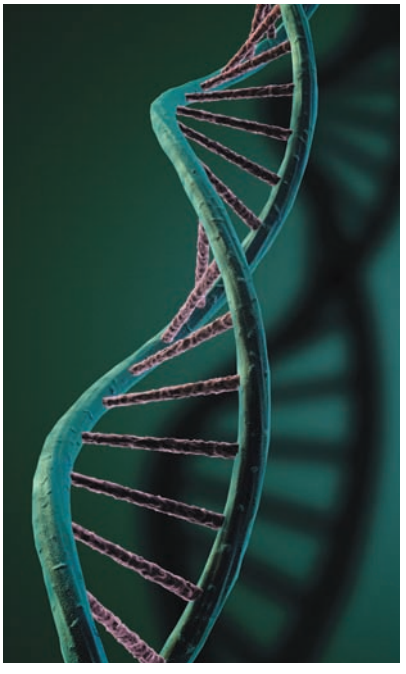
response to certain drugs. A slightly different DNA sequence in a patient's genome can affect the response to a medication in either a therapeutic or adverse manner.[37,38]

Nurses have many responsibilities in the administration of medications, including administering medications safely and monitoring patients for therapeutic effects and adverse reactions. The genomic era has given clinicians a better understanding of atypical responses to certain drugs that have been observed over the years.

For example, much attention has focused on the CYP family of enzymes and their effect on the metabolism of drugs.[39] Enzymes produced from cytochrome P450 genes affect drug metabolism. Each cytochrome P450 gene is labeled with CYP, identifying it as a part of the cytochrome P450 gene family. Variations (polymorphisms) change how these genes function, with the result that certain drugs are metabolized more quickly or more slowly than usual. This can significantly affect a patient's response to treatment. Nurses need to recognize that patients may metabolize drugs differently based on genetic variants in the enzyme system.[40]

We also have a better understanding of how ethnicity can influence a patient's response to medications. People of different ethnicities have genetic variations in metabolic enzyme activity that can significantly affect how they respond to certain drugs.[15] Clinical trials for cardiovascular medications, including antihypertensive drugs, traditionally included subjects who were mainly White, with few of Black, Asian, or Hispanic ethnicity. This sometimes resulted in dosing recommendations that weren't optimal for many nonwhite patients.[40]

As illustrated by the case studies in the beginning of this article, warfarin and codeine are two common drugs that can be affected by a patient's genetic variations. Due to variations in CYP enzyme function, ethnically Asian patients may need a lower dose of warfarin initially. These patients and others who carry the CYP2C9*2 allele and/or the CYP2C9*3 allele, and those who have the VKORC1 AA genotype, may have a higher risk for bleeding with the usual dose, so lower initial doses may be indicated.[41]



Patients may metabolize certain drugs differently based on genetic variants in their enzyme system.

Codeine is a prodrug that requires metabolism by the CYP2D6 enzyme to convert to morphine and produce an opioid effect. Patients who have increased activity by that enzyme due to their genotype may be ultra-rapid metabolizers and demonstrate an unusual response to the codeine. This can result in opioid intoxication at even modest doses.[42] Signs and symptoms include severe abdominal pain, sedation, and respiratory depression. People of Greek descent may be at an increased risk for this response; about 10% of persons from that ethnic group are ultra-rapid metabolizers.[15]

Addressing privacy issues

Nurses are remiss in meeting their responsibilities to patients and families if they're not informed and prepared to discuss the Genetic Information Nondiscrimination Act (GINA). Because the HGP provided the possibility for revealing much about an individual's risk for genetic disorders, the scientific and healthcare communities were concerned about the potential for discrimination by employers and health insurers. To address that possibility, President Bush signed GINA into law in 2008. The provisions of the law prevent employers or health insurers from using genetic information to make decisions about a person's work opportunities or premiums for health insurance. However, GINA doesn't restrict the ability of long-term care, disability, or life insurance companies to make decisions based on genetic risk. Other nonprotected groups are individuals serving in the military, military veterans cared for by the Veteran's Administration, and American Indians served by the Indian Health Service.[43]

New York Congresswoman Louise Slaughter reminded nurses about the importance of providing accurate information about the law and its protections to those who are concerned about discrimination based on genetic testing or family history of a genetic disease.[44] Inform them, for example, that GINA doesn't prohibit employers or insurers from considering information about a genetic disorder if the person already has evidence of a genetic disease.[45] Nurses can find a guide for discussing GINA with patients on the NCHPEG website (www.nchpeg.org/).

Genetic testing technologies

The HGP has resulted in development of new technology in the form of genetic testing, whole-genome sequencing, whole-exome sequencing, GWAS, epigenomics, gene expression profiling, and biorepositories.[2,43] Discovering differences in the genetic makeup of individuals is what will enable "personalized medicine" to become a reality instead of science fiction. With advances in technology, clinicians have more opportunity to test and treat patients with common diseases such as diabetes, cancer, and heart disease, which are affected by many genetic and environmental factors. Genetic testing takes many forms, and a complete discussion is beyond the scope of this article.

When receiving lab reports, nurses may begin to see acronyms used for genetic technology such as SNP (single nucleotide polymorphism) and FISH (fluorescence in situ hybridization).[46] SNPs refer to the most common type of human genetic variation. They can occur normally and have no effect on a person's health. SNPs have been used to help predict response to drugs and susceptibility to environmental influences and to track disease genes within families. Research continues to identify the relationship of SNPs to complex health disorders including heart disease, diabetes mellitus, and cancer.[47]

FISH provides visualization and mapping of genetic material in a human cell, including specific genes. It's helpful for understanding chromosomal abnormalities and genetic mutations.[48]

Four technological advances that are used in genomics are genome sequencing, GWAS, gene expression profiling, and epigenomics.[2] GWAS and epigenomics were addressed previously in this article; genome sequencing and gene expression profiling will be described in this section.

Introducing the Precision Medicine Initiative

In January 2015, President Obama announced the Precision Medicine Initiative, a research effort intended to "pioneer a new model of patient-powered research." Designed to help healthcare providers better understand and manage diseases and improve patient outcomes, it's described as "an innovative approach to disease prevention and treatment that takes into account individual differences in people's genes, environments, and lifestyles." Anticipated benefits include reducing adverse reactions and improving cancer survival. Using a Precision Medicine approach, healthcare providers consider pharmacogenomics in prescribing medication for a specific patient to avoid the "one size fits all" approach, which can result in adverse drug reactions and treatment failure.

Funding for the Precision Medicine Initiative is included in the President's 2016 budget, which provides a $215 million investment for the National Institutes of Health, together with the FDA and the Office of the National Coordinator for Health Information Technology, to support this effort.

Objectives of the Precision Medicine Initiative include:

- more and better treatments for cancer
- creation of a voluntary national research cohort
- commitment to protecting privacy
- regulatory modernization
- public-private partnerships involving, for example, academic medical centers, researchers, foundations, medical product innovators, and medical ethicists.

Source: The White House. Fact Sheet. President Obama's Precision Medicine Initiative. News release. January 30, 2015. www.whitehouse.gov/the-press-office/2015/01/30/fact-sheet-president-obama-s-precision-medicine-initiative.

Newer approaches to DNA sequencing have increased efficiency in production of data, reducing the time and cost of sequencing the human genome. The original approach, called whole genome sequencing, has been made less costly in terms of time and money with a method known as whole exome sequencing, which represents sequencing the 1% that is the protein-coding regions of the DNA and contains more than 85% of the genetic variations that cause or have strong impact on disease. Diagnosis and treatment can be based on these data.

One clinical practice implication for genome sequencing involves treatment for breast cancer. Genome sequencing can help clinicians predict how a patient with breast cancer, based on her genotype, will respond to different drugs that may be used to treat the tumor.[2]

Gene expression profiling can also promote a personalized approach to breast cancer treatment. The human genome contains over 20,000 genes; not all are active or expressed at any one time. Making a determination for breast cancer treatment can be guided by the tumor's gene expression pattern.[2] Microarray (or chip) technology can detect and analyze thousands of genes simultaneously to seek patterns of expression.[26] Using DNA microarray techniques, researchers are making some progress in being able to predict patient outcomes based on this technology. Much more development of the techniques is needed for gaining accuracy and definitive outcomes in clinical practice.

Funded by the NIH, the Clinical Genome Resource (ClinGen), has a mission to build an authoritative central resource that defines the clinical relevance of genomic variants for use in precision medicine and research. Through this central resource, the working group aspires to build a genomic knowledge base to standardize testing and improve patient care.[49-51]

ClinGen has a patient portal, GenomeConnect, whose purpose is "to partner researchers, clinicians, and patients with each other to learn about the effects of genetics on human health and disease. *Patients* will be the source of data studied, as they are the experts on their own health history and have (or can access) their genetic test results."[52]

Nursing roles in the genomic era

A new role for nurses in the era of genetics and genomics is the advanced genetics nurse. As of December 2014, the American Nurses Credentialing Center (ANCC) has the responsibility for credentialing genetic nurses.[53] Nurses who were credentialed under the Genetic Nurses Credentialing Commission (GNCC) and the International Society of Nurses in Genetics (ISONG) will remain credentialed through the expiration date on their credential.

The current credential, Advanced Genetic Nursing-Board Certified, is awarded upon completion of the peer-review and portfolio process as described on the ANCC website.[54] Nurses who were certified through the GNCC can find information about renewing their certifications at www.nursecredentialing.org/AdvancedGenetics.

The role of the generalist nurse in genomic care doesn't require additional education or credentialing. Clinical nurses who have ongoing interaction with patients in various settings must be prepared to observe for "red flags" that can help them identify patients or families at risk for a genetic disorder. In addition, because patients and families may have questions about genetics or concerns about their family, the nurse must be prepared to provide accurate information or refer the patient and family to the healthcare provider for a possible consultation with a genetic counselor. A genetic counselor has education and expertise to advise the patient and family regarding specific genetic disorders and possible genetic testing.[15]

Whether at the baccalaureate or graduate education level, nurses have a lot to learn in this era of genomic medicine. Many opportunities are available for independent learning through the ISONG website and also through formal coursework, such as those offered by the Cincinnati Children's Hospital Medical Center through their Genetics Education Program for Nurses. This Web-based Genetics Institute (WBGI) is an 18-week, online asynchronous learning experience with rigorous assignments and online discussions. The course provides many opportunities for nurses to interact with colleagues in various roles and to learn from expert educators and clinicians in genetics and genomics.

Another look at the case studies

Returning to the patients presented at the beginning of this article, could you now explain the genomic implications for their clinical situations? Because Mr. C is of Asian descent, he may have a genotype that predisposed him to excessive bleeding from warfarin. He was treated with a lower dose of warfarin and careful monitoring. Mrs. K, a patient of Greek descent, may have the genotype of an ultra-rapid metabolizer of the prodrug codeine to morphine. This rapid metabolism of codeine resulted in high blood levels of morphine leading to sedation and respiratory depression.

Your ability to incorporate information about genomic medicine into your nursing practice can yield clinical insights that improve care and outcomes for many patients. ■

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