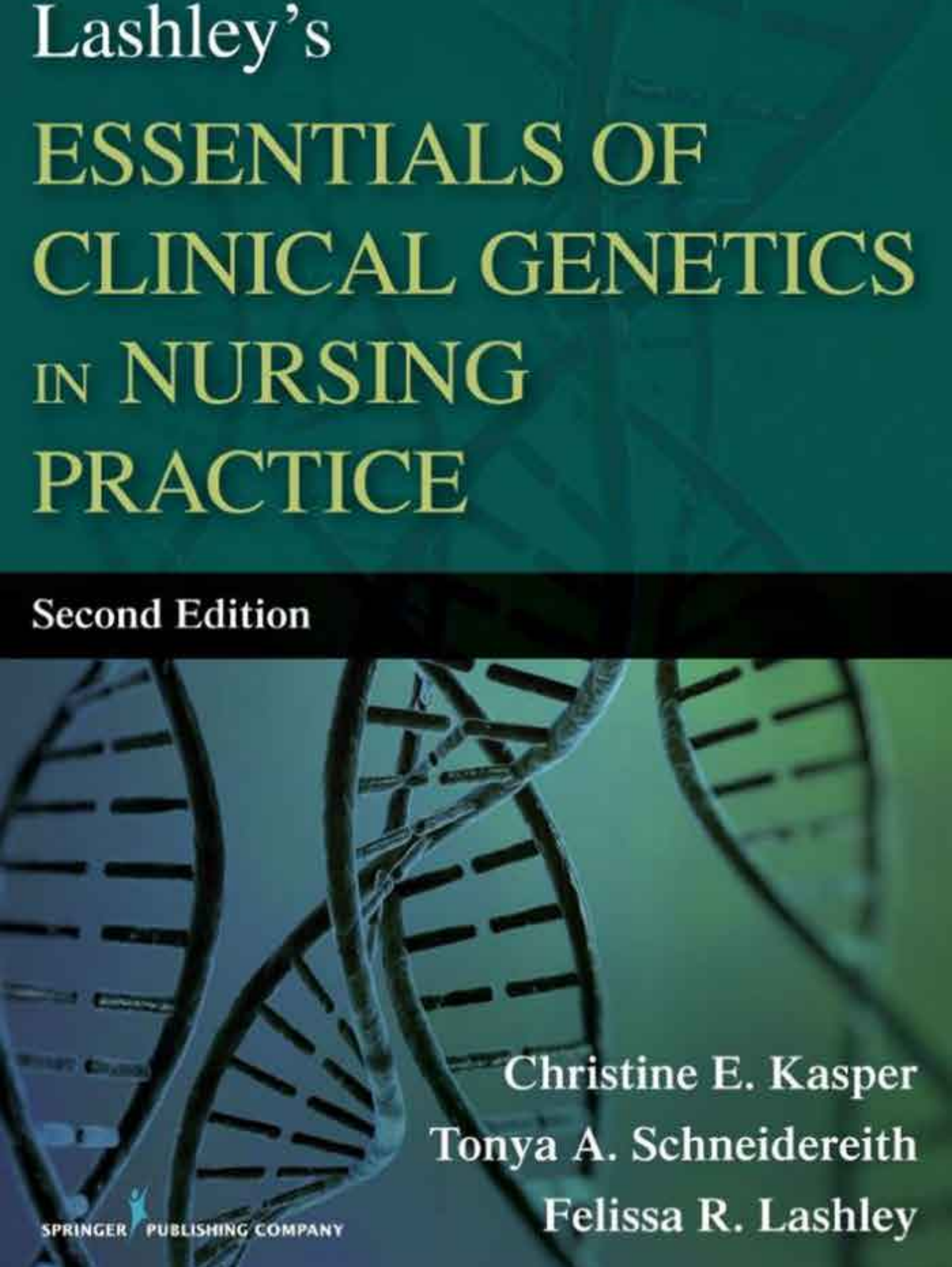
Lashley's

# ESSENTIALS OF CLINICAL GENETICS IN NURSING PRACTICE

Second Edition



Christine E. Kasper
Tonya A. Schneidereith
Felissa R. Lashley

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# LASHLEY'S ESSENTIALS *of* Clinical Genetics *in* Nursing Practice

**Christine E. Kasper, PhD, RN, FAAN, FACSM,** is a senior research scientist and professor at the U.S. Department of Veterans Affairs and Uniformed Services University of the Health Sciences. Prior to that, she held the Adelaide M. Nutting Endowed Research Chair at the Johns Hopkins School of Nursing and was also a faculty member at the University of California—Los Angeles and the University of Wisconsin—Madison. Dr. Kasper received her BSN from the University of Evansville, her MSN from Rush University, and her PhD from the University of Michigan.

Dr. Kasper has published more than 110 research papers, book chapters, reviews, and editorials in highly respected nursing and scientific journals. She was the founding editor of *Biological Research for Nursing* and is the current editor of the *Annual Review of Nursing Research*. She was a co-author of the ground-breaking book, *In Search of Nursing Science*, used in many nursing programs as a philosophy of science text. Her research has included funding from the National Institutes of Health (NIH), the National Aeronautics and Space Administration (NASA), and the Department of Veterans Affairs as the principal investigator on 10 grants. Additionally, she has received funding for 11 studies from foundations and universities, and has participated as a co-investigator on 14 additional interdisciplinary grants ranging from clinical genomics in nursing practice to genotoxic changes arising from embedded military-relevant heavy metals.

Dr. Kasper has been inducted as a fellow of the American Academy of Nursing and the American College of Sports Medicine. In 2015, she received the distinctive honor of becoming an inductee of the Sigma Theta Tau International Nurse Researcher Hall of Fame.

**Tonya A. Schneidereith, PhD, CRNP, PPCNP-BC, CPNP-AC,** is an assistant professor of nursing at Stevenson University. Dr. Schneidereith received her BSN from Florida State University, her MSN from the University of Pennsylvania, and her PhD from the Johns Hopkins University. While at Johns Hopkins, she studied under the direction of Dr. Christine Kasper, Dr. Gayle Page, Dr. Kirby Smith, and Dr. George Dover on the pharmacogenetics of fetal hemoglobin and F-cell variation. She was a summer intramural research assistant at the National Human Genome Research Institute under the direction of Dr. Wendy Fibison and has received funding as the primary investigator from the National Institute of Nursing Research for her work in molecular genetics.

Among her national and international presentations, Dr. Schneidereith has published in the *Annual Review of Nursing Research, Experimental Hematology,* and *Human Molecular Genetics*. She also serves as a reviewer for the *Journal of Nursing Education* and *Nursing Education Perspectives*.

Additionally, Dr. Schneidereith maintains a clinical practice as a pediatric nurse practitioner, with over two decades of experience in pediatric acute and primary care.

**Felissa R. Lashley, PhD, RN, FABMGG**, is former dean and professor at the College of Nursing at Rutgers, The State University of New Jersey. Prior to that, she was a dean and professor at the Southern Illinois University—Edwardsville and a clinical professor of pediatrics at the School of Medicine, Southern Illinois University—Springfield. She is the first nurse to be certified as a PhD medical geneticist by the American Board of Medical Genetics, and is a founding fellow of the American College of Medical Genetics. She began her practice of genetic evaluation and counseling in 1973.

# LASHLEY'S ESSENTIALS *of* Clinical Genetics *in* Nursing Practice

SECOND EDITION

Christine E. Kasper, PhD, RN, FAAN, FACSM
Tonya A. Schneidereith, PhD, CRNP, PPCNP-BC, CPNP-AC
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*To my parents, David and Betty Andrade, who developed and fostered my passion for learning. To my wonderful husband, Scott, and my children, Sam and Lauren, without whom this would not be possible.*

*—TAS*

*To my earliest mentor in science, my father, John M. Kasper, and to my first professional mentor, Luther Christman, PhD, RN, FAAN, who made it all possible. And to my family: Ray, and my talented daughters, Alexandra and Gabrielle, for their constant encouragement and support.*

*—CEK*

*To my very special children (Peter, Heather, and Neal) and grandchildren (Ben, Hannah, Jacob, Grace, and Lydia). You make everything brighter.*

*—FRL*

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# EDITOR'S NOTE

It is hard to believe that my interest in genetics began more than 50 years ago, when I chose genetics, as opposed to a less demanding course, as a needed free elective at Adelphi University. At the same time, one of my nursing instructors noted that I was "too interested in the unusual." In the 1960s, it was at New York University, influenced by Dr. Martha Rogers and Inga Thornblad, where I really discovered that I was a critical thinker and that even as a woman and a mother, the sky was the limit. Yet, it was not until I began my doctoral work in the Department of Biological Sciences at Illinois State University, when I had to repeat a genetics course because of all the changes that had occurred since my undergraduate course, that I saw the potential that genetics held for people's health and how necessary that knowledge would be for the health professions.

So I set my sights in that direction, switching my major to genetics, specifically human and medical genetics, with a minor in biochemistry. That was in 1970. I especially am grateful for the shared knowledge and professionalism of two of the genetics faculty there, Dr. Herman Brockman and Dr. William (Bill) Daniel, who were wonderful role models of scholarship and decency.

And if the potential was visible then, surely all of the applications available now were only a dream. Over my career, however, understanding human genetic variation interacting with the environment, along with its implications, has led to exciting applications not only in health and illness, but also in fields such as forensics and law. Genetics and genomics have truly permeated all aspects of our lives, and even young schoolchildren are conversant in the terminology and concepts, if not the societal implications.

It hardly needs to be said that the increasing importance of genetics and genomics translates to all fields of nursing, as well. By now, I hope that nurses are truly "thinking genetically" and looking at their clients with a "genetic eye." To do otherwise would be a failure to practice nursing in the way that it should be practiced by the professional nurse.

I am now happily retired from active practice and am lucky enough to be able to spend my time doing the things I love most, especially spending time with family and friends. Throughout my past genetic evaluation and counseling practice, I met so many wonderful people affected by genetic variation. I am grateful for the lessons I learned from them.

I have had wonderful friends and colleagues in and out of nursing, and there is not space to mention all of them; however, two long-time nursing friends and colleagues deserve special mention: Dr. Jerry D. Durham and Dr. Wendy M. Nehring.

I marvel at how thoroughly genetics is now integrated into our culture and society. Being in the genetics field has always been an honor for me and my contributions have been a labor of love.

And how many more amazing things in genetics there are to come … the excitement has just begun.

*Felissa R. Lashley*
Overland Park, Kansas

# PREFACE

The practice of clinical genetics and genomics has infiltrated nearly every area of health care. Currently there are over 3,000 genetic and genomic tests available to health care providers to query a wide range of diagnostic and pharmacogenetic needs, such as individual patient heredity and metabolic responses to drug treatment. Today's nurses not only participate in pedigree construction and risk identification, but are increasingly responsible for referral to genomic medical services. The formal academic process of bringing genetics into nursing began in 2000 and has since resulted in the 2009 publication of the American Nurses Association (ANA) Consensus Panel on Genetic/Genomic Nursing Competencies. These establish genomics as a core competency for all registered nurses (RNs), regardless of academic preparation, clinical role, or practice specialty. The endorsement of these guidelines by most professional nursing organizations leads to the hope that soon the study of genetics in the undergraduate curriculum will be as ubiquitous and required as anatomy and physiology are today.

Being able to assess clients and families with a "genetic eye" has become critical for all nurses. Advances from genetic and genomic research have influenced all areas of health care and all periods of the life cycle. Genetic factors are responsible in some way for both indirect and direct disease causation; for variation that determines predisposition, susceptibility, and resistance to disease; and for response to treatment. When we look into the future, we can see that the application of genetic knowledge, including genetic screening and personalized drug therapy, will have a direct influence on health care.

Nurses must be able to "think genetically" to help individuals and families, in all practice areas, that are affected in some way by genetic disease or are contemplating genetic testing. Each person's state of health and risk for developing diseases may be based on genetic variation. This includes not only diseases thought of as genetic but also more common disorders such as cancer and heart disease.

Becoming competent in the use of genetic content begins in undergraduate and generic nursing education programs. It was with this in mind that ***Lashley's Essentials of Clinical Genetics in Nursing Practice*** was originally written. Given the rapid progress of genetic and genomic science, the original work has been revised and extensively updated as ***Lashley's Essentials of Clinical Genetics in Nursing Practice, Second Edition.*** Part I of the book discusses the place of genetics in health care and the health care trends related to genetics. This is followed by a review of basic and molecular biology, a discussion of human variation and diversity, and gene action and types of inheritance. The topics of prevention of genetic disease, genetic testing, and treatment are presented, including aspects of genetic counseling. Part II applies these principles to areas of clinical nursing practice. Specific application of genetics and genomics in regard to pharmacology, history taking and physical assessment, maternal–child nursing, adult health and illness and medical–surgical nursing, psychiatric mental health nursing, policies, and social and ethical issues are

all discussed. The broad concepts are presented in a nursing context with selected disease examples and case examples. Many key concepts, questions, and examples from Dr. Lashley's practice appear liberally throughout this new edition. **Qualified instructors may obtain access to ancillary materials, including PowerPoints and a test bank, by contacting *textbook@springerpub.com***.

Within this book, the term ***normal*** is used as it is by most geneticists—to mean free from the disorder or condition in question. Genetic terminology does not generally use apostrophes (e.g., Down syndrome instead of Down's syndrome), and this pattern has been followed.

The writing of this book in a manner that allows students to understand and apply genetics is an important step toward early educational preparation. Thinking inclusively about genetics in all types of disease conditions will help nurses preserve the optimum function and health of patients. All nurses, as health care providers and as citizens, are charged with understanding advances in genetics and the resultant implications on health care and social decisions. In the words of Florence Nightingale (1859), "[T]he knowledge of nursing…of how to put the constitution in such a state as that it will have no disease, or that it can recover from disease, takes a higher place. It is recognized as the knowledge which everyone ought to have." For today's nurses, this is genetics.

*Christine E. Kasper*
*Tonya A. Schneidereith*
*Felissa R. Lashley*

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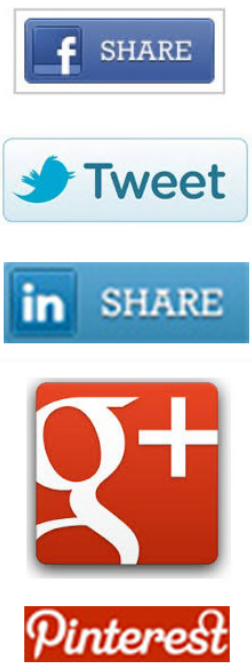

# PART I

## The Basics

# CHAPTER 1

# Genomics in Health Care

*Tonya A. Schneidereith and Christine E. Kasper*

Since the inception and completion of the Human Genome Project (HGP), the field of genetics has experienced unimaginable growth. The identification of approximately 30,000 human genes, coupled with advancements in molecular techniques, has created an opportunity to delve deep into every part of the human life span. No longer confined to the sciences and health care, discussions on genetics and the role of genes in disease are part of everyday conversation. From television and mainstream media to the grocery store and genetically modified foods, society is deluged with genetic information. The chromosomal locations for known diseases can now be found with the click of a mouse, making information accessible for everyone.

## HUMAN GENOME PROJECT

Much of the detailed information now known about human genetics evolved from the HGP. Started in 1990, the HGP was a collaborative research program coordinated through the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH) and the Department of Energy (DOE). David Smith directed the program at the DOE, and James Watson and Francis Collins were the first and second directors at NIH, respectively. Although the primary focus of research included gene sequencing and mapping of the human genome, a major contribution of the HGP was the development of large-scale molecular technologies. These contributions, along with the development of computer technologies to handle the enormous amount of sequencing data, have allowed for the continued, rapid advancements in all areas of genetic research.

In April 2003, the full sequence of the human genome was published in *Nature*. The complexity of the genome highlighted the discovery that only 1% to 2% of bases encode proteins, meaning that the role of 98% of human DNA is unknown. The total number of identified genes that code for proteins is approximately 30,000, fewer than what was originally expected. Some of the other unexpected findings included "the more complex architecture of human proteins compared to their homologs in worms and flies, the profoundly important lessons that could be learned from the human repeat sequences, and the discovery of apparent horizontal transfer from bacterial species" (Collins, 2001, p. 643).

The HGP also led to the establishment of the ethical, legal, and social implications (ELSI) programs of genetic research. The ELSI programs fund research in four main categories: genomics research; genomic health care; broader societal issues; and legal, regulatory, and public policy. To date, the major impact from ELSI research includes policies related to the conduct of genomics research, mostly involving informed consent. The future role of the ELSI program includes frequent reassessment of research priorities due to this constantly emerging science and protection of researcher autonomy and independence in a field filled with policy implications.

## INCREASING GENETIC LITERACY

Educators have recognized the importance of an informed public that is able to understand genetic risk and predisposition. Historically, aspects of genetics were taught in middle/high school and primarily included the basics of Mendelian inheritance. None of the complexities involved in disease were taught, leading students to believe that genetics followed only the primary inheritance patterns. The American Society for Human Genetics recognized these limitations and suggested a curriculum for K-12 education, increasingly focused on improving genetic literacy.

In today's health care, there is an expectation that providers are capable of understanding and translating findings from genetic screening and testing into language that is easily understood. This requires incorporation and comprehension of genetic content in both undergraduate and graduate education that is commensurate with the rapidly expanding gains toward understanding genetic risk and predisposition.

### Knowledge and Competencies

Many of the challenges and applications of new genetic information are still unknown, but health professionals in all areas of practice will encounter clients with disorders that have either a known genetic etiology or genetic component. Preparation of the provider will aid in recognition of the role of genomics in many conditions and the application of gene-based diagnostic tests and therapies. This includes a breadth of genetic and genomic knowledge regarding testing and assessment of risk, as well as the ability to interpret results and provide education and counseling.

However, staying current with genetic and genomic knowledge is, in itself, a seemingly insurmountable challenge for educators. A study of over 7,700 practicing nurses revealed knowledge deficits in genetics and genomics, while more than 50% of the group identified genetics in their curriculum (Calzone, Jenkins, Culp, Caskey, & Badzek, 2014). This suggests an inadequacy in genetic curricula and inappropriate academic preparation for both students and educators. Making academic preparation a priority is essential for future nurses.

The NHGRI and the National Cancer Institute (NCI) collaborated on a series of articles to help nurse educators focus on genetics and genomics (Mjoseth, 2012). Additionally, in 2006, an esteemed consensus panel comprising nurses from national organizations (NHGRI, American Nurses Association [ANA], Centers for Disease Control and Prevention [CDC], Health Resources and Services Administration

[HRSA], American Nurses Credentialing Center [ANCC], Sigma Theta Tau International, etc.), universities, and nurses' associations (Society of Pediatric Nurses, National Association of Hispanic Nurses, National Alaska Native American Indian Nurses Association, etc.) established essential competencies and curriculum guidelines. These guidelines were updated to include outcome indicators in the second edition, published in 2009 as the ***Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators*** (Jenkins, 2008). This document identifies essential competencies including:

- Professional responsibilities
  - Demonstrating understanding of genetics as applied to health prevention and screening
  - Ability to obtain three-generation family health history and construct a pedigree
  - Critically analyzing history for risk factors
- Applying/integrating genetic and genomic knowledge
- Identification of those who may benefit from genetic services
- Referral activities
- Provision of education, care, and support

Although the importance of these competencies is irrefutable, their implementation in nursing education is still inadequate. The ***Essentials***, along with integration of genetics in core science courses, provide the very basic components to best prepare future nurses to provide safe, cost-effective care that will improve health outcomes.

## NURSING ROLES IN A GENOMIC ERA

Traditionally, nurses were expected to interview clients, obtain an accurate history over three generations, and identify risk based on pedigree. However, the information gained from the HGP has added layers of complexity, including the idea of relatedness. As previously determined through a three-generation pedigree, inheritance and risk were measured through identity by descent (IBD). However, IBD does not account for molecular variability, including meiotic recombination, thereby making it an imprecise way to establish inheritance risk. The availability of molecular testing and analysis of genome-wide single-nucleotide polymorphism (SNP) data allows for more accurate diagnosis, limiting the value of the traditional pedigree. Will nurses forego the pedigree for whole-genome analysis (WGA)? Does this mean that teaching the art of eliciting a pedigree has become obsolete? Regardless, nurses should be prepared to explain and interpret correctly the purpose, implications, and results of genetic tests.

The role of the nurse will vary depending on the disorder, the needs of the client and family, and the nurse's expertise, role, education, and job description. Nurses will treat adults with genetic diseases of childhood who present with common

health problems and people with traditional adult-onset disorders, such as hemochromatosis and Huntington disease. Technological advances have increased life expectancy for many chronic diseases, including sickle cell disease and cystic fibrosis. This will lead to greater knowledge of the effects of illness across the life span. Different mutational changes within a gene may produce different phenotypic outcomes with varying responses to treatment and prognosis. Persons with specific genotypic mutations already are known to have preferential responses to certain medications or therapeutic approaches. Large-scale genome-wide association studies (GWAS) are shedding insight into the role of SNPs in complex diseases such as cancer, chronic obstructive pulmonary disease, diabetes mellitus, and heart disease. Additionally, access to whole-genome sequencing may be available within the next decade, making the idea of personalized medicine in diagnosis and treatment of disease a real possibility.

Consideration of the family unit is important for nurses. Identification of a genetic disorder in one member can allow others in the family to receive appropriate preventive measures, detection, and diagnosis or treatment and to choose reproductive and life options concordant with their personal beliefs. Also, there is a toll on the community and society. Although mortality from infectious disease and malnutrition has declined in the United States, the proportion due to disorders with a genetic component has increased, assuming a greater relative importance. Furthermore, nurses must be aware of potential increases in health disparities, especially among the poor and disadvantaged from various ethnic backgrounds, as the demand for genetic services continues to grow.

Nurses are in an ideal position to apply principles of health promotion, maintenance, and disease prevention. Coupling an understanding of cultural differences, technical skills, family dynamics, growth and development, and other professional skills with the person and family unit threatened by a genetic disorder, nurses can ensure an appropriate outcome.

For those interested in learning more about genetics, the International Society of Nurses in Genetics (ISONG; www.isong.org) offers various certifications for nurses related to genetics, depending on their education and experience. Additional certifications are available through the American Board of Medical Genetics.

## SUMMARY

Nurses are uniquely positioned to assess, treat, and educate individuals and their families on the presence, absence, or future possibility of disease. As members of the profession, it is the responsibility of the nurse to remain up to date on testing and therapies related to genetics and disease.

To paraphrase Francis Collins, the payoff of the HGP for health care professionals is a better ability to diagnose, treat, and prevent disease. Understanding the role of genetics and genomics throughout the life span, increasing genetic literacy, and applying new technologies in diagnosis and treatment is a great place to start.

## KEY POINTS

- Health care and society are increasingly influenced by genetics and genomics.
- Many genetic disorders that appear to follow Mendelian patterns of inheritance and were ascribed to a single mutant gene are now known to be more complex than formerly thought.
- The influence of genetic testing for screening and diagnosis has a greater weight now than once prior to the HGP.
- Genetic disorders may appear in any phase of the life span.
- Nurses will encounter clients/patients with genetically influenced disorders in every area of clinical nursing practice.
- Nurses play many roles in caring for persons and families affected by genetically influenced disorders.
- Nurses, as well as educators, should have basic genetic and genomic knowledge, competencies, and literacy.
- Personalized medicine may be a reality within the next decade.

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# CHAPTER 2

# Basic Concepts in Molecular Biology

*Wendy L. Kimber*

This chapter introduces the fundamental concepts in molecular biology that underlie the principles of genetics and inheritance. It begins with a discussion of the way that genetic material is organized into genes and chromosomes and the mechanisms by which these are transmitted to the next generation. This is followed by an explanation of the molecular nature of genes and the processes of DNA replication and gene expression. The flow of information from DNA to RNA to protein is described along with the consequences of mutations on this system. Finally, a brief survey of current genetic technologies is presented, including the burgeoning field of genomics. This chapter provides the foundational concepts on which subsequent chapters are built.

## CHROMOSOMES

The term ***chromosome*** is derived from the Greek words for color (***chroma***) and body (***soma***) as chromosomes were first observed as colored threads inside the nucleus of stained cells by scientists in the 1800s. These thread-like structures are present in the nucleus of all cells and are the basic units of heredity that are passed from parents to their offspring.

Chromosomes are composed of a single molecule (double strand) of DNA, which is wrapped around histone proteins (Figure 2.1). The association of DNA with histone proteins is known as ***chromatin***. Chromosomes exist in the cell in one of two forms, condensed (closed) or relaxed (open). For most of the time, the DNA in chromosomes is only loosely wound around histone proteins so that the genes on the chromosomes are accessible to the transcriptional machinery of the cell. In this form, chromosomes exist as long slender threads that are not visible under a light microscope. Only when a cell is getting ready to divide does the DNA become compacted to take on the characteristic shape and form of a chromosome.

Before a cell divides it makes a duplicate copy of each chromosome; both chromosome copies remain temporarily stuck together, with each individual chromosome referred to as a *chromatid* (Figure 2.2). During cell division the DNA of both chromatids are wound tightly around histone proteins so that it forms a short tight bundle.