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Basic Genetics and Genomics: A Primer for Nurses

[Dale Halsey Lea, MPH, RN, CGC, FAAN](#)

Abstract

The completion of the Human Genome Project is leading to a new type of medicine, called personalized medicine. The goal of personalized medicine is to create a healthcare plan that encompasses not only traditional factors, such as patient preferences and cholesterol levels, but also a person's genetic and genomic information. This approach should allow providers to prescribe more specific and individualized treatment and to avoid adverse drug and treatment reactions. A basic knowledge of genetic terms and concepts and an understanding of genomics can provide nurses with a foundation that will enable them to provide competent, personalized healthcare. This article will review the basic genetic structures and functions, explain cellular and genetic changes, discuss genetic inheritance, describe chromosomal variations, and share applications of genetics and genomics for nursing practice.

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The Human Genome Project (HGP) was an international research effort to read and map all of the genes in the human body, which together are known as the human genome. The HGP, completed in April of 2003, gave scientists the ability, for the first time, to read the complete genetic blueprint for building a human being ([National Human Genome Research Institute \[NHGRI\], 2008b](#)). As a result of human genome discoveries, it is now known that genetic factors play a role in nine of the ten leading causes of death in the United States, including heart disease, cancer, and diabetes. Human genome research is also leading to a better understanding of the interactions between genes and the environment and helping to find better ways to improve health and prevent disease ([NHGRI, 2008c](#)).

The completion of the Human Genome Project...is leading to a new type of medicine, called personalized medicine.

The completion of the HGP, by opening new doors for understanding the underlying causes of rare and common diseases, is leading to a new type of medicine, called personalized medicine. Personalized medicine is transforming healthcare. Learning about the influence of genetic and genomic factors on health and disease is leading towards earlier diagnosis, more effective and individualized prevention and treatment of disease, better response to treatments, and improved health outcomes ([NHGRI, 2008i](#)).

Nurses will increasingly be called upon to use genetic- and genomic-based approaches and technologies in client care.

For many, the term genetics brings up memories of pea plants and rare disorders caused by single genes. A newer term, genomics, refers to all of the genes in the human genome and their interactions with each other, the environment, and other cultural and psychosocial factors ([Consensus Panel, 2006](#)). The goal of personalized medicine is to create a healthcare plan that encompasses not only traditional factors, for example patient preference and attributes such as the Body Mass Index and cholesterol levels, but also a person's genetic and genomic information. This approach should allow providers to prescribe more specific and individualized treatment and to avoid adverse drug and treatment reactions.

Nurses will increasingly be called upon to use genetic- and genomic-based approaches and technologies in client care. As stated in the *Essential Nursing Competencies and Curricula*

Guidelines for Genetics and Genomics:

Genomics is a central science for all nursing practice because essentially all diseases and conditions have a genetic or genomic component. Health care for all persons will increasingly include genetic and genomic information along the pathways of prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness ([Consensus Panel, 2006, p. 1](#))

Having a basic knowledge of genetic and genomic terms and concepts is a crucial first step for nurses in building a foundation to provide competent and personalized healthcare. This article will review the basic genetic structures and functions, explain cellular and genetic changes, discuss genetic

inheritance, describe chromosomal variations, and share applications of genetics and genomics for nursing practice.

Basic Genetic Structures and Functions

This section will describe the structure and function of genes and deoxyribonucleic acid (DNA), the chemical found inside the nucleus of the cell that carries the genetic instruction for making living organisms and that is delivered by RNA from the cell's nucleus to the cell's cytoplasm where proteins are made. Chromosomes, which are the structures that contain genes, will also be discussed. [Table 1](#) provides a listing of common genetic and genomic terms and concepts related to this discussion.

Table 1. – Glossary of Genetic and Genomic Terms
<ul style="list-style-type: none"> ● Deoxyribonucleic acid (DNA) – The chemical inside the nucleus of a cell that carries genetic instructions for making living organisms. ● Double Helix - The structural arrangement of DNA, which looks something like an immensely long ladder twisted into a helix or coil. The sides of the "ladder" are formed by a backbone of sugar and phosphate molecules, and the "rungs" consist of nucleotide bases joined weakly in the middle by hydrogen bonds. ● Family History – A family's health history that helps one identify genetic heritage and risks, and guides in making healthy environment and lifestyle choices. ● Gene – The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein. ● Genetics - Genetics is a term that refers to the study of genes and their role in inheritance - the way certain traits or conditions are passed down from one generation to another. ● Genomics - Genomics is a relatively new term that describes the study of all of a person's genes including interactions of those genes with each other and the person's environment. ● Genetic disorders - A genetic disorder is a disease caused in whole or in part by a "variation" (a different form) or "mutation" (alteration) of a gene. ● Genetic testing - Genetic tests look for alterations in a person's genes or changes in the level of key proteins coded for by specific genes. Abnormal results on these tests could mean that someone has an inherited disorder. Types of genetic tests include: gene tests, chromosomal tests, and biochemical tests. <ul style="list-style-type: none"> ○ Gene tests - Gene tests look for signs of a disease or disorder in DNA taken from a person's blood, body fluids, or tissues. ○ Chromosomal tests - Chromosomal tests look at features of a person's chromosomes, which are structures in the nucleus of a cell that contain DNA, including their structure, number, and arrangement. ○ Biochemical tests - Biochemical tests look at the level of key proteins. This level signals genes that are not working normally. These types of tests are used for newborn screening. ○ Direct-to-consumer (DTC) genetic tests - A variety of genetic tests that are being offered directly to consumers, often over the Internet. Such genetic testing usually involves scraping a few cells from inside the cheek and mailing the sample to a test laboratory, which performs the test. The major types of DTC tests are health-related, ancestry, nutrigenetic, and general (test scans of a person's genome for genetic variants related to many aspects of life, from physical traits to ancestry and to personality). ● Mendelian Inheritance – Manner in which genes and traits are passed from parents to children. Examples of Mendelian inheritance include autosomal dominant, autosomal recessive, and sex-linked genes. ● Mitochondrial DNA – The genetic material of the mitochondria, the organelles that generate energy for the cell. ● Pharmacogenetics - Pharmacogenetics is the field of study dealing with the variability of responses to medications due to genetic variation. Pharmacogenetics takes into account a person's genetic information regarding how drugs are transported and metabolized by the body and their specific drug receptors. The goal of pharmacogenetics is to create an individualized drug therapy that allows for the best choice and dose of drugs. ● Pharmacogenomics - Pharmacogenomics is the research area that involves the search for genetic variations that are associated with drug efficiency. The term comes from the words pharmacology and genomics, and is the intersection between pharmaceuticals and genomics. Pharmacogenomics is leading to drugs that can be tailor

made for individuals, and adapted to each person's own genetic makeup.

- **Protein** - A large complex molecule made up of one or more chains of amino acids. Proteins perform a wide variety of activities in the cell.
- **Ribonucleic acid (RNA)** - A chemical similar to a single strand of DNA. In RNA, the letter U, which stands for uracil, is substituted for T in the genetic code. RNA delivers DNA's genetic message to the cytoplasm of a cell where proteins are made.

(NHGRI, 2008e; 2008m)

Genes, DNA, and RNA

A gene is the functional and physical unit of heredity passed from parents to their children. Genes make up the DNA; each gene is one length of DNA. Most genes have instructions for making a specific protein. Genes are responsible for the types of proteins made and, in part, the rate and time points at which they are made. Genes have regions called exons and introns. Exons are the regions of a gene that hold the code for making the gene's protein. Each exon codes for a specific piece of the complete protein. Introns are the noncoding sequences of DNA that are not involved in making the gene's protein. [Figure 1](#) [pdf] illustrates the basic structure of a gene. (NOTE: pdf figures must be printed individually).

Genes make up the DNA...The structure of DNA is called the double helix, and is shaped like a winding staircase.

DNA is the chemical inside the nucleus of a cell that carries the genetic instructions for making a living organism. The structure of DNA is called the double helix, and is shaped like a winding staircase. The DNA molecule is made up of sugar-phosphate molecules and pairs of nitrogenous bases. [Figure 2](#) [pdf] illustrates the DNA structure.

Ribonucleic acid (RNA) is a chemical similar to a single strand of DNA. RNA's role is to deliver DNA's genetic message to the cytoplasm of a cell where proteins are made. The proteins made are large complex molecules consisting of one or more chains of amino acids. The amino acids, called the "building blocks" of proteins, are a group of twenty different types of small molecules. The proteins that are made perform a wide variety of activities in the cell. [Figure 3](#) [pdf] describes the structure of amino acids.

There is another special type of DNA called mitochondrial DNA, or mtDNA. Mitochondrial DNA is genetic material found in mitochondria, the structures within cells that change energy from food into a form that cells are able to use. Each body cell has hundreds to thousands of mitochondria that are present in the fluid that surrounds the cell nucleus (cytoplasm). Mitochondria control energy production, help regulate cell death (called apoptosis), and help with the production of cholesterol and heme (a part of hemoglobin, the molecule that carries oxygen in the blood). Some mitochondria are responsible for making molecules called transfer RNA and ribosomal RNA, the chemical cousins of DNA. These two types of RNA help to put together the amino acids necessary to create working proteins ([Genetics Home Reference \[GHR\], 2008h](#)).

Chromosomes

Genes are arranged and lined up in threadlike structures called chromosomes.

Genes are arranged and lined up in threadlike structures called chromosomes. The number of chromosomes depends on the type of organism. [Figure 4](#) [pdf] portrays a chromosome consisting of genes that make up the DNA, which is made from sugar-phosphate molecules and pairs of nitrogenous bases. Humans have 46 chromosomes arranged in 23 pairs. Forty-four of these chromosomes are called autosomes. The remaining two are called the sex chromosomes. Females have two X chromosomes, and males have one X and one Y chromosome. Children get half of their chromosomes from their mother and half from their father. A person's chromosomal makeup is called a karyotype, a word that is also used for a picture of a person's chromosomes ([GHR, 2008g; NHGRI, 2008g](#)).

Cellular and Genetic Changes

This section will explain how cells normally divide. It will also describe how an unexpected change in the structure of DNA can sometimes cause harm to the body. New tools to study genetic variations of common diseases and to identify genetic variations common to specific diseases will also be presented.

Cell Division

Humans grow and develop as a result of a process called cell division. There are two types of cell division – mitosis and meiosis. Mitosis is the process of cell division that involves cell growth, differentiation, and repair. During mitosis, the chromosomes in each cell are duplicated resulting in two cells called daughter cells. Each of the daughter cells contains the same number of chromosomes as the parent cell, namely 46 chromosomes. The daughter cells are called diploid because they have 46 chromosomes in 23 pairs. Mitosis takes place in all cells of the body except for egg and sperm cells.

...an unexpected change in the structure of DNA can sometimes cause harm to the body.

Meiosis takes place only in reproductive cells, and is the cell

division process by which egg and sperm are formed. During the cell division process of meiosis, there is a reduction in the number of chromosomes that results in egg and sperm cells that contain 23 chromosomes, or half of the usual number of 46. Egg and sperm cells are called haploid cells because they have a single copy of each chromosome instead of the usual two copies (GHR, 2008i).

Meiosis allows for genetic variation through a natural process of breaking and rejoining DNA strands to create new combinations of genes and therefore generating genetic variation (GHR, 2008d). During meiosis, the paired chromosomes come together to prepare for cell division, and during this process portions of the chromosomes cross over so that there is an exchange of genetic material before the chromosomes separate. This process is called recombination, and it creates greater diversity among the eggs and sperm.

When meiosis takes place, a pair of chromosomes may fail to separate properly, creating a sperm or egg that has either two copies or no copy of a specific chromosome. This is a sporadic event and it is called nondisjunction. Nondisjunction can lead to an extra chromosome, called trisomy, or a missing chromosome, called monosomy (GHR, 2008l). Down syndrome is an example of trisomy. Individuals who have Down syndrome have an extra chromosome number 21. Turner syndrome is an example of monosomy. Girls who have Turner syndrome have only one X chromosome. This causes them to have short stature and be infertile (NHGRI, 2008l).

Gene Mutations

Sometimes a mutation can improve an organism's chance of surviving and passes the beneficial change on to its descendants.

Within the cells of the human body many complex interactions take place that regulate and express human genes. Changes in the structure and function of a gene and the process of protein synthesis may affect a person's health. A permanent change in the structure of DNA is called a mutation. Most of the time DNA changes either have no effect or else cause harm. Sometimes a mutation can improve an organism's chance of surviving and passes the beneficial change on to its descendants. There are several different types of gene mutations. These include: deletion (loss), duplication (multiplication), inversion, insertion (addition), translocation (rearrangements), and point mutations (changes in base pair sequences) (NHGRI, 2008n). Examples of conditions caused by gene mutations include Huntington Disease, Fragile X syndrome, and Duchenne muscular dystrophy (NHGRI, 2008l).

Gene mutations can be inherited, spontaneous, or acquired. Inherited gene mutations are called germline mutations and they are present in all body cells. Inherited gene mutations are passed on from parent to child in reproductive cells, the egg and sperm, and are passed on to all of the cells in that child's body when the body cells reproduce. This is described in the Genetics Home Reference under Germline Mutation (2008e). The gene alteration that causes cystic fibrosis is an example of a germline mutation.

A spontaneous mutation can occur in individual eggs or sperm at the time of conception. A person who has the new, spontaneous mutation has the risk of passing the gene mutation on to his or her children. Examples of genetic conditions that may take place in a single family member as a result of a spontaneous mutation include Marfan syndrome and Achondroplasia (NHGRI, 2008l).

Acquired mutations, also called somatic mutations, occur in body cells other than egg or sperm. They involve changes in DNA that take place after conception, during a person's lifetime. Acquired mutations happen as a result of cumulative changes in body cells that are other than egg or sperm and are called somatic cells. Somatic gene mutations are passed on when they reproduce to daughter cells (GHR, 2008m).

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Gene mutations take place in the human body all of the time. Cells have special mechanisms that help them to recognize mutations in DNA. In most situations, the cells are able to correct the mutation before it is passed on by cell division. Over time, however, genes may lose the ability to repair damage from gene mutations. This can lead to an accumulation of genetic changes that may result in diseases such as cancer or other conditions of aging, such as Alzheimer disease (Jenkins & Lea, 2005).

Gene Variations

As a result of human genome research, researchers now have new tools and technologies to identify the genetic variations of common diseases such as heart disease, stroke, diabetes, asthma, and common cancers. Genome-wide association studies (GWAS) are one such research approach. GWAS involve the use of gene chips that can look at the complete sets of variable DNA markers (up to about a million!) in many people to find genetic variations associated with a particular disease. When these new genetic associations are identified, researchers can then use the information to develop better ways to detect, treat, and prevent the disease. Successes have been reported using GWAS to identify genetic variations that contribute to risk of type 2 diabetes, Parkinson's disease, heart disorders, obesity, Crohn's disease, and prostate cancer, as well as genetic variations that influence response to anti-depressant medications. GWAS studies are laying the foundation for personalized medicine, in which the current one-size-fits-all approach to medical care will be transformed to more individualized and customized strategies (NHGRI, 2008f).

Genetic Inheritance

Genes are inherited in families in a number of ways. These various ways of genetic inheritance are

called patterns of inheritance. This section will discuss various patterns of inheritance, specifically the Mendelian inheritance patterns of autosomal dominant inheritance, autosomal recessive inheritance, X-linked inheritance, mitochondrial DNA inheritance, and multifactorial inheritance.

One important type of inheritance pattern is called Mendelian inheritance. Mendelian disorders are genetic conditions that are passed on in families in fixed proportions. They are caused by gene mutations that are present on one or both chromosomes of a pair. One gene inherited from one or both parents can cause a Mendelian disorder. Mendelian disorders are classified according to how they are inherited in families. These disorders are autosomal dominant, autosomal recessive, and X-linked disorders ([NHGRI, 2008h](#)). The terms dominant and recessive refer to the disorder, trait, or physical presentation of the disorder. These terms do not refer to the specific gene or genes that can cause the observable characteristics.

Autosomal Dominant Inheritance

Autosomal dominant inheritance is a pattern of Mendelian inheritance in which an affected individual has one copy of a gene mutation and one normal version of the gene. Individuals who have autosomal dominant disorders have a 1 in 2 (50%) chance to pass on the gene mutation and therefore the disorder to their children. Examples of autosomal dominant diseases include Huntington's disease, polycystic kidney disease, and neurofibromatosis ([GHR, 2008c](#)). Autosomal dominant inherited disorders frequently present with varying degrees of severity. Some individuals may have significant symptoms, while others may have mild symptoms. This aspect of dominantly inherited disorders is called variable expression, and results from the influence of other genetic and environmental influences, such as other medical conditions or diseases, diet, and medications, on the clinical presentation ([GHR, 2008e](#); [NHGRI, 2008a](#)).

Autosomal Recessive Inheritance

Genetic disorders that are inherited in an autosomal recessive manner occur only in patients who have received two copies of a gene mutation, one from each parent ([NHGRI, 2008k](#)). Parents who are carriers do not show any symptoms, as they have a copy of one normally functioning gene. Examples of autosomal recessive inherited disorders include cystic fibrosis, sickle cell anemia, and phenylketonuria (PKU).

X-Linked Inheritance

X-linked genetic disorders (also called sex-linked) are caused by gene mutations on the X chromosome. Most often X-linked genetic disorders are seen in males. Males inherit the X chromosome from their mother and the Y chromosome from their father. Because males have only one X chromosome, if they inherit a gene mutation on the X chromosome from their mother, they will have the disorder. Examples of X-linked genetic disorders occurring in males include hemophilia and Duchenne muscular dystrophy ([GHR, 2008o](#)).

Mitochondrial DNA Inheritance

Another type of inheritance pattern is called mitochondrial inheritance. Mitochondria are small, energy-producing centers inside of body cells. Each mitochondria contains a small amount of DNA. Disorders that follow a mitochondrial inheritance pattern result from mutations in mitochondrial DNA (also called mtDNA). Mitochondrial disorders can affect both males and females, but only females can pass mutations in mitochondrial DNA to their children. The reason that only females pass on mitochondrial DNA to their children is that their ova contain mitochondrial DNA whereas sperm do not ([GHR, 2008h](#)). A woman with a disorder caused by changes in mitochondrial DNA will pass the mutation to all of her daughters and sons. The children of a man with a mitochondrial disorder, however, will not inherit the mutation ([GHR, 2008f](#)).

Multifactorial Inheritance

Multifactorial inheritance involves the combined contribution of multiple genes and environmental factors, often unknown, as the cause of a particular disease or trait ([GHR, 2008j](#)). Examples of environmental factors that may contribute to multifactorial diseases include chemicals, such as metals and solvents; biological agents, such as toxins released from mold and bacteria; and lifestyle factors, such as diet and physical activity. Common examples of multifactorial disorders include neural tube defects, such as spina bifida and anencephaly ([GHR, 2008k](#)). Multifactorial disorders are also called complex medical conditions or disorders, for example diabetes, heart disease, and obesity. Multifactorial disorders and complex conditions and traits may cluster in families, but they do not have a characteristic pattern of inheritance like autosomal dominant and recessive inherited disorders.

Chromosomal Variation and Genetic Conditions

Differences in the number or structure of chromosomes are a significant cause of birth defects, mental retardation, and malignancies, as described earlier under Cell Division. Chromosomal differences usually involve an extra or missing chromosome. These chromosomal differences are generally not inherited but take place as random events during the formation of egg or sperm. An error that occurs in the cell division process, while either the egg or sperm is being formed, can result in an abnormal number of chromosomes. When one of these reproductive cells contributes to the genetic makeup of a baby, that baby will have an extra or missing chromosome in each of his or her cells. The most common example of this type of chromosomal abnormality is Down syndrome ([GHR, 2008a](#)).

Chromosomal disorders can also be caused by changes in the structure of one or more chromosomes. These are not as common as chromosomal disorders that are caused by a missing or extra chromosome. Some structural chromosome changes can be inherited. For example, an individual may carry what is called a "balanced" chromosome rearrangement. This means the individual has all of their chromosomal material, but it is rearranged. A woman who carries a balanced chromosome rearrangement has an increased chance of having a spontaneous pregnancy loss and of having children with an unbalanced chromosome rearrangement that can cause physical and/or mental disabilities. Other chromosomal rearrangements occur as a result of an accidental event during the formation of egg or sperm, or during early fetal development. These are not commonly inherited ([GHR, 2008b](#)).

Application of Genetics and Genomics to Nursing Practice

Genetics and genomics discoveries are leading to new and improved methods for screening, diagnosis, prevention, and treatment of rare and common diseases. Members of the general public will increasingly expect that all registered nurses, including RNs, APNs, and nurse specialists, will use genetic and genomic information and technologies when they are providing nursing care ([Consensus Panel, 2006](#)). Nurses at all levels and in all areas of practice will soon be participating in risk assessment for genetic conditions and disorders, explaining genetic risk and genetic testing, and supporting informed health decisions and opportunities for early intervention ([Skirton, Patch, & Williams, 2005](#)). The *Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics* states:

To care for persons/families/communities and/or populations throughout the life span, registered nurses will need to demonstrate proficiency with incorporating genetic and genomic information into their practice ([Consensus Panel, 2006, p.7](#)).

Examples of applications of genetics and genomics in nursing practice include the following:

- recognizing a newborn infant that is at risk for morbidity or mortality due to genetic metabolism errors
- identifying an asymptomatic adolescent who is at high risk for hereditary colorectal cancer based on his or her family history
- identifying a couple that is at risk for having a child with a genetic condition
- assisting with the selection of a drug or dose of a drug, based on genetic markers, in the treatment of an adult who has cancer
- helping any patient or family that has questions about genetic or genomic information or services to find reliable information. ([Consensus Panel, 2006](#)).

Nurse educators, with guidance from the *Essential Nursing Competencies and Curricula Guidelines in Genetics and Genomics (2006)*, will soon be designing and implementing learning experiences that will help students at all levels, along with practicing nurses achieve the genetic and genomic competencies they will need for practice. It is expected that each nursing curriculum preparing nurses for practice at any level will incorporate genetics and genomics topics and learning experiences into existing classes. To help support this integration, the *Essential Nursing Competencies and Curricula Guidelines in Genetics and Genomics* includes a comprehensive listing of resources for nurse educators ([Consensus Panel, Appendix A, 2006](#)).

It is expected that each nursing curriculum preparing nurses for practice at any level will incorporate genetics and genomics topics and learning experiences into existing classes.

This section will now focus on several examples of nursing roles at all levels of practice in genetic and genomic healthcare including family history collection and pedigree construction, referral for genetic consultation, genetic testing, pharmacogenetics and pharmacogenomics, and a new type of genetic testing called direct-to-consumer genetic testing.

Family History

Knowledge of the role of family history in common and rare genetic conditions and disorders is a first step in genetic and genomic risk assessment and early intervention. The United States (U.S.) Surgeon General, in cooperation with the U.S. Department of Health and Human Services and other government agencies, launched a national public health campaign in 2004 called the *U.S. Surgeon General's Family History Initiative* in acknowledgement of the important role of family history in health and disease. The Initiative encourages all American families to learn more about their family history. It offers a computerized tool to help families create a portrait of their family health. Although Americans know that family history is important to their health, as many as one-third have not ever tried to gather and write down their family's health history ([U.S. Department of Health and Human Services, 2008](#)). Nurses can help families learn about their family health history by offering them information about the U.S. Surgeon General's Family History Initiative.

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Nurses will also need to be prepared to collect a three-generation, family-health history and construct a pedigree from the collected family-history information, using the standardized symbols and terminology ([Consensus Panel, 2006](#)). When collecting family history, nurses need to be alert for clients who present with family histories having multiple generations affected with a particular disorder (autosomal dominant inheritance), or those with multiple siblings who have a genetic disorder (autosomal recessive inheritance), or those affected with a disease or condition at an early age (multiple generations with early onset breast and ovarian cancer). When a client is identified as having a family history of a possible inherited genetic disorder, the nurse discusses the family history with the healthcare team and lets the client know that he or she has a risk factor for a disease. The nurse and healthcare team can then offer the patient genetic resources and referral to genetic specialists, including medical geneticists, genetic counselors, and genetics nurse specialists ([Consensus Panel, 2006](#)).

Genetic Testing

Genetic and genomic discoveries have led to the development of an increasing number of genetic tests that can be used to identify a trait, diagnose a genetic disorder, and/or identify individuals who have a genetic predisposition to diseases, such as cancer or heart disease. There is now genetic testing for more than 1,600 genetic disorders ranging from single-gene disorders, such as cystic fibrosis, to complex disorders, such as diabetes ([GeneTests, 2008](#)). Nurses in all practice settings and at all levels of practice will increasingly be involved with identifying patients who could benefit from genetic

testing, educating patients about the nature and purpose of a genetic test, and using genetic testing to guide prevention and treatment of rare and common diseases ([Consensus Panel, 2006](#)).

Genetic testing is now used in prenatal, pediatric, and adult populations. Prenatal genetic testing is used to screen for and diagnose genetic conditions, such as Down syndrome. Carrier testing helps to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder, such as cystic fibrosis. Carrier testing is offered to people who come from certain ethnic groups that have an increased risk of specific genetic disorders, such as Tay-Sachs disease among Ashkenazi Jewish populations. When both parents are tested, the test gives information about a couple's chance of having a child with a genetic disorder ([GHR, 2008n](#)).

Newborn screening has expanded to include an increasing number of genetic disorders that could prove to be dangerous if left untreated. Currently many states offer screening for over 29 conditions through their newborn screening programs. Hence, more genetic disorders are being identified in the early newborn period so that interventions and treatments can be administered promptly. Phenylketonuria (PKU) is an example of a genetic disorder for which newborn screening is available ([GHR, 2008n](#)).

Diagnostic genetic testing is used to confirm a diagnosis of a particular genetic disorder when it is suspected based on physical symptoms or positive newborn screening results. Results of a diagnostic test can influence the management of the genetic disorder and a person's choices about healthcare ([GHR, 2008n](#)).

Another use of genetic testing is preimplantation genetic testing, also called preimplantation genetic diagnosis (PGD). PGD is used to identify genetic changes in embryos that are created using assisted-reproductive technologies, such as in-vitro fertilization. During in-vitro fertilization, a woman's egg cells are removed from her ovaries and fertilized with sperm outside of her body. A small number of cells are then taken from the embryos and tested for gene mutations or variations. Those embryos without the gene mutations or variations are then implanted in the woman's uterus to begin her pregnancy ([GHR, 2008n](#)).

Genetic testing is increasingly offered to adults who are at risk for early onset disorders, such as breast cancer, colon cancer, and hemochromatosis ([NHGRI, 2008i](#)). This type of genetic testing is called presymptomatic or predisposition genetic testing. A presymptomatic genetic test is a test for a completely penetrant single-gene disease (meaning that if a person has the disease-causing gene, he or she will develop the disorder). Predisposition genetic testing is a test for a genetic predisposition (incompletely penetrant conditions). Not all individuals who have a positive genetic test result will develop the disease during their lifetime. Presymptomatic and predisposition genetic tests provide asymptomatic individuals with information about whether they have a gene mutation and/or their risk to develop a particular disorder. Presymptomatic and predisposition genetic testing are offered to individuals who come from families with a known adult-onset condition. Information gained from genetic testing indicates an increased or decreased risk for developing the disease or disorder, and also allows for earlier interventions and treatment ([GHR, 2008n](#); [NHGRI, 2008j](#)).

Pharmacogenetics and Pharmacogenomics

Pharmacogenetics looks at a person's genetic information regarding how drugs are transported and metabolized in their body...

Our knowledge about the role of genetics, in regard to the way in which a person's body breaks down and responds to certain medicines, is continually increasing. Two new fields of the study and application of genetics and genomics for medical treatment of disease are evolving. One new field is that of pharmacogenetics which deals with the variability of individuals' responses to medications due to genetic variation. Pharmacogenetics looks at a person's genetic information regarding how drugs are transported and metabolized in their body and their specific drug receptors. The aim of pharmacogenetics is to create an individualized drug therapy program, thereby allowing for the best choice and dose of drugs. For example, genetic testing for certain genes (CYP2C9 and VKORC1) that affect the metabolism of warfarin, a blood thinner, can now be done to guide warfarin dosage and management and to help prevent adverse side effects ([Lea, Feero, & Jenkins, 2008](#)).

Another new field is that of pharmacogenomics which looks for genetic variations that are associated with drug discovery and development. Pharmacogenomic research is leading to the development of drugs that can be tailor made for specific individuals and adapted to each person's own genetic makeup. An individual's environment, diet, age, lifestyle, and state of health can all affect the person's response to medicines. In addition, understanding an individual's genetic makeup can help create personalized drugs that are most effective, and have fewer side-effects, for a specific person ([NHGRI, 2008c](#)). For example, it is now known that patients who have advanced colorectal cancer, and who have a mutation in the gene called KRAS in their tumors, should not receive both chemotherapy and the medication called cetuximab (Erbix), because they are not likely to benefit from the combined treatment. Avoiding this combined treatment could spare them the side effects and cost of this treatment. Experts predict that it will soon become standard practice to test all colorectal tumors for mutations in the KRAS gene before starting those patients who have advanced disease on treatments that involve cetuximab ([National Cancer Institute, 2008a](#)).

There is also a test currently used to determine whether a medicine called Herceptin will be an effective treatment in breast cancer. The test determines whether a woman who has metastatic breast cancer tests positive for Human Epidermal Growth Factor Receptor 2 (HER2). Those women who have HER2-positive breast cancer have been found to have a more aggressive form of the disease, a greater likelihood of recurrence, a poorer prognosis, and a decreased chance of survival, when compared with those women who have HER2-negative breast cancer. Herceptin, which is designed to target and block the function of HER2 protein overexpression, is given

Pharmacogenomic research is leading to the development of drugs that can be tailor made for specific individuals...

to those women who test positive for HER2 ([National Cancer Institute, 2008b](#)).

Direct-to-Consumer Genetic Testing

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Genomic research has also opened the door for private, for-profit companies to use genomic information to develop and market genetic testing directly to consumers via the Internet. There are a growing number of companies that offer direct-to-consumer (DTC) genetic testing to assess genetic health risks. DTC genetic tests usually involve scraping cells from the inside of the cheek and mailing the sample to a testing laboratory that will perform the test. The company analyzes the gene-sequence data, interprets it for the risk information it reveals, and creates a report. The report is sent to directly to the consumer. In some situations, a healthcare provider is not involved. In other situations DTC genetic testing means that healthcare providers, including nurses, will have to deal with the complex genetic test results for common diseases without knowledge, training, or clear guidelines on the subject ([Hunter, Khoury, & Drazen, 2008](#)). Other concerns raised by DTC genetic testing include the possibility that the public may

misinterpret the genetic information that is given directly to them, or the possibility that they may order genetic tests that are inappropriate. Several organizations, including the National Institutes of Health, are developing programs and resources to educate the general public and healthcare professionals about genetic testing ([Feero, Guttmacher, & Collins, 2008](#)).

Nursing practice will increasingly involve these new directions in healthcare. Nurses will need to expand their knowledge and skills in tailoring genetic and genomic information and testing to their patients. Having knowledge of both the benefits and limitations of emerging genetic tests will enable nurses to assist consumers to be informed, healthcare decision makers ([Lea, Feero, & Jenkins, 2008](#)).

Summary

Genetics and genomics are increasingly being integrated into the screening, prevention, diagnosis, and treatment of rare and common diseases. Nurses are on the frontlines of patient care and need to become knowledgeable about and competent in genetic- and genomic-related healthcare ([Consensus Panel, 2006](#)). Nurses also need to know how to find reliable genetic and genomic information for themselves and their patients, and where to refer patients and families for further genetic information and counseling. [Tables 2](#) and [3](#) provide resources for nurses regarding these activities. Knowledge of basic genetics, genetic principles, and genomic applications and resources will give nurses the foundation to provide competent genomic healthcare today and in the future.

Table 2: Basic Genetics Resources
<p>Genetics and Genomics for Health Professionals - Genetics and Genomics for Health Professionals provides reliable, up-to-date genetics and genomics information related to patient management, curricular resources, new National Institutes of Health and NHGRI research activities, and ethical, legal, and social issues. www.genome.gov/27527599</p>
<p>Genetics 101 for Health Professionals, National Institutes of Health - The resources in this section provide basic information about genetics and genomics in clear language and provide links to online resources. www.genome.gov/27527637</p>
<p>Talking Glossary of Genetic Terms, National Human Genome Research Institute - The Talking Glossary helps people without scientific backgrounds understand the terms and concepts used in genetic research. Simply click on the term of interest to open a page with a wealth of information, including the term's pronunciation, audio information, images, and additional links to related terms. Students, teachers, and parents will find the glossary an easy-to-use, always available learning source on genetics. www.genome.gov/page.cfm?pageID=10002096</p>
<p>From the Blueprint to You, National Human Genome Research Institute - This educational resource explores the world of genetics, DNA, the Human Genome Project, the ethical, legal and social implications of genetic research, and the future of genomics. www.genome.gov/Pages/Education/Modules/BluePrintToYou/Blueprint3to4.pdf</p>
<p>Handbook: Help Me Understand Genetics, Genetics Home Reference, National Library of Medicine - Presents basic information about genetics in clear language and provides links to online resources. http://ghr.nlm.nih.gov/handbook</p>
<p>Learn.Genetics, Genetic Science Learning Center, University of Utah - Learn.Genetics delivers educational materials on genetics, bioscience, and health topics. They are designed to be used by students, teachers, and members of the public. http://learn.genetics.utah.edu/</p>
<p>DNA from the Beginning - Provides an animated primer on the basics of DNA, genes, and heredity. www.dnafb.org/dnafb/</p>

Table 3 – Clinical Genetics Resources

Genetics and Rare Diseases Information Center (GARD) - Established by the National Human Genome Research Institute (NHGRI) and the Office of Rare Diseases Research (ORDR), the Genetic and Rare Diseases Information Center (GARD) employs experienced information specialists to answer questions in English and Spanish from the general public, including patients and their families, health care professionals, and biomedical researchers. www.genome.gov/10000409

Genetics and Genomics for Patients and the Public - Genetics and Genomics for Patients and the Public provides everything from detailed information about genetic disorders, background on genetic and genomic science, the new science of pharmacogenomics, tools to create your own family health history, and a list of online health resources. www.genome.gov/19016903

Genetics Home Reference - Genetics Home Reference provides consumer-friendly information about the effects of genetic variations on human health. <http://ghr.nlm.nih.gov/>

Gene Reviews – Features expert-authored, peer-reviewed, current disease descriptions that apply genetic testing to the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions. www.genetests.org/servlet/access?id=8888891&key=etn1HekazydBi&fcn=y&fw=8PQs&filename=/about/content/reviews.html

Office of Rare Diseases Research (ORDR) – ORDR coordinates research and information on rare diseases at the NIH and for the rare diseases community. The ORDR Web site provides information for patients and their families with rare diseases and about NIH- and ORDR-sponsored biomedical research and scientific conferences. <http://rarediseases.info.nih.gov/>

U.S. Surgeon General's Family History Initiative, Family History Tool - This web-based tool helps users organize family history information and then print it out for presentation to the family doctor. In addition, the tool helps users save their family history information to their own computer and even share family history information with other family members. www.hhs.gov/familyhistory The tool can be accessed at <https://familyhistory.hhs.gov/>.

Author

Dale Halsey Lea, MPH, RN, CGC, FAAN
E-mail: lead@mail.nih.gov

Dale Halsey Lea is a registered nurse and a Board Certified genetic counselor with more than 20 years experience in clinical and educational genetics. She is currently the Health Educator with the Education and Community Involvement Branch and the Genomic Healthcare Branch, National Human Genome Research Institute. As Health Educator, Ms. Lea develops consumer and health professional genetics and genomics health education and community involvement programs and resources; translates genetic and genomic research results into terms understandable by lay audiences; conducts genetics research for the Education and Community Involvement Branch; and provides administrative support for public and health professional education and community involvement programs. Ms. Lea is widely published in the nursing and genetics/genomics literature in the area of integrating genetics into nursing practice. In 2001, Ms. Lea was inducted as a new Fellow in the American Academy of Nursing, and serves on their Expert Panel on Genetics.

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