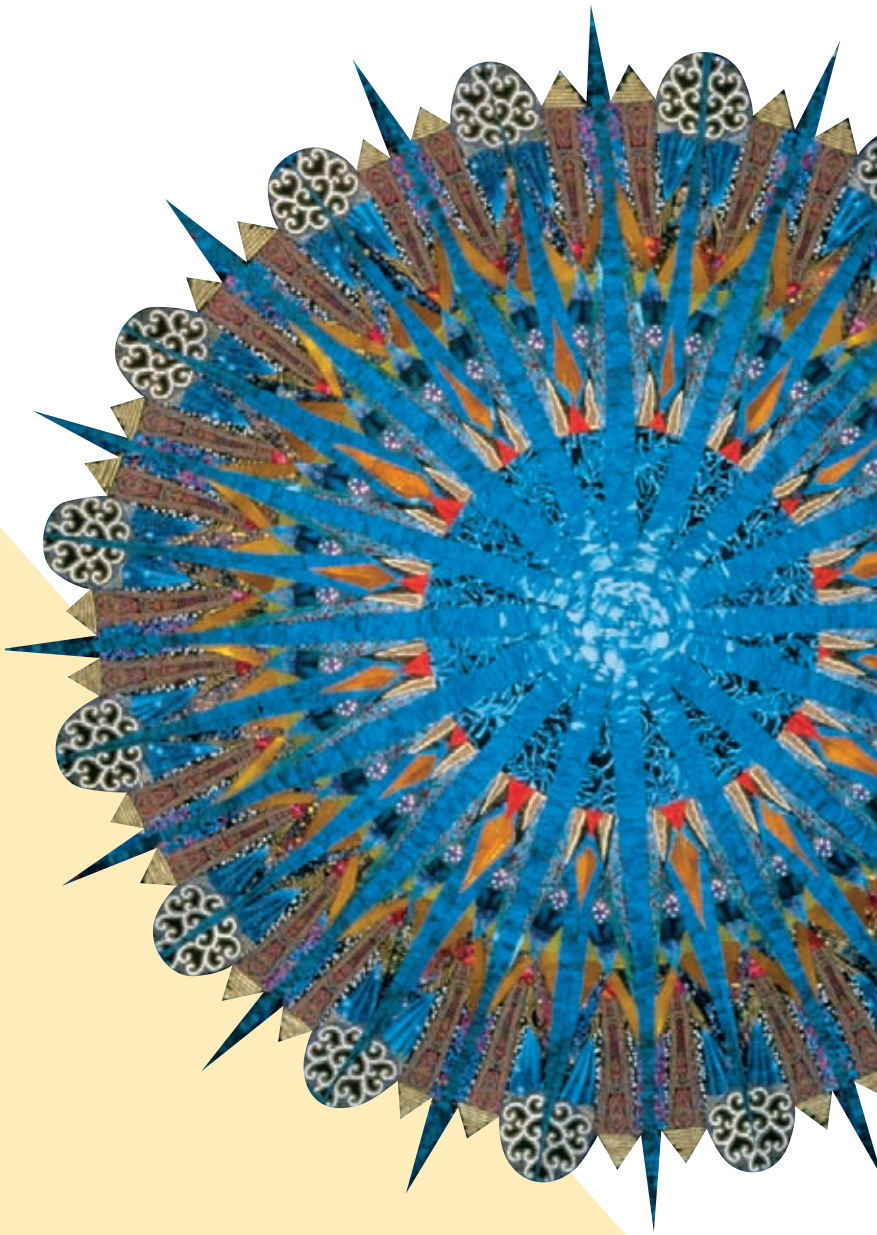
UNIT 3 Pathophysiology and Patterns of Health



CHAPTER 8 Genetic Implications of Adult Health Nursing

LEARNING OUTCOMES

- Discuss the role of genetic concepts in health promotion and health maintenance.
- Apply knowledge of the principles of genetic transmission and risk factors for genetic disorders.
- Describe the significance of delivering genetic education and counseling follow-up in a professional manner.
- Identify the implications of genetic advances on the role of nurses with particular attention to spiritual, cultural, ethical, legal, and social issues.
- Identify the significance of recent advances in human genetics and the impact on healthcare delivery.

CLINICAL COMPETENCIES

- Integrate genetic physical assessment and the use of a pedigree family history into delivery of nursing care.
- Identify clients or families with actual or potential genetic conditions and initiate referrals to a genetics professional.
- Prepare clients and their families for a genetic evaluation and facilitate the genetic counseling process.
- Integrate basic genetic concepts into client and family education and the reinforcement of information provided to clients by genetic professionals.

MEDIALINK

Resources for this chapter can be found on the Prentice Hall Nursing MediaLink DVD-ROM accompanying this textbook, and on the Companion Website at http://www.prenhall.com/lemone

KEY TERMS

alleles, *151*
autosomes, *149*
carriers, *153*
chromosomes, *149*
DNA-based tests, *157*
gene, *151*
gene expression, *155*
genetic locus, *151*
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The genetic knowledge obtained from the Human Genome Project (Box 8–1) has changed not only the way disease treatment is approached but, more importantly, how nurses look at health promotion and health maintenance. DNA is at the center of the state of our health (Figure 8–1 ■). We now know that good health is associated with genes functioning properly and if genes are not functioning properly, disease or an increased risk for disease can result. This includes not only the traditional genetic disorders, but also common complex diseases such as heart disease, stroke, diabetes, and several kinds of cancer. The knowledge gained from **human genome** research has and will have a profound impact on the prevention, diagnosis, and treatment of genetic disorders and complex diseases.

MEDIALINK Human Genome Project Animation

INTEGRATING GENETICS INTO NURSING PRACTICE

Genetic knowledge will continue to revolutionize how persons perceive themselves as well as their health status and their health potential. Therefore, nurses must integrate new genetic knowledge into their nursing practice. This expectation has been defined in the ANA/ISONG *Statement on the Scope and Standards of Genetics Clinical Nursing Practice* (Box 8–2). In addition, a national interdisciplinary group known as the National Coalition for Health Professional Education in Genetics (NCHPEG) has developed competencies to encourage health-

BOX 8–1 Human Genome Project

In 1986, the U.S. Department of Energy (USDOE) announced the Human Genome initiative, and in 1990 the USDOE joined with the National Institutes of Health (NIH) to develop the Human Genome Project (HGP). The ultimate goal was to sequence the human genome and to identify all human genes. The completion of a high-quality reference sequence was announced in April 2003. Information obtained through the sequencing of the human genome has had a tremendous impact on finding the genes associated with human disease. Future research will now be directed toward understanding the complex functions of cellular regulation, human variation, and the interplay of genes and environment and how all the cell organelles, genes, and proteins work together in life's functions (USDOE Genome Programs, 2003).

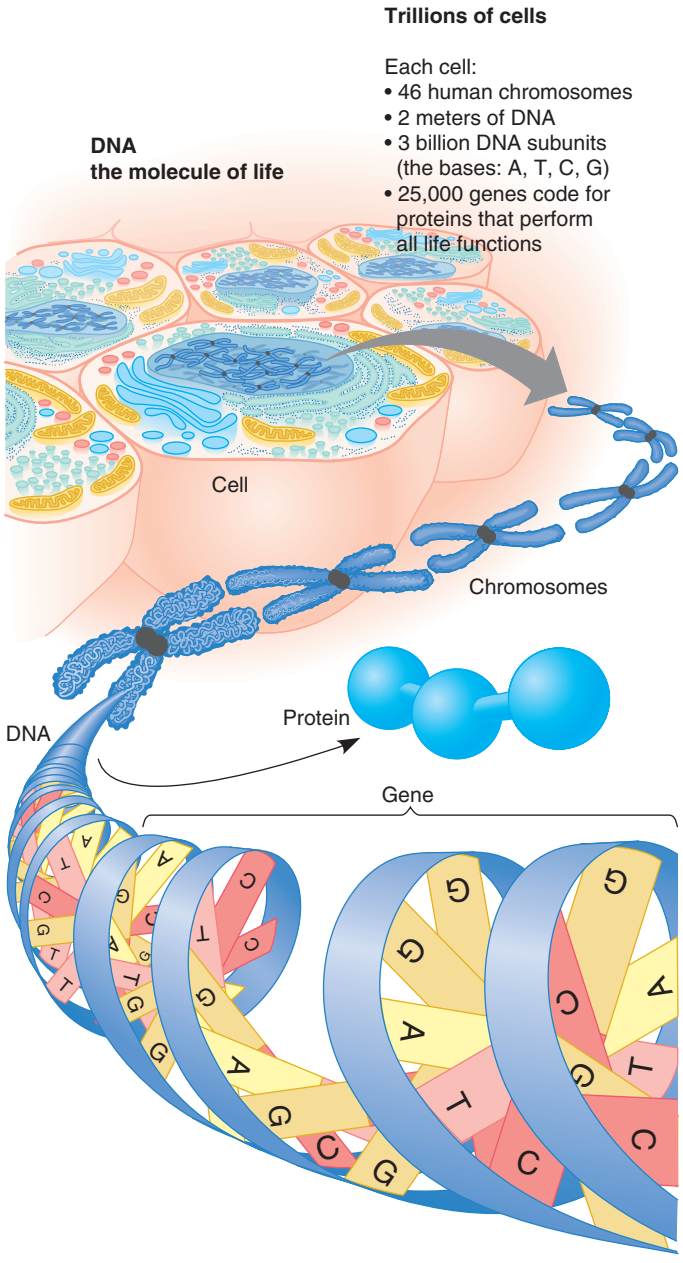


Figure 8–1 ■ Each cell nucleus throughout the body contains the genes, DNA, and chromosomes that make up the majority of an individual's genome. The remaining portion of the human genome is in the mitochondria.

BOX 8–2 ANA/ISONG Standards of Genetics Clinical Nursing Practice Statement

ANA/ISONG *Statement on the Scope and Standards of Genetics Clinical Nursing Practice:*

> All licensed registered nurses, regardless of their practice setting, have a role in the delivery of genetics services and the management of genetic information. Nurses require genetic knowledge to identify, refer, support, and care for persons affected by, or at risk for manifesting or transmitting, genetic conditions. As the public becomes more aware of the genetic contribution to health and disease, nurses in all areas of practice will be increasingly asked to address basic genetics-related questions, service needs, or both.

Source: Used with permission from American Nurses Association and International Society of Nurses in Genetics. Copyright 1998, p. 2. Washington, DC: American Nurses Publishing.

care providers to integrate genetic knowledge, skills, and attitudes while delivering care to clients.

Nurses must have basic genetic knowledge to care for the needs of clients and their families with known or suspected genetic disease. Basic level nursing practice that meets the standard of genetic nursing includes:

- Identifying simple risk factors by completing a genetics focused family history and also drawing a three-generation pedigree
- Incorporating a genetics focus into physical assessments
- Applying concepts of health promotion and health maintenance to assist the client and family to make informed decisions and lifestyle choices
- Collaborating with the client to perform interventions that include advocacy, supporting the client's decisions, teaching, making referrals, clarifying information, and providing information about available resources and services
- Partnering with the community to educate the public and supporting legislation that protects genetic information and those with genetic conditions from discrimination
- Completing an evaluation of the delivery of care for the client
- Applying knowledge of the ethical, legal, and social implications of genetic information.

Through the application of fundamental genetic concepts, nurses can significantly improve the nursing care provided to clients.

GENETIC BASICS

A basic knowledge of the cell, DNA, cell division, chromosomes, and genes is essential to deliver the genetic standard of care to the adult client (Box 8–3).

The cell is the basic unit of life and it is the working unit of all living systems. Life starts as a single cell, but the developed human body is made up of trillions of cells. These cells share common features such as a nucleus that contains 46 chromosomes, and organelles such as mitochondria. There are many different types of "specialized" cells that function differently depending on their location. For example, pancreatic cells have a very different function than that of nerve cells.

BOX 8–3 Using the People-First Approach

The nurse must incorporate a "people-first" philosophy and use genetic terminology that is sensitive to the maintenance of an individual's positive self-image. This can be accomplished by using "unaltered" or "wild-type" gene instead of "normal" gene and "altered" gene; "altered, disease-producing gene" or "gene alteration," instead of the term "mutated" or "abnormal" gene when communicating genetic concerns to clients, other healthcare providers, or the public.

wild type = normal = expected = unaltered
versus
mutated = abnormal = defective = unexpected = altered

All human cells, except mature red blood cells, contain a complete set of deoxyribonucleic acid (DNA) molecules. DNA molecules consist of long sequences of nucleotides or bases represented by the letters A, G, T, and C. The order of these bases gives the exact instructions for the functioning of that particular cell. Writing the correct order of the bases using the above abbreviations represents the sequence of the bases in DNA. All of the DNA in a human cell is referred to as the human genome, or the complete set of inheritance for an individual. The human genome includes the DNA in the cell nucleus as well as the DNA found in the mitochondria, which will be discussed later in this section. Each person's genome is unique. Identical (monozygotic) twins are the exception because they develop from only one fertilized ovum and share identical DNA (Guttmacher & Collins, 2002).

The cell nucleus contains about 6 feet of DNA that is tightly wound and packaged into 23 pairs of chromosomes making a complete set of 46 **chromosomes**. The structure and number of chromosomes can be shown by a karyotype, or picture, of an individual's chromosomes (Figure 8–2 ■) There are two copies of each chromosome. One copy, or half of the complete set of these 46 chromosomes, is inherited from the mother and the other copy, or the other half of the 46 chromosomes, is inherited from the father. For example, an individual will have two of chromosome 1, one inherited from her mother and one inherited from her father. These two copies or pairs of inherited chromosomes are called **homologous chromosomes**. Chromosomes are numbered according to size, with chromosome 1 being the largest and chromosome 22 being the smallest. The first 22 pairs of chromosomes, known as **autosomes**, are alike in males and females. The 23rd pair, the **sex chromosomes**, determines an individual's gender. A female has two copies of the X chromosomes (one copy inherited from each parent) and a male has one X chromosome (inherited from his mother) and a Y chromosome (inherited from his father).

Cell Division

Mitosis and meiosis are the two types of cell division in human cells. **Mitosis** is the process of making new cells and it takes place in the **somatic**, or tissue, **cells** of the body. Cell division through mitosis heals wounds and replaces cells lost daily on skin surfaces and in the lining of gastrointestinal and respiratory

MEDIALINK National Coalition for Health Professional Education in Genetics (NCHPEG) Core Competencies

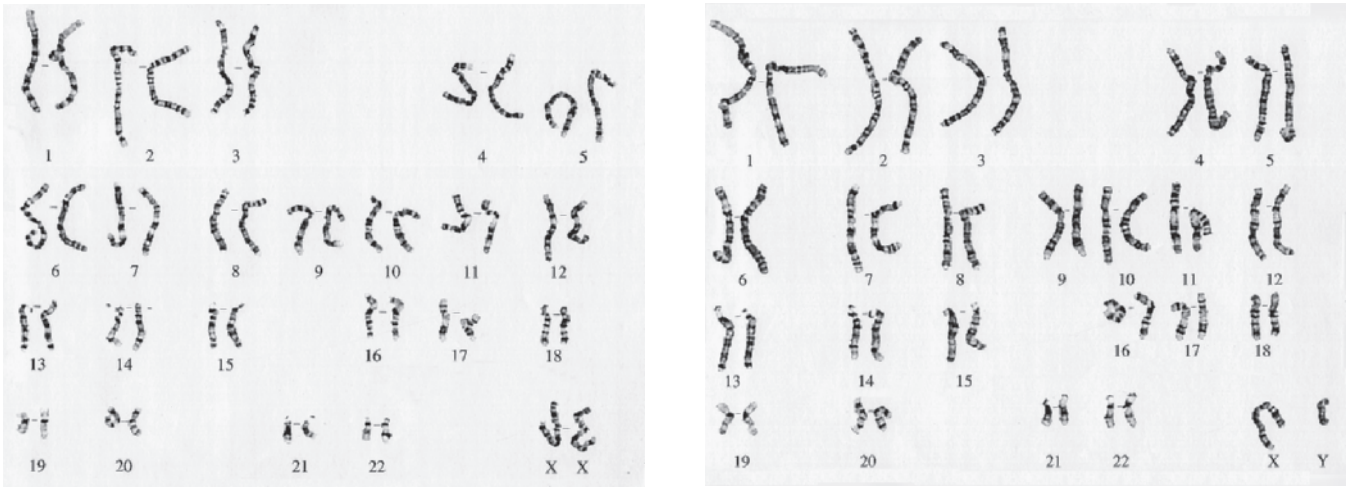


Figure 8–2 ■ A karyotype is a picture of an individual's chromosomes. It shows the chromosomal structure and number of the 22 pairs of autosomes and the sex chromosomes.

tracts. In addition, mitosis is responsible for development. The mitotic activity of the zygote and its daughter cells is the foundation for a human's growth and development. The zygote undergoes mitosis to form a multicellular embryo, then fetus, then infant. Cell division through mitosis results in two cells called *daughter cells* that are genetically identical to the original cell, or *mother cell*, and each other.

Meiosis is also known as the reduction division of the cell. Meiosis occurs only in the sex cells of the testes and ovaries and results in the formation of the sperm and oocyte (gametes). Meiosis is very similar to mitosis in that it is a form of cell division; however, through a series of complex mechanisms, the amount of genetic material is reduced in half (23 chromosomes). This is very important because when the two sex cells combine during fertilization, the total number of chromosomes (46) is present in the offspring's cells. The purposes of meiosis is to produce gametes, to reduce the number of chromosomes by half, and also to make new combinations of genetic material from crossing over and independent assortment processes, which allows diversity in the human population.

Chromosomal Alterations

Alterations in chromosomes often occur during cell division (meiosis or mitosis) and are classified as either alterations in the number of chromosomes or structural alterations. They involve either part of or the whole chromosome. The clinical consequences of number and structural changes in the chromosomes in an individual vary depending on the amount and type of DNA affected by the alterations.

Alterations in Chromosome Number

An increase or decrease in chromosomal numbers can occur during meiosis or mitosis (Box 8–4). Alterations occur often during meiosis because meiosis is a highly specific and complex process and each new daughter cell must contain exactly one chromosome from each homologous pair of chromosomes. During meiosis, the paired chromosomes may fail to separate, resulting in daughter cells with either two copies or no copies of that chromosome. This is known as **nondisjunction**. This creates an egg or sperm cell with either two copies or no copies of a particular chromosome. When these egg or sperm cells are fertilized by a normal gamete that contains 23 copies of all of the chromosomes, a zygote that is monosomic (one member of the chromosome pair is missing) or trisomic (having three chromosomes instead of the usual two) results. These circumstances produce such conditions as Turner syndrome (**monosomy**) or **trisomy 21** (Down syndrome).

BOX 8–4 Variations in Chromosomal Number

Aneuploidy—the condition when extra or missing chromosomes exist; if there is an addition or deletion of chromosome number and the individual lives, physical abnormalities and/or mental retardation is common.
- *Monosomy*—the loss of a single chromosome from a pair; i.e., Turner syndrome (45,XO)
- *Trisomy*—the gain of a single chromosome, making a total of three copies of a certain chromosome; i.e., trisomy 21 or Down syndrome

Euploidy—the presence of the normal number of 46 chromosomes

Polyploidy—the condition when more than two pairs of all the chromosomes are present

Alterations in Chromosome Structure

Alterations in chromosome structure include inversions, deletions and duplications, and translocations. In a chromosomal inversion a segment of a chromosome is reversed, changing the DNA sequence for that portion of the chromosome. It occurs when a chromosome breaks in two places and the piece between the breaks turns upside down and reattaches within the same chromosome. The clinical consequences of an inversion depend on how much chromosomal material is involved, where the inversion occurs, and what type of inversion is present.

A chromosomal alteration that includes a missing (deletion) or additional (duplication) whole chromosome or segment of a

chromosome is an unbalanced rearrangement. An unbalanced rearrangement can result in missing genes, confusing directions from the genes, or too much gene product, which often results in a condition that is not compatible with life or altered physical and/or mental development. An example is *cri du chat* syndrome (mental retardation, crying that sounds like a cat mewing, and low-set ears), which results from a large deletion on chromosome 5.

Translocation (chromosomal reshuffling) occurs when a segment of a chromosome transfers or moves and attaches itself to another chromosome. An example is the reciprocal translocation that is found in 95% of clients with chronic myelogenous leukemia (CML). The contributing translocation occurs between chromosomes 9 and 22 and is known as the Philadelphia chromosome (Ph 1). Unlike the translocation responsible for Down syndrome, which occurs in the germ cells, the translocation responsible for CML occurs in somatic cells and therefore is not inheritable (National Cancer Institute, 2005; Nussbaum et al., 2001).

Genes

The nurse must also have knowledge of genes—what they are, the role genes play in homeostasis, as well as the consequences of gene alterations. How these gene alterations are inherited is also important for nursing interventions and teaching the client who is at risk for or who has a known gene (DNA-based) condition. Knowledge of the function and inheritance of genes is implicit in health promotion as well as health maintenance of the client and his or her family.

A **gene** is a small portion (segment) of the nucleotide (base) sequence of a chromosome DNA molecule that can be identified as having a particular function or characteristic. These segments of DNA within each gene have specific directions for the functioning of the gene. This specific sequence of nucleotides (the genes and the variations therein) is referred to as the individual's **genotype**. Each chromosome contains numerous genes arranged in a linear order. Researchers currently believe there are about 20,000 to 25,000 genes in the human genome. The number of genes present on each chromosome varies. Chromosome 1 is the largest chromosome and has the largest number of genes with 2,968. The Y chromosome has the smallest number of genes with 231 (Human Genome Project [HGP], 2004a).

All genes come in pairs because chromosomes come in pairs. The only exception to all genes being paired are the genes on the sex chromosomes (X and Y) present in males. All genes have a specific location on a specific chromosome. This is known as the **genetic locus**. For example, one of the many genes located on chromosome 19 is a gene for eye color. There may be slight variations or different forms of a gene, for instance, green versus blue eye color, and these different forms or versions of genes are called **alleles**. When an individual has two identical forms (alleles) of a gene they are said to be **homozygous** (homo = same). If an individual has two different forms (alleles) of the gene, they are said to be **heterozygous** (hetero = different). Genes can be described as *altered* or *mutated,* when a change has taken place, or *expressed*, when the gene has an impact on the outward appearance of an individual and/or the functioning of cells. The observable, outward expression of an individual's entire physical, biochemical, and physiologic makeup, as determined by their genotype (alleles) and environmental factors, is referred to as **phenotype**. Phenotype may be expressed or observed as curly or straight hair or the presentation of signs and symptoms of a disease.

Function and Distribution of Genes

Although the function of more than 50% of the genes in the human genome is unknown, we do know that about 2% of the genes give directions to parts of the cell for how to make proteins, what type of proteins to make, and how much of a protein to make (HGP, 2004a). These protein-directing genes are very important to life and functioning as a human being because proteins are very specialized and perform a variety of functions within the cell. These functions include transmitting messages between cells, fighting infection, directing genes to turn "on" or "off," forming structures, as well as sensing light, taste, and smell (Jegalian, 2000). Some gene activities change from moment to moment in response to tens of thousands of intra- and extracellular environmental signals (USDOE Genome Programs, 2003). An example of this is the feedback mechanism that stimulates a cell to produce insulin after eating a candy bar. After eating, a gene on chromosome 11 directs pancreatic cells to produce, modify, and secrete insulin. Although the gene for producing insulin is present in all nucleated cells of the body, it is only functional in insulin-secreting pancreatic cells (Guttmacher & Collins, 2002).

Mitochondrial Genes

Chromosomes in the cell nucleus are not the only site where genes reside. Several dozen that are involved in energy metabolism are located in the cell mitochondria (the "powerhouse" of the cell). **Mitochondria** are concerned with energy production and metabolism. Some cells contain more mitochondria than others, but each mitochondrion contains its own copies of DNA identified as mitochondrial DNA (mtDNA). Because ova have many mitochondria and sperm do not (most mitochondria are located in the tail of the sperm that detaches after fertilization), mtDNA is primarily inherited from the mother. Therefore, mitochondrial genes and any diseases due to DNA alterations on those genes are transmitted through the mother in a matrilineal pattern. This pattern of inheritance is very different from the pattern of inheritance of genes found in the nucleus of the cell (Guttmacher & Collins, 2002). Thus, an affected female will pass the mtDNA mutation to all of her children; however, an affected male will not pass the mtDNA mutation to any of his children (Nussbaum et al., 2001). Signs and symptoms of conditions as a result of mitochondrial gene alterations are primarily involved in high-energy tissues and organs such as skeletal muscles, liver, kidney, brain and nerve cells, ears, eyes, endocrine system, and heart muscle. Symptoms develop over years as unhealthy or dying cells are not replaced. Hypertrophic cardiomyopathy, heart block, seizures, and deafness are also associated with mtDNA gene alterations (Nussbaum et al., 2001).

Gene Alterations and Disease

A protein will malfunction and in many cases cause disease if any kind of alteration (mutation or change) is present in the order of the DNA sequence within a gene. These gene alterations can be inherited from one or both parents or they can be acquired. *Mutations* inherited from a parent (hereditary mutations) are also known as *germline mutations* because the mutation exists in the reproductive sperm or ova of the parent. Consequently, the DNA in every cell of that offspring will have the gene alteration and also can be inherited from generation to generation.

The second kind of gene alteration is an *acquired mutation* or *somatic mutation*. These alterations occur in the DNA of cells of the individual throughout their life. They can result from errors during cell division (mitosis) or from environmental influences such as radiation or toxins (National Institutes of Health [NIH], 1995).

Today, we know that gene alterations are responsible for approximately 6000 hereditary diseases. However, different gene alterations within a particular gene can result in a wide variety of signs and symptoms. For example, the *CFTR* gene for cystic fibrosis is a very large gene located on chromosome 7. More than 800 different mutations of this gene have been reported to cause cystic fibrosis (Wine et al., 2001). The area of the *CFTR* gene that controls mucous production can have more than 300 different gene alterations resulting in a variety of symptoms ranging from mild, to severe, or no symptoms at all (NIH, 1995). Gene alterations, not the genes themselves, cause genetic diseases and conditions.

Other situations where gene alterations cause illness and disease are through gene interaction with the environment. These genes and conditions are referred to as multifactorial (Jegalian, 2000). Alterations in regulatory genes may also occur. Regulatory genes play a part in maintaining homeostasis or normal functioning. A regulatory gene mutation might lead to the loss of expression of a gene, to unexpected expression in a tissue in which it is usually silent, or a change in the time when a gene is usually expressed. An example of a regulatory gene mutation associated with disease includes the insulin gene region that increases the risk of type 1 diabetes (Guttmacher & Collins, 2002).

Gene Alterations That Decrease Risk of Disease

Although it is common to associate gene mutations with disease, it is important to remember that gene mutations can also be helpful and decrease the risk of disease. Gene alterations and genetic variations may also have a protective role in the expression of diseases. A common example is the protective value of the gene alteration that causes sickle cell disease. Those individuals with this gene alteration have protection against malaria. Another, less common example of a "protective" gene alteration is the one on the receptor gene named *CCR5*. This mutation consists of a deletion within the DNA sequence. Persons who are homozygous for the *CCR5* mutation (have two copies of the altered gene) are almost completely resistant to infection with HIV type I, and those who are heterozygous for the deletion (have one copy of the altered gene) progress much slower from the stage of HIV infection to AIDS (Guttmacher & Collins, 2002). As genomic research continues, more and more of these types of beneficial gene alterations will be identified.

Single Nucleotide Polymorphisms

Single nucleotide polymorphisms, or SNPs ("snips"), are one-letter (base pair) variations in the DNA sequence that occur in greater than 1% of the population. In all people, 99.9% of the DNA is identical; SNPs are responsible for differences among individuals. **Polymorphisms** are DNA sequences that have many forms but give the genetic "directions" for the same thing. Most of these differences have no effect on the individual. Some cause subtle differences in numerous characteristics in appearance such as widow's peak, tongue rolling, and attached ear lobes. Other SNPs, however, affect an individual's risk for certain diseases and have a major impact on how the individual responds to environmental factors such as toxins, microbes, and medications. Scientists are mapping these areas of SNPs in order to move to the next step of identifying the multiple genes that are associated with diseases that are not caused by single-gene alterations, but the complex diseases caused by multiple genes such as cancer, cardiovascular disease, some forms of mental illness, and diabetes (HGP, 2004c; Jegalian, 2000).

PRINCIPLES OF INHERITANCE

Knowledge of inheritance allows the nurse to not only offer and reinforce genetic information to clients and their families but also to assist them in managing their care and in making reproductive decisions. The basic underlying principles of inheritance that nurses can apply to inheritance risk assessment and teaching include (1) all genes are paired, (2) only one gene of each pair is transmitted (passed on) to an offspring, and (3) one copy of each gene in the offspring comes from the mother and the other copy comes from the father. Understanding the Mendelian patterns of inheritance is made easier by relating these principles.

Mendelian Pattern of Inheritance

Conditions that are caused by a mutation or alteration of a single gene are known as *monogenic* or *single-gene disorders*. There are more than 6000 known single-gene disorders occurring in about 1 per 200 births (HGP, 2003). The most common gene alterations that result in genetic disorders are categorized into Mendelian inheritance patterns, because they are predictably passed on from generation to generation following Mendel's laws of inheritance. These single-gene mutations follow an autosomal dominant, autosomal recessive, **X-linked recessive**, or **X-linked dominant** inheritance pattern. The first three of these patterns are the most common. Modes of transmission or inheritance for thousands of conditions resulting from monogenic alterations have been identified (Online Mendelian Inheritance in Man, 2003).

Recessive vs. Dominant Disorders

The distinction between recessive and dominant phenotypes or disease presence (expression) is in the amount of gene product (usually proteins) from the unaltered (**wild-type** or normal) gene. When the individual is heterozygous (has one unaltered

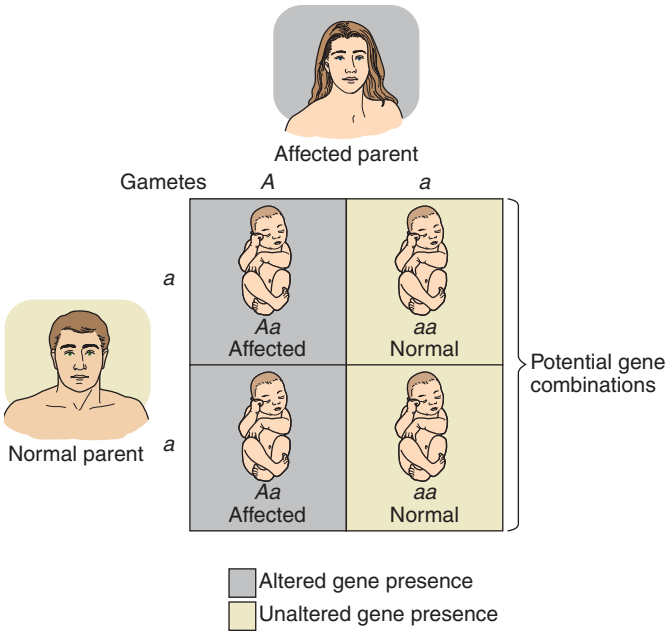


Figure 8–3 ■ This Punnett square shows potential gene combinations (genotypes) and resulting phenotypes of children from parent genotypes with an autosomal dominant altered gene. Phenotypes are expressed (affected) when a male *or* female has one copy of the gene alteration.

gene and one altered gene), the altered gene as well as the disease is classified as recessive if half of the product produced from the unaltered gene is enough to maintain homeostasis and perform the expected function. Therefore, two altered genes must be present to cause a diseased state. If the altered gene causes disease even though the unaltered gene is producing the gene product, then the altered gene as well as the disease is classified as dominant (Nussbaum et al., 2001).

Autosomal Dominant

Autosomal dominant (AD) conditions are the result of an altered gene on any of the 22 autosomes or non–sex chromosomes (Figure 8–3 ■). More than half of the known Mendelian conditions are autosomal dominant. In AD conditions, disease occurs in spite of the fact that there exists one unaltered or normal gene. Also, homozygous dominant conditions are generally much more severe than heterozygous dominant conditions and are often lethal. Because homozygous dominant conditions are usually lethal and would result from *both parents being affected*, the nurse should consider an individual exhibiting an autosomal dominant condition as heterozygous. See Box 8–5 for characteristics of an AD pattern of inheritance.

Autosomal Recessive

A gene or genetic condition is considered recessive when two copies of altered genes are needed to express the condition. Autosomal recessive (AR) conditions are the result of an altered gene on any of the 22 autosomes or non–sex chromosomes (Figure 8–4 ■). An individual with a recessive condition has inherited one altered gene from his mother and one from his father. In most cases, neither of the parents is affected and, therefore,

BOX 8–5 Autosomal Dominant Mendelian Inheritance Characteristics

(Examples: neurofibromatosis, breast and ovarian cancer, autosomal dominant polycystic kidney disease, Marfan syndrome, Huntington disease, familial hypercholesterolemia)

When the nurse gathers a family history, the nurse should assess for any of the following characteristics of autosomal dominant inheritance:

1. Both males and females are affected.
2. Males and females are usually affected in equal numbers.
3. An affected child will have an affected parent and/or all generations will have an affected individual (appearing as a vertical pattern of affected individuals on the family pedigree).
4. Unaffected children of an affected parent will have unaffected offspring.
5. A significant proportion of isolated cases are due to a new mutation.

each of the parents must have a single gene alteration on one chromosome of a pair and the normal, wild-type or unaltered form of the gene on the other chromosome. These parents would be known as **carriers** of the condition and they do not usually exhibit any signs and symptoms of the condition. Because the gene alteration occurs on a non–sex chromosome, both males and females have an equal chance of inheriting the altered gene from their parent. Generally, conditions that are autosomal recessive are more severe and have an earlier onset than conditions with other patterns of inheritance. Most inborn errors in metabolism or metabolic conditions are autosomal recessive. Many are enzyme defects and the functioning of the unaltered gene is sufficient to

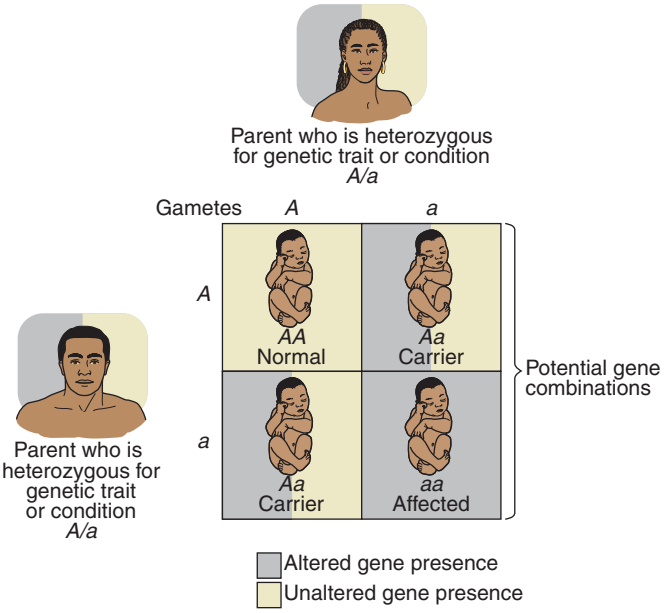


Figure 8–4 ■ This Punnett square shows potential gene combinations (genotypes) and resulting phenotypes of children from parent genotypes with an autosomal recessive altered gene. Phenotypes are expressed (affected) when a male *or* female has two copies of the gene alteration.

provide normal functioning in the person who is heterozygous or the carrier of one copy of the altered gene (Lashley, 2005). See Box 8–6 for characteristics of an AR pattern of inheritance.

X-Linked Recessive

X-linked conditions are the result of an altered gene on the X chromosome. Unlike the autosomes, the sex chromosome, X, is unevenly distributed to males and females. The female has two X chromosomes and the male has only one. Thus, the family history and pattern of inheritance has a characteristic distribution pattern among the males and females in the family (Figure 8–5 ■). Because the male has only one copy of any gene on the X chromosome, it becomes the only copy available to give direction for those particular functions of these genes regardless of whether it is considered dominant or recessive in the female. Thus, if any of these genes are altered, an unaltered counterpart is not present to "override" the altered functioning gene.

The consequences of the altered gene on an X chromosome will be expressed in all males. Females, on the other hand, will have two copies and the unaltered gene generally compensates for the altered gene, making the female a carrier. The male receives his X chromosome from his mother and his Y chromosome from his father. The female offspring receives an X chromosome from each of her parents. Thus, all affected males will pass on the altered X chromosome to all of his daughters who will be carriers of the altered gene. A male can never transmit an altered gene on the X chromosome to his sons because the male will transmit only the Y chromosome to his sons. Because of these transmission patterns, the most commonly occurring transmission of an X-linked condition is through a female who is a carrier of an altered gene. See Box 8–7 for characteristics of an X-linked recessive pattern of inheritance.

BOX 8–6 Autosomal Recessive Mendelian Inheritance Characteristics

(Examples: hemochromatosis type 1, cystic fibrosis, phenylketonuria, sickle cell anemia)

When the nurse gathers a family history, the nurse should assess for any of the following characteristics of autosomal recessive inheritance:

1. Both males and females are affected.
2. Males and females are usually affected in equal numbers.
3. An affected child will have an unaffected parent but may have affected siblings (appearing as a horizontal pattern of affected individuals on the family pedigree).
4. The condition may appear to skip a generation.
5. The parents of the affected child may be consanguineous (close blood relatives).
6. The family may be descendants of a certain ethnic group that is known to have a more frequent occurrence of a certain genetic condition.

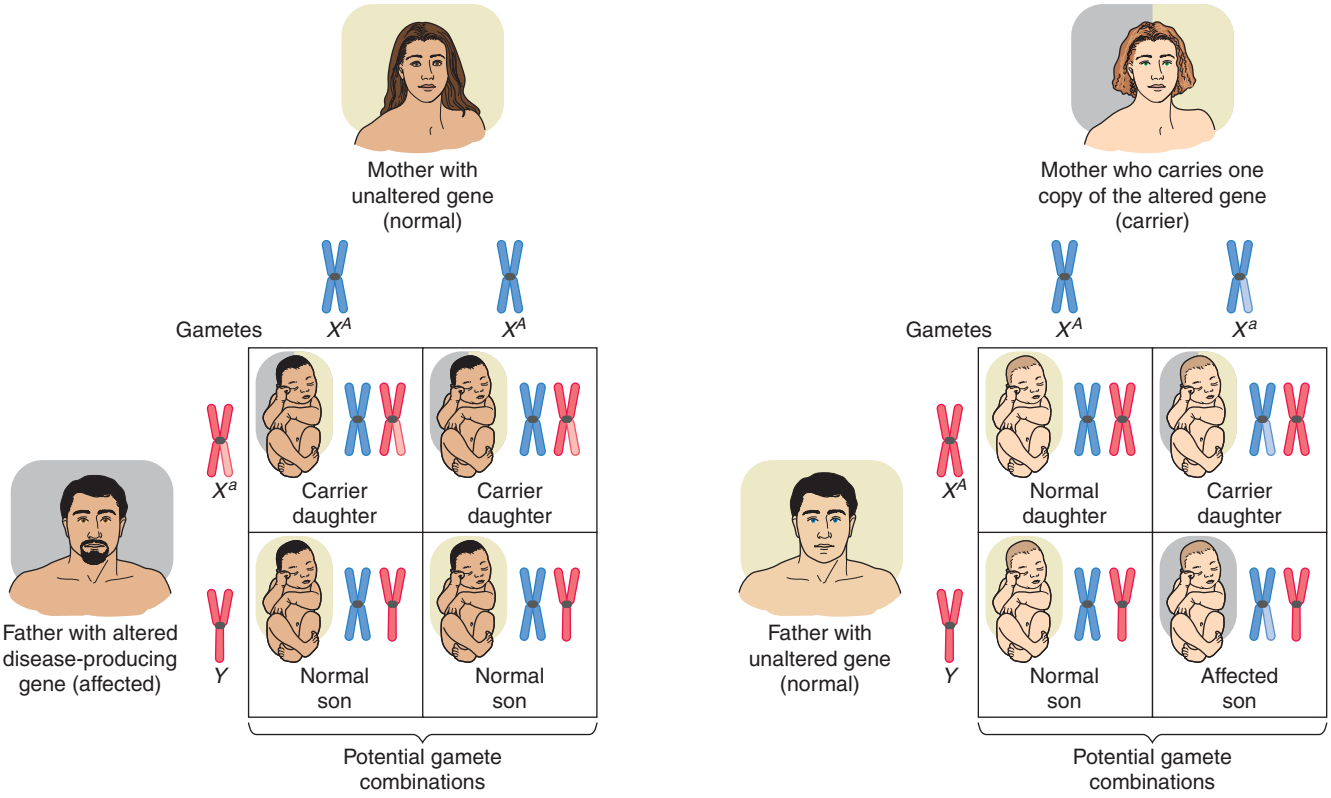


Figure 8–5 ■ These Punnett squares show potential gene combinations (genotypes) and resulting phenotypes of children from different parent genotypes with an X-linked recessive altered gene. Phenotypes are expressed (affected) in a male with only one copy of the gene alteration and in a female with two copies of the altered gene.

BOX 8–7 X-Linked Recessive Mendelian Inheritance Characteristics

(Examples: hemophilia A; Duchenne muscular dystrophy)

When the nurse gathers a family history, the nurse should assess for any of the following characteristics of X-linked recessive inheritance:

1. More males will be affected than females; rarely seen in females.
2. An affected male will have all carrier daughters.
3. There is no male-to-male inheritance.
4. Affected males are related by carrier females.
5. Females may report varying milder symptoms of the condition.
6. A new sporadic case could be due to a new mutation.

X-Linked Dominant

X-linked dominant conditions also exist but these conditions are very rare. If a male is affected, the condition is severe and often lethal. A family history of multiple male miscarriages may be a sign of an X-linked dominant condition.

Variability in Classic Mendelian Patterns of Inheritance

Along with understanding the classic Mendelian inheritance patterns, several other concepts are also important for families to understand when the nurse is assisting clients with or at risk for inheriting a genetic disorder. These include the following exceptions or variations to the traditional Mendelian patterns of inheritance.

Penetrance

Penetrance is the probability that a gene will be expressed phenotypically. It is an "all or none" concept in that, either the gene will be expressed (even if mildly expressed) or it will not be expressed at all. Penetrance can be measured in the following way. In a certain group of individuals with the same genotype, what percentage of them will exhibit at least some signs and/or symptoms of the condition? If the number is less than 100%, then that condition is said to show reduced penetrance. For example, the gene alterations that cause achondroplasia exhibit 100% penetrance and all individuals with one copy of the gene alteration will exhibit signs and symptoms of the disease (Nussbaum et al., 2001).

New Mutation

When there is no previous history of a condition including even subtle signs and symptoms of the disease in any other immediate or distant family member, the disease may be caused by a spontaneous new mutation. This case is usually called *de novo* mutation. New mutations of a gene are most frequently seen in autosomal dominant conditions because one copy of an altered gene is all that is necessary to elicit a state of altered health. Autosomal dominant diseases known to have high mutation rates include neurofibromatosis, achondroplasia (dwarfism), and Marfan syndrome. New mutations are also possible in autosomal recessive diseases although rarely expressed because two altered genes are necessary for signs and symptoms to appear. Finally, new mutations are often seen in X-linked recessive disorders, such as hemophilia A, since the male with just one altered gene will demonstrate the disease.

Anticipation

Anticipation is said to occur when successive generations of a family exhibit more severe signs and symptoms of certain diseases and the disease often has an earlier onset. An example is myotonic dystrophy type 1, an autosomal dominant condition characterized by a range of signs and symptoms including myotonia, muscle weakness, cataracts, and cardiac dysrhythmias. The congenital form is severe, causing mental retardation and may be life threatening. Most children with this congenital form of myotonic dystrophy have a mildly affected mother who may not even be aware she has the disease (Nussbaum et al., 2001). The severity of the condition as well as the age of onset is determined by the number of trinucleotide repeats. Trinucleotide repeats are short DNA sequences such as the CTG base sequence of the gene *DMPK* that are repeated to greater than 2000 times, which results in alterations in the protein products produced by the gene and varying signs and symptoms.

Variable Expressivity

Expressivity is used to describe the severity of the **gene expression** of the phenotype. When people with the same genetic makeup (genotype) exhibit signs and/or symptoms with varying degrees of severity the phenotype is described as *variable expression* (Nussbaum et al., 2001).

Variable expression is common in the autosomal dominant condition, neurofibromatosis. Although neurofibromatosis has 100% penetrance, variable expressivity can occur within family members with each family member exhibiting a variety of signs and/or symptoms.

Multifactorial (Polygenic or Complex) Disorders

Many birth defects such as cleft lip and palate, as well as many adult-onset conditions such as cancer, mental illnesses, asthma, diabetes, obesity, heart disease, and Alzheimer's disease, have a multifactorial cause. **Multifactorial conditions** occur as a result of several gene (polygenic) variations, lifestyle, and environmental influences that work together. The polygenic concept is illustrated with the multiple genes involved in an individual's susceptibility for breast cancer. These genes have been identified on chromosomes 6, 11, 13, 14, 15, 17, and 22. Exactly which genes interrelate and how many environmental influences are enough to cause the presentation of many of the common complex diseases or conditions is not known. However, it is believed that by 2010, the major contributing genes for many common complex conditions will be identified (HGP, 2004a).

Multifactorial conditions accumulate in families but these conditions do not follow the characteristic Mendelian pattern of inheritance seen with single-gene conditions. Inheritable recurrence risks vary in multifactorial conditions. With information gathered from a family history, basic occurrence risks can be assessed for an individual. For instance, premature death in a first-degree relative, two affected first-degree relatives, and two

second-degree maternal or paternal relatives with at least one individual having premature onset of the disease are all considered high inheritance risks. Moderate risks include an individual having a first-degree relative with late or unknown disease onset or two second-degree relatives from the same lineage with late or unknown disease onset. An individual having no affected relatives or a negative family history, only one affected second-degree relative from one or both sides of the pedigree is considered average or general population risk (Scheuner et al., 1997).

Recurrence risks refer to whether or not a condition will occur again in subsequent pregnancies. Because a Mendelian pattern is not present, statistical percentages can be used to represent the chance that parents have a condition that will occur in another child. The risk of recurrence is higher when more than one family member is affected. The recurrence risk after the first affected child is 4.0%, whereas the recurrence risk after a second affected child increases to 10%. It is also known that the recurrence risk increases with an increase in severity of the defect.

INTERDISCIPLINARY CARE

Many health professionals work together in screening, diagnosis, identification, and treatment of genetic disorders. The goals of collaborative care are early diagnosis through testing and assessment, and development of an effective treatment plan, combined with psychosocial support to enhance coping and referral to a genetic specialist when needed.

Genetic Testing

Genetic testing may be used for a client's clinical management, for making personal decisions, or for assisting in reproductive choices. A genetic test is very different from other types of clinical tests. Genetic tests involve the analysis of DNA, RNA, chromosomes, and serum levels of specific enzymes or metabolites. Enzymes and metabolites are part of the protein products that genes produce. DNA, RNA, and/or chromosomes are very unique for each individual and the results have personal, social, financial, and legal implications. Some genetic tests are diagnostic, while others are predictive or inform the individual of an increased risk of acquiring a disease or condition. A "positive" genetic test may indicate that the asymptomatic individual will develop a genetic condition, but a prediction of the onset or severity of the condition cannot be made.

Complications also arise in client understanding because a negative test result cannot guarantee the disease or condition might not develop in the future, often because environmental influences cannot be measured or controlled. Also, the genetic test may have only been able to detect the most common gene mutations and not all of the disease-producing gene alterations are known or available for inclusion in clinical testing. Clients may learn they will develop a genetic condition such as Huntington disease, for which there is no treatment. Clients may find out through genetic testing that they are a carrier and they have unknowingly passed the altered disease-producing gene on to their children.

Finally, the implications of genetic test results are far reaching. While confidentiality and autonomy for the client always are foremost for the nurse, the implications to the client's children, grandchildren, siblings, and other extended family members who share a percentage of the same genes can be life altering. This information may be very confusing and very different from how they have perceived health care in the past and how they perceived the implications of a simple "blood test."

TYPES OF GENETIC TESTS The nurse should understand that genetic tests can be classified into two categories: screening and diagnostic. A positive screening genetic test result notifies the client of an increased risk or probability but must always be confirmed by diagnostic testing. Screening genetic tests are most commonly completed in prenatal, newborn, and carrier circumstances. In contrast, a diagnostic test can definitively validate or eliminate a genetic disorder in the symptomatic client and then direct clinical management. Box 8–8 lists some of the positive and negative aspects of genetic testing.

Several categories of genetic tests included as subcategories of screening and diagnostic genetic tests follow:

- *Newborn screening* is carried out on large sections of the newborn population and provides a means to identify children who have an increased risk for developing a genetic disease such as phenylketonuria, sickle cell disease, or maple syrup urine disease.
- *Carrier testing* is completed on asymptomatic individuals who may be carriers of one copy of a gene alteration that can be transmitted to future children in an autosomal recessive or X-linked pattern of inheritance. This may be part of a couple's premarriage or preconception planning if they belong to a particular ethnic group with known incidence to genetic disorders such as sickle cell anemia and Tay-Sachs disease. It may be necessary to determine the exact gene mutation

BOX 8–8 Positive and Negative Outcomes Related to Genetic Testing

Benefits of Genetic Testing

Provide for:

- Early screening and preventive measures
- Future planning and life preparation
- Lifestyle adaptations
- Decreased confusion and anxiety
- Psychologic stress relief
- Reproductive choices
- Informed extended family members
- Early medical and/or surgical intervention
- Cost of medical follow-up reduced (if negative result)

Possible Negative Outcomes of Genetic Testing

- Survivor guilt
- Loss of identity
- No treatment may exist
- Employability and insurability affected
- Confusion about accessing health care and resources
- Risk for invasion of confidentiality and privacy
- Social stigmatization

Source: Data from Secretary's Advisory Committee on Genetic Testing (SACGT), National Institutes of Health, 2000.

from an affected family member prior to carrier testing. This is often completed through lineage analysis.

- *Preimplantation genetic diagnosis (PGD)* involves the detection of disease-causing gene alterations in human embryos just after *in vitro* fertilization and before implantation in the uterus, thus providing an opportunity for preselection of unaffected embryos for implantation. This type of genetic testing is most often used by parents who are both carriers of a single-gene recessive disorder and who wish to implant into the uterus only the embryo(s) without the disease-causing gene alteration. PGD is usually not covered by insurance, is very costly, and is available at only a small number of centers and for only a small number of disorders (GeneTests, 2004). More recently it has also been used to determine tissue type for donation of tissue such as bone marrow to a sibling or parent (Javitt et al., 2004).
- *Predictive genetic testing* is usually made available to the asymptomatic individual and includes both predispositional and presymptomatic testing. A positive predispositional testing result will indicate there is an increased risk that the individual might eventually develop the disease. Common examples include breast cancer and hereditary nonpolyposis colorectal cancer. A presymptomatic test is performed when development of the disease is certain if the gene alteration is present. These tests are medically indicated when the seriousness and mortality of the disease can be reduced with knowledge of the gene alteration. An example of this would be hereditary hemochromatosis or familial hypercholesterolemia. Life planning and lifestyle choices can be influenced by predictive testing.
- Other uses of genetic testing include organ transplantation tissue typing and pharmacogenetic testing, which involves predicting or studying the client's response to particular medications (Javitt et al., 2004). For example, pharmacogenetic testing has shown that individuals who have Alzheimer's disease and carry two copies of a particular altered gene do not respond well to a drug frequently used in the treatment of Alzheimer's disease. However, if the individual only has one copy of the altered gene, the drug is effective in slowing the progression of the disease (Secretary's Advisory Committee on Genetic Testing, 2000a).

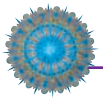
DIAGNOSING CHROMOSOMAL ALTERATIONS Microscopic examination of chromosomes through a karyotype can reveal chromosomal alterations such as chromosomal additions, deletions, gross breaks, and rearrangements or rejoinings (translocations) (USDOE Genome Programs, 2003). Among other things, these chromosomal alterations are responsible for many forms of cancer and, more importantly, particular tumor types. Chromosomal diagnostic examination can be accomplished with a simple blood sample and skin or buccal cell sampling. A karyotype is completed in a cytogenetics laboratory. Chromosomes can be identified by their size and unique light and dark banding patterns. The pairs of autosomal chromosomes are arranged from 1 to 22 according to each chromosome's size, unique banding patterns, and centromere position. The sex chromosomes complete the picture, with the X chromosome(s) first, then the Y chromosome (if present). The karyotype shows all of the chromosome pairs lined up and positioned on a piece of paper allowing for visual chromosomal analysis. (See Figure 8–2 earlier in this chapter.) The final report contains numerical data that includes the total number of chromosomes present. If there is an additional or deleted chromosome, it is identified with a plus (+) or minus (−) symbol. For example, the male individual with 47, XY, +18 has 47 chromosomes (instead of the expected number of 46) that include an additional chromosome 18.

DIAGNOSING GENE ALTERATIONS With the rapidly expanding advances in technology and the identification of genes in the human genome, the availability of genetic, DNA (gene)-based tests has grown tremendously. Currently, more than 900 genetic tests are available with more becoming available each day (USDOE Genome Programs, 2003). **DNA-based tests** involve new, sophisticated technology that permits the examination of the DNA itself. Genetic testing that is DNA based can be obtained from blood, bone marrow, amniotic fluid, fibroblast cells of the skin, or buccal cells from the mouth. Genetic testing includes different types of DNA-based tests. The appropriate genetic tests may be testing for a specific mutation. This would be used if a family member was known to have a genetic condition and could therefore be tested for that particular gene alteration. Another way to examine DNA is by running a panel of mutations. This is done when there are a specific number of identified genes that the majority of individuals with a genetic condition have, for instance, a panel of the three mutations on the *BRCA1* gene that are common in the Ashkenazi Jewish population. A third type of DNA-based test is a complete gene sequence (GeneTests, 2004).

QUALITY AND ACCURACY OF GENETIC TESTS Genetic nurses have expressed concerns that genetic tests are becoming available too quickly with no regulation of the companies that are offering genetic tests. The quality, accuracy, and reliability of genetic test results are not measured against any common standard. Individuals often make hard and irrevocable life-altering decisions after receiving test results so accuracy and reliability are essential. Also, in most cases, minimal or no education is provided for the individual undergoing testing, nor is there any quality counseling or follow-up after the results are given to the individual. Genetic tests are often offered by laboratories before the tests have been proven safe, effective, and practical. Because the majority of genetic conditions are rare, there is often only one laboratory offering the genetic test that is needed. Recently, concerns have been voiced about "direct to consumer" genetic testing. Genetic tests are being offered at "walk-in" locations and also via the Internet. Individuals can receive results of genetic tests in private without a physician's order and fear of discrimination, but also without education or knowledge of the implications of the test results.

Concerns also exist related to test validity, test sensitivity and specificity, the quality of the laboratory performing the test, and the competence of the client's healthcare provider to interpret the test results. **Test sensitivity** refers to how specifically the test identifies (positive test result) individuals who are affected and/or who have the disease phenotype. A test with a high degree of sensitivity has very few false negatives and many true positives. **Test specificity** refers to how specifically

the test does not identify (negative test result) individuals who are unaffected or do not have the disease phenotype. A test with a high degree of specificity has very few false positives (SACGT, 2000). The laboratory selected for the genetic test should have a CLIA88 (Clinical Laboratories Improvement Amendments of 1988) certification (Javitt et al., 2004).



NURSING CARE

The Role of the Nurse in Genetic Testing

With knowledge of available genetic tests and the many implications related to genetic testing, the nurse can assist clients as they weigh choices regarding genetic testing. As consumers of multimedia, clients often have unreliable sources for information related to genetic testing. They may form many misconceptions about the types of genetic tests available and what information these different types of genetic tests are able and not able to provide. When managing genetic information about genetic testing, the nurse must include education for the individual and the family. Communication with the client about genetic testing should include an assessment of the positive and negative outcomes of the test. Are there existing treatments for the condition being tested? Psychologic issues should also be emphasized. Who will be affected by the test results? Will the test results be shared with extended family members?

The nurse is responsible for alerting clients of their right to make an informed decision prior to any genetic testing with consideration of the special circumstances arising from the family, culture, and community life. All genetic testing should be voluntary and it is the nurse's responsibility to ensure that the consent process includes discussion of the risks and benefits of the test, including any physical harm as well as potential psychologic and societal injury by stigmatization, discrimination, and emotional stress (Beskow et al., 2001; International Society of Nurses in Genetics, 2000).

Above all, nurses have a responsibility to fully educate clients about the multiple issues related to genetic testing. Clients should engage in genetic testing with full knowledge, confidentiality, and autonomously. Informed consent may be given verbally, although some laboratories require written consent. Prior to the testing, the client should have an idea of the probability for a positive or negative result if one can be determined by the client's or family history (GeneTests, 2004). To deliver the expected standard of care, it is imperative that the nurse include these issues when developing an educational plan for the client anticipating the use of genetic testing and as part of the informed consent process.

Ensuring Confidentiality and Privacy for Genetic Testing

Although confidentiality and privacy are integral parts of delivery of care for all nurses, this issue is of even more concern as it relates to genetic information. Results of genetic tests can be far reaching and can affect employment and insurance options. Will the results affect the client's ability to obtain and/or maintain insurance coverage? Can an employer refuse to hire or promote an individual because of genetic testing results? Can genetic information be released to the courts, military, schools, or adoption agencies? Would a client with a known gene alteration for Huntington disease be offered a college scholarship to the best law school? There is debate over whether genetic privacy is different from medical privacy. The nurse should inform clients of their rights and responsibility to know who will have access to the genetic test results. Those providing the genetic tests must provide the client with assurance that the results will be handled confidentially, and that there will be no access to the genetic information by a third party without written permission of the individual being tested.

Results of genetic tests should only be communicated directly to the individual who gave the consent. No outside governmental, employment, or insurance organizations should ever have access to genetic test results without the written permission of clients. Clients should confirm how they will receive the test results. Clients should ask who will have access to the test results and what will happen to the DNA sample after the test is completed. In the majority of cases, results of genetic tests should not be shared with extended family members without written permission. Healthcare providers are legally liable to maintain that confidence. Exceptions to the individual's privacy may be made only when the individual refuses to inform extended family members when a very high probability of irreversible harm exists for the extended family member and informing the family member can prevent the harm (National Human Genome Research Institute, 2005). Every effort should be made to educate the individual about the benefits of informing extended family members if applicable. Genetic testing should ideally be accompanied by pretest and post-test counseling by genetic specialists or by another knowledgeable healthcare provider.

Psychosocial Issues

Although family and individual anxiety may be decreased with a negative test result, potential problems do exist and the nurse must be prepared to address them. Concerns about carrier status may interfere with development of intimacy and interpersonal relationships. Nonpaternity may be revealed through genetic testing. For example, the parents of a child born with an autosomal recessive condition will be considered carriers of the altered gene the majority of the time. To counsel the parents about future pregnancies, the parents would be tested to confirm their genotype, and nonpaternity may become an issue. A positive test result may lead to feelings of unworthiness, confusion, anger, depression, and self-image disturbance. Survivor guilt may affect adults with negative results if their siblings are positive. The individual carrying a gene alteration for a late-onset disease may have an increased tendency for risky behaviors and may choose not to be a positive member of society. Relatives of an individual affected with a genetic dis-

order may be very frightened when they realize what their own future might be (SACGT, 2000a). The individual who has inherited an altered disease-producing gene may foster deep resentment toward the parent who carries the altered gene. Parents and older generations may feel tremendous guilt for passing the altered gene to their children and grandchildren (Wertz et al., 1994).

Economic Issues

The nurse should consider the cost of genetic tests, which can range from hundreds to thousands of dollars, depending on the size of the gene being tested. Most insurance companies do not cover genetic tests but if there is insurance coverage, the individual must weigh the cost of allowing the insurance company to have access to the genetic information (HGP, 2005).

Genetic tests differ from routine medical tests in many ways. The risks and benefits of genetic testing are numerous and complicated. Nurses have an obligation to maintain their knowledge regarding genetic testing to advocate for the client as well as maintain ethical standards of care. Above all, nurses must be able to recognize the limits of their expertise and how to refer a client to genetic specialists and additional resources.

Assessment

Health Promotion and Health Maintenance

Health promotion and health maintenance of the client are viewed as the foundations of all nursing care. However, most individuals do not know their complete genetic makeup. Some know they carry an altered gene that causes a specific disease, but the majority of individuals do not know with certainty what their future health status will be. With no sure knowledge of genetic makeup or whether a certain alteration in health status will occur (heart disease, for example), healthy lifestyles are not always a priority. Imagine, then, if people knew their statistical risks for developing or inheriting disease by having complete access to the types of genes in their cells? Health promotion and health maintenance teaching and nursing interventions would be based on specific genes. The nurse could provide important, life-saving nutritional information to their clients based on their specific risks, and clients then might be more inclined to maintain a proper diet, give up their sedentary lifestyle, increase exercise, and decrease fast-food intake. Personal lifestyle choices would become more personal and monitoring health might take on a new meaning.

With knowledge of genetic conditions, the nurse can ensure health teaching and early detection of complications from genetic conditions with emphasis on primary and secondary care interventions. For example:

- A woman with a strong family history and/or mutations in the *BRCA1* and *BRCA2* tumor suppressor genes should begin monthly self breast exams, and have screening clinical breast exams and mammographies at an earlier age than the general population.
- A man with a strong family history and/or mutations in the *BRCA1* and *BRCA2* tumor suppressor genes should report any mass, tenderness, or swelling in the breast tissue and maintain early screening for prostate cancer.
- Aggressive colonoscopy screening every 1 to 2 years beginning at age 25 is important for the individual with a positive family history and/or mutations in the *MLH1/MSH2* gene, which increases the risk for hereditary nonpolyposis colorectal cancer.

Clients receiving early intervention and health promotion-focused care can live longer and with a much better quality of life that those who do not. The nurse must be able to identify both community-based and genetic-based resources that are available to assist the client in strategies to support both health promotion and health maintenance activities.

By simply integrating into practice the genetic aspects of assessment, observation, and history gathering, the nurse can improve the standard of care delivered and have a very positive impact on the client. The nurse does not need to be a genetic expert, but with heightened awareness, appropriate inquiries and referrals to genetic specialists can be completed.

Client Intake and History

Nurses can improve the standard of nursing care and have a positive impact on clients by integrating genetic concepts into their existing practice of inspection, observation, and history gathering. Nurses do not need to be genetic experts, but they should be able to recognize genetic features of physical assessment, basic patterns of inheritance, and predisposition to development of disease. As nurses integrate genetic concepts into their delivery of care, appropriate inquiries and referrals to genetic specialists can be completed.

Although family history has long been a part of nursing assessment, the relative importance of this assessment piece has recently increased as our knowledge of the interaction of genes and the environment has expanded. In phenotypically "healthy" individuals, an accurate and complete family history can identify a single-gene (Mendelian) disorder or a mitochondrial, multifactorial, or chromosomal inheritance pattern as well as guide the prevention, diagnosis, and treatment of common complex diseases such as cardiovascular disease and cancer. A family history illustrates the interaction of genes and the environment for an individual and consequently provides a basis for individualized disease prevention (Guttmacher et al., 2004). Although an individual's inheritance risks from his or her genotype are nonmodifiable, knowledge of an individual's increased risk for chronic disease can influence lifestyle choices, clinical management, and sometimes risk reduction and prevention of the disease. Knowledge of a family history can also guide diagnostic workups and clinical treatment (Guttmacher et al., 2004).

Pedigrees

A nurse should know how to take a family history, record the history in a pedigree, and think "genetically." A pedigree is a pictorial representation or diagram of the medical history of a family. Multiple symbols are utilized to present this picture (Figure 8–6 ■) and the finished pedigree presents a family's medical data and biologic relationship information at a glance

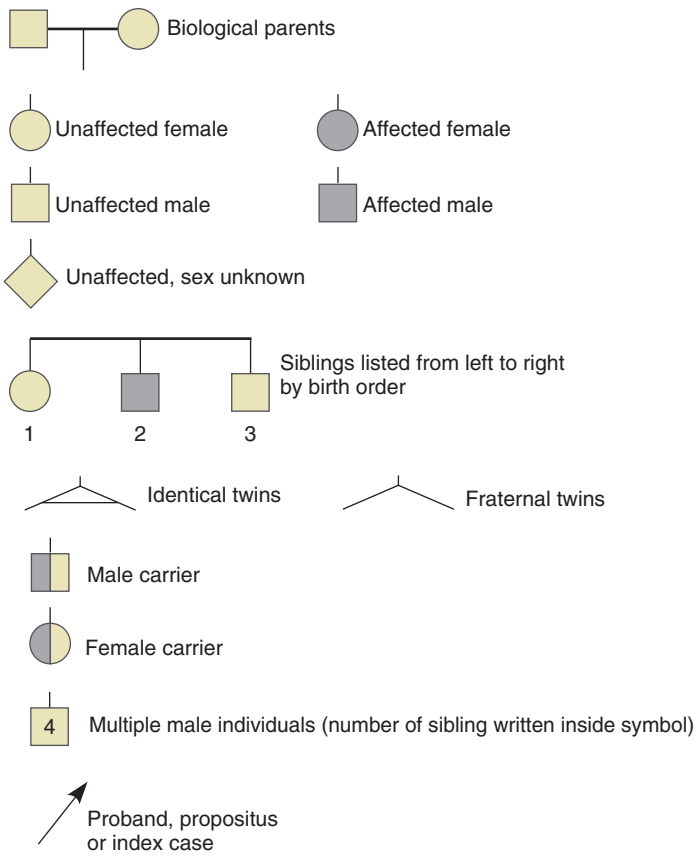


Figure 8–6 ■ Selected standardized symbols for use in drawing a pedigree.

(Figure 8–7 ■). A pedigree provides the nurse, genetic counselor, or geneticist with a clear, visual representation of relationships of affected individuals to the immediate and extended family. It can identify other individuals in the family who might benefit from a genetic consultation. It also can identify a single-gene alteration pattern of inheritance or a cluster of multifactorial conditions, and a referral and/or reproductive risk teaching for the individual and family can result. A family's learning can be enhanced by the visual teaching contribution a pedigree can provide and also clarify any inheritance misunderstandings or misconceptions. Box 8–9 lists steps for creating a family pedigree. If completed correctly and completely, a pedigree allows all healthcare professionals working with the client or family to quickly see what history and background information has been collected (Box 8–10).

It is important to gather a three-generation family pedigree even if the nurse believes this is a first occasion of the condition within a family (Figure 8-7). A condition without any identifiable inheritance pattern on the pedigree may be due to a new mutation or variable expressivity. Throughout the process of gathering family history assessment data, the nurse must remember family confidentiality at all times: All information related to a pedigree is confidential information. The history may reveal sensitive details that include infertility problems, elective termination of pregnancies, or nonpaternity. This information may not even be known by a current partner or immediate and extended family members. Other sensitive issues include pregnancies conceived by technology, a history of suicides, drug or alcohol abuse, and same-sex relationships. Box 8–11 lists ethical implications of genetic information.

Challenges inherent in recalling the family history include the client's inability to remember any conditions that may have been surgically repaired and then forgotten, or reporting conditions that may have been attributed incorrectly to other causes. Also, the family history may contain information previously unknown to extended family members. Reproductive decisions may have been made that were against the family's religious or cultural beliefs. Both immediate and extended family members may be unaware of these "family skeletons" and the client may be very reluctant to reveal this information (Bennett, 1999; Bowers, 2002).

Genetic Physical Assessment

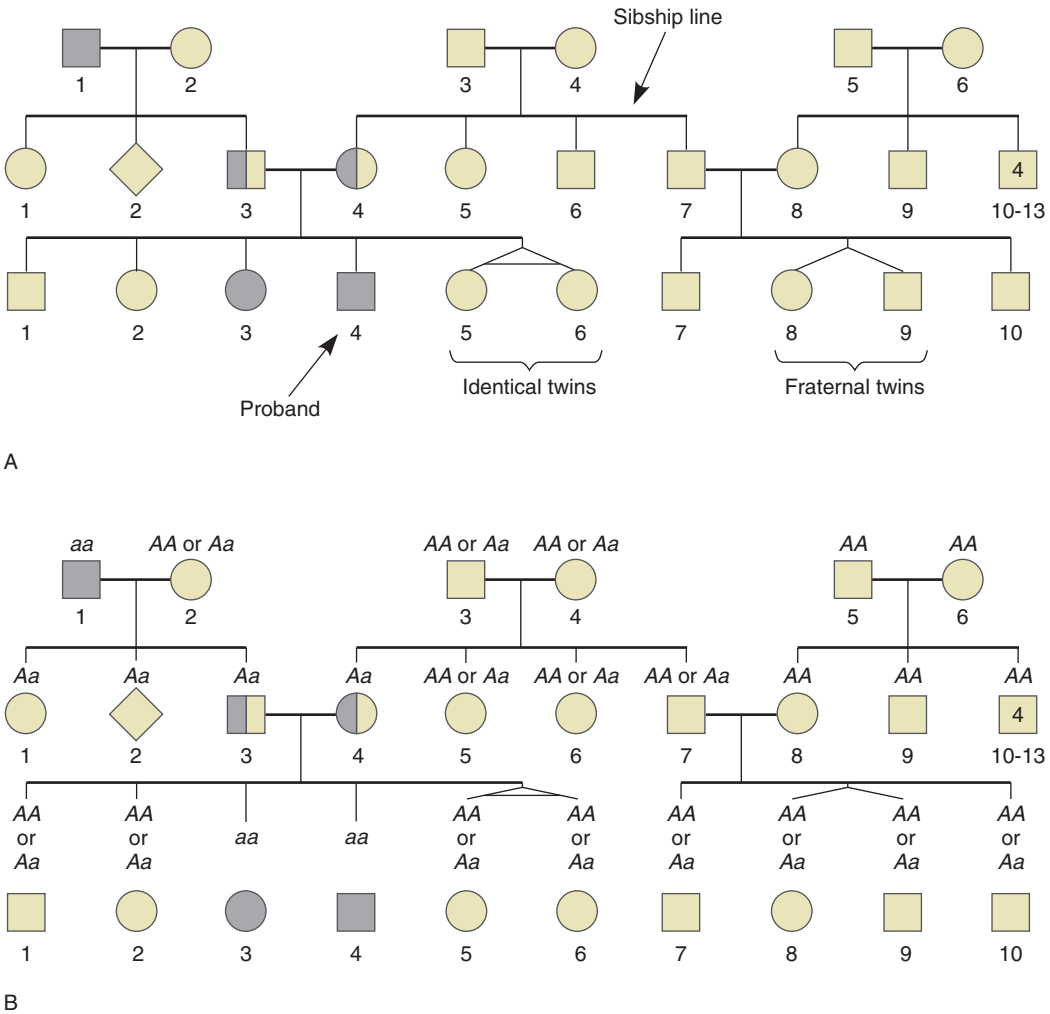
An assessment of newborns and children with a genetics focus of minor and major anomalies is essential. A minor anomaly or malformation is an unusual or morphologic feature that in itself is of no serious medical or cosmetic concern to the individual or family. A major anomaly is a serious structural defect at birth that may interfere with normal functioning of body systems and may lead to a lifelong disability or even an early death (Aase, 1992).

Some children grow into adults whose physical features may or may not have been recognized as potential links to genetic conditions. Anomalies that exist after a person is beyond expected developmental and cognitive milestones may go unnoticed, but they may still be important. The nurse can pick up cues to genetic problems by inspecting the client and other family members. If practical, nurses should ask to look at family photographs and examine them for common dysmorphic features and family traits. Subjective assessment data can also be valuable. A client's complaints of fatigue and joint pain may indicate the onset of hereditary hemochromatosis. A physical assessment that includes a genetics focus is important for nurses when caring for any age client. An undiagnosed genetic condition may also have implications for reproductive decisions the client makes. However, genetic assessments and genetic referrals are important throughout the life span.

Nursing Diagnosis and Interventions

Nurses are responsible for comprehensively delivering the correct standard of care to clients, but at the same time being aware of the limitations of their own knowledge and expertise. In addition to the continuous integration of genetic aspects into nurses' assessments of family history and physical assessment, nurses are also responsible for carrying out interventions that include initiating referrals to genetic specialists and delivering care to the individual or family in any of the following ways. Nursing diagnoses to consider include:

- *Anticipatory Grieving*
- *Anxiety*
- *Disturbed Body Image*
- *Ineffective Coping*
- *Decisional Conflict*



(A) A representative pedigree for a single character or genetic condition through three generations.
(B) The most probable genotypes of each individual in the pedigree for an autosomal recessive condition, represented by AA, Aa, or aa.

Figure 8–7 ■ Sample three-generation pedigree.

Source: Redrawn from Klug, W. S., & Cummings, M. R. (2003). *Concepts of genetics* (7th ed., p. 63). Upper Saddle River, NJ: Prentice Hall.

- *Interrupted Family Processes*
- *Ineffective Health Maintenance*
- *Deficient Knowledge*
- *Powerlessness*
- *Spiritual Distress*

Genetic Referrals and Counseling

After gathering assessment data that incorporates genetic concepts, the nurse is able to initiate a referral to genetic specialists if there are indicators for a genetic referral (Box 8–12). The nurse should provide the client with information about the advantages of a referral to genetic specialists, and the disadvantages of not following through with the referral. The nurse should inform the client that a genetic referral can provide information and answer many questions they may have concerning genetic health. Questions regarding the conditions, inheritance, availability of treatment, as well as economic, insurance, and future implications can be addressed.

Clients who are concerned about genetic disease may benefit from a genetic consultation whether or not genetic testing is available for that condition. Many people seek information and coping strategies as much as they do test results. Referral of a client with a suspected genetic problem to a geneticist, genetic clinical nurse specialist, or genetic clinic is an expected nursing responsibility in the same way as referrals to a dietitian or a social worker. When in doubt, the nurse should contact the advanced practice genetic clinical nurse, genetic counselor, or geneticist to discuss concerns.

CLIENT PREPARATION FOR GENETIC REFERRALS AND GENETIC COUNSELING Not knowing what to expect from a genetic referral is common, and the fear of the unknown may cause anxiety for the client. To facilitate the referral to a genetic specialist, the nurse should educate the client so that he or she knows what to expect during as well as after a genetic evaluation.

BOX 8–9 **Steps in Drawing a Pedigree**

I. How to:
  1. Work in pencil.
    a. Family historians often remember additional relatives and details only after questioning is almost completed.

II. Organization:
  1. Begin recording data in the middle of the sheet of paper.
    a. Allow enough room for both the maternal and paternal sides of the family.
  2. Use only standard pedigree symbols.
    a. For example, males are represented by squares and females by a circle.
  3. The male individual in a couple is placed on the left of the relationship line and the paternal side of the family also goes on the left-hand side of the paper.

III. Determining Family Relationships:
  1. The nurse should determine the relationships within the family by asking questions such as:
    - Do you have a partner or are you married?
    - How many biologic brothers and sisters do you have?
    - How many children do you have?
    - Do all the children have the same biologic father?
    - Do all the children share the same mother and father?
  2. Referral to "the baby's father or mother" can be helpful until a relationship or marriage is established between the parents.
  3. Referral to a "union" if marriage does not exist can also help communication.
  4. Determine if each individual is married, has children, signs and symptoms, etc., before continuing on to the next individual.
  5. Always ask if there is any chance the mother could be related to the father or if any other parents in the family are blood relatives.
    - To determine consanguinity.

IV. Who Should or Should Not Be Included:
  1. To ensure accuracy, the pedigree should include the parents, offspring, siblings, aunts, uncles, grandparents, and first cousins of the individual seeking counseling.
  2. Detailed information about the spouses of the proband's family can be omitted unless there is a history of some kind of disorder or condition.
  3. Eliminating persons or any information that does not contribute any valuable information can help keep the pedigree small and more manageable.

V. Recording the Family History:
  1. Determine the approximate size of the family.
  2. Record the family's ethnic background at the top of the page.
  3. The initial drawing should begin with the proband, or the person who is affected with the genetic condition.
    a. Usually the reason someone is seeking a genetic referral.
  4. The proband is marked with an arrow on the pedigree.
  5. Draw and mark the symbols for the brothers and sister of the proband. Draw the relationship line, the line of descent, marriage or union line, and symbols for parents of the proband.
    a. Repeat this step for any children of the proband or children of the proband's brothers and sisters.
  6. Children resulting from a mating (siblings) should be recorded in descending order of their birth with oldest sibling on the left.
  7. Continue with symbols for all immediate relatives drawn previously and then draw and mark symbols for paternal grandparents and indicated relatives followed by the same for the maternal grandparents and relatives.
  8. A legend key should contain all of the correct symbols for each indicated disease.
  9. Record the age of onset of common and complex diseases and/or conditions such as coronary heart disease; diabetes mellitus; hypertension; colon, breast, ovarian, or endometrial cancer; and stroke.
  10. The pedigree should include at least three generations.
    a. Generations are symbolized by Roman numerals along the left-hand side of the paper with the first generation marker, I, at the top.
    b. Each person in the generation should follow an imaginary horizontal line from left to right.
  11. The names of each individual (maiden names in case of married women) and their dates of birth should be included along with half-siblings, pregnancy losses, still births, previous marriages, and adopted children.

VI. Other:
  1. Consanguinity may be suspected if the historian repeatedly gives the same last name on both sides of the family.
    a. Consanguinity can be confirmed by asking if any relatives in the family have ever had a child together.

VII. Completing the Pedigree:
  1. When completed, the pedigree should be dated and signed with the name, credentials, and position of the person drawing it.

Source: Data from Bennett, R. L. (1999). *The practical guide to the genetic family history*. New York: Wiley-Liss.

MEDIALINK Create and Analyze a Family Pedigree

Usually before the first genetic evaluation visit, the client will be contacted to provide a detailed medical and family history and to make an appointment for genetic consultation. The client should be prepared to give as exact a family history as possible so that a detailed three-generation pedigree can be constructed. The client should be informed that a genetic consultation usually lasts several hours. During the appointment, a genetic clinical nurse, genetic counselor, and/or a physician will perform an initial interview with the client. A geneticist will examine the client in order to establish an accurate diagnosis. Tests may be ordered. These may include chromosome (cytogenetic) analysis, DNA-based testing, x-rays, biopsy, biochemical tests, and linkage studies (Lashley, 2005). After the exam and the completion of any applicable testing, the

BOX 8–10 Specific Facts and Health Information to Include in a Pedigree

- Age/birth date or year of birth
- Age of death (year, if known)
- Cause of death
- Age at diagnosis
- Full siblings versus half or step-siblings
- Pregnancy with gestational age (LMP) or estimated date of delivery (EDD)
- Infertility versus no children by choice
- Pregnancy complications with gestational ages noted (e.g., 6 wks, 32 wks)
- Miscarriage (SAB)
- Stillbirth (SB)
- Pregnancy termination (TOP)
- Relevant health information (e.g., height and weight)
- Affected/unaffected status—define shading of symbols in a legend key
- Ethnic background
- Consanguinity
- Date pedigree taken or updated
- Name of person who took pedigree and credentials
- Key or legend

geneticist and/or genetic counselor will discuss the findings with the client and make recommendations. The discussion will include the natural history of the condition, the inheritance patterns, the current preventive or treatment options, and the risks to the client and/or family. The visit will also include opportunities for questions and answers as well as the assessment and evaluation of the client's understanding. It is typical for the information retention of a client facing a new genetic diagnosis to be very low. This makes it imperative for the nurse to take advantage of opportunities to reinforce genetic concepts at a later time when the client is ready.

As the visit concludes, the client can expect appropriate referrals to be made, discussion of available services or research studies, and possible scheduling of a follow-up visit. A summary of the information is usually sent to the client and the client's healthcare provider will receive a report if requested by the client.

Genetic healthcare providers present the client with information to promote informed decisions. They are also sensitive to the importance of protecting the individual's autonomy. A challenge during any visit to a genetic specialist is providing nondirective counseling. Clients should be permitted to make decisions that are not influenced by any biases or values from the nurse, counselor, or geneticist. Many clients are accustomed to practitioners and nurses providing direction and guidance in their decision making, and clients may be very uncomfortable with the nondirectional approach of the nurse. They may believe that the nurse or healthcare provider is withholding very bad news. The nurse should discuss the positives and negatives of each decision and present as many options as possible through the use of therapeutic listening and communication skills (Cunniff, 2001).

Client Teaching

The nurse must be aware of available genetic resources and participate in the education of genetic disorders as well as health promotion and prevention. Informing clients of what to expect from a genetic referral as well as clarifying and/or reinforcing information obtained during a genetic referral or from genetic test results is also important.

The cultural and religious beliefs and values of the client must be assessed by the nurse prior to teaching. Are the gene alterations viewed as uncontrollable and believed to be occurring secondary to cultural belief such as a stranger looking at the client? Or, are the gene alterations considered a "punishment"? A client's readiness to learn can be influenced by cultural or religious beliefs and values. Obtaining educational materials in the native language of the client will also help facilitate the teaching-learning experience.

The nurse must be aware of common inheritance misconceptions such as a client's belief that with a 25% recurrence risk, after one child is affected, the next three children will be unaffected or with a 50% recurrence risk, every other child will be affected. The recurrence risk *for each pregnancy* should be continually stressed by the nurse. Clients often believe that a family member has inherited the genetic condition because they look or "take after" a relative with a genetic condition. When new gene alterations or mutations are discussed, clients will often exhibit surprise because no one else in the family has the condition so they perceive that the trait or condition cannot possibly be inherited (Bennett, 1999). Helping clients to understand these genetic concepts is fundamental to delivering the standard of genetic nursing care.

Psychosocial Care

To meet the client's psychosocial needs, the nurse should identify the client's expectations and needs as well as the client's cultural, spiritual, value, and belief system. From where does the client receive strength? Denial of the genetic diagnosis is common and nurses must be aware of the client's state of acceptance. Individuals often will not believe that a chronic genetic condition exists. Nurses must also provide care to help alleviate any anxiety and/or guilt in the client. Anxiety of the unknown is common when awaiting diagnosis or test results, but individuals also experience anxiety from not understanding the future implications of a confirmed genetic disease. Guilt may be associated with knowledge of the existence of a genetic condition in a family. The nurse must support clients as they contemplate telling extended family members, friends, and neighbors about a confirmed diagnosis. Clients often do not want to tell extended family members until they are ready. The nurse should encourage open discussions and the expression of fears and concerns. Guilt and shame are very common as a client deals with the loss of the expectation and dream of a healthy, productive life.

BOX 8–11 Ethical Implications of Genetic Information

The nurse must consider the enormity of the ethical issues facing all families who have knowledge of their genetic makeup. The ethical issues a nurse may have to discuss with the client are numerous. A few of the issues are listed below.

Access to Information

- Who should have access to personal genetic information, and how will it be used?
- Do insurers, employers, courts, law enforcement, schools, universities, adoption agencies, and the military have a right to access this information?

Self-Perception

- How does personal genetic information affect an individual's perception of self?
- How does personal genetic information affect society's perceptions of that individual?
- How does personal genetic information affect an individual's cultural identity?
- How is self-identity and self-worth affected by a confirmed genetic risk or condition?

Family Roles and Relationships

- Should an individual be tested for an autosomal dominant condition if the siblings and/or parents are opposed to knowing if they, themselves, have the altered gene?
- Should potential mates have genetic information?
- Should two people with increased genetic risk be prohibited from having children?
- Should a child be tested?
- Should the father be told if genetic testing and/or genetic counseling reveals nonpaternity?
- Should adoption records contain a complete genetic history of the biologic parents?
- Is there an obligation to tell other family members if an altered gene that demands a change in lifestyle (nutrition, exercise, smoking, etc.) is diagnosed?
- Is there an obligation to tell other family members if an altered gene that causes early debilitation and/or death is diagnosed?

Informed Consent

- Are all individuals receiving true informed consent and do they understand all of the consequences of agreeing to even a simple blood test in the doctor's office that may reveal a diagnosis or increased risk for a genetic condition?

Health and Life Insurance

- Should insurance companies have access to genetic test results?
- Should medical insurance costs be higher for persons with a known gene disease-producing alteration?
- Should medical insurance costs be higher for persons with a known increased risk for disease because of any gene alteration?
- Should medical insurance costs be higher for persons with known increased risk for disease because of any gene alteration if they make unhealthy lifestyle choices and do nothing to lower their risk?
- Should the individual be covered by medical insurance at all?
- Should individuals pay higher costs if they have children?
- Should individuals be required to have a large life insurance policy to financially protect their families?

Financial

- Should the child be eligible for government grants or any scholarship money?
- Should society be expected to financially support children through government programs or private insurance?
- What is the motivation to save money for the future?

Employment

- Should an employer have access to an individual's genetic profile?
- Should a young adult be hired even though she/he will burden the company with multiple sick days, higher insurance financial support, etc.?
- Should the individual receive promotional opportunities and increased job responsibilities if the employer knows there will be a great deal of lost work days?
- Will the individual's productivity be affected by the genetic condition?

Reinforce to clients that genetic alterations are caused by changes within a gene and not by superstitions related to sin or other cultural beliefs. However, it is important to remember that everyone has superstitions or beliefs and the nurse must remain nonjudgmental. As mothers, fathers, siblings, and extended family members provide continuous care for the client with a genetic condition, depression can result. Depression also can occur in the individual with the chronic condition. The nurse must maintain awareness of the possibility of depression and be proactive in obtaining support for the client or family.

The nurse also is responsible for assessing the client's coping mechanisms as well as available family, spiritual, cultural, and community support systems. Genetic conditions can cause a permanent strain on family dynamics and relationships. The nurse may need to help the client reaffirm his or her self-worth and value (Lashley, 2005). If seen in an academic setting, clients may feel they are part of a "production line" even though they are present for a very private problem (Cunniff, 2001). Nurses must be sensitive to these perceptions, provide open communication, and encourage discussion of feelings. Growth and development and meeting adult developmental milestones can be altered by actual or potential genetic disorders. Especially unique is the potential or actual inheritance of a late-onset condition such as Huntington disease. The client with this altered gene may not meet any of the developmental tasks in moving through adulthood. Should the client get married, attend college, save money, or worry about future? The nurse must identify the impact of genetic knowledge on activities of daily living but also movement through developmental milestones. Both client and family strengths need to be identified.

BOX 8–12 Adult Indicators for a Referral to a Genetic Specialist

Adult History Assessment Data Concerns

- Several closely related individuals affected with the same or related conditions:
  - Breast and ovarian cancer
  - Colon and endometrial cancer
  - Diabetes
  - Hypertension
  - Coronary heart disease
  - Thyroid cancer
  - Colon polyps
- A common disorder with earlier age of onset than typical (increase concern if it occurs in more than one family member):
  - Breast cancer: < 45–50 years of age or premenopausal
  - Colon cancer: < 45–50 years of age
  - Prostate cancer: < 45–60 years of age
  - Vision loss: < 55 years of age
  - Hearing loss: < 50–60 years of age
  - Dementia: < 60 years of age
  - Heart disease: < 40–60 years of age
  - Stroke: < 60 years of age
- A sudden or unexpected death in someone who "seemed" healthy
- Renal disease
- Asthma
- Suicides

An Individual with:

- Two or more conditions
- A medical condition and dysmorphic features
- Developmental delay with dysmorphic features and/or physical birth anomalies
- Learning disabilities
- Behavioral problems
- Unexplained:
  - Movement disorders
  - Seizures
  - Hypotonia
  - Ataxia
  - Infertility
- Disproportionate tall or short stature
- Proportionate short stature with dysmorphic features
- Atypical sexual development
- Premature ovarian failure

Source: American Medical Association. (2004). *Family medical history in disease prevention*. Retrieved March 12, 2005, from http://www.ama-assn.org/go/familyhistory

The nurse can refer the client to a support group. However it is important to have permission from the client if the nurse is providing a support group with the client's name and contact information.

Another key role for the nurse is to help clients with the often difficult task of communicating genetic information such as inheritance patterns to extended family members. Cultural values of autonomy and privacy are impacted when a client must consider whether to communicate genetic information to extended family members who may also carry the altered gene. The history of a genetic alteration that may or may not cause disease can be extensive within a family, affecting multiple family members. Family members often have difficulty understanding that some genetic conditions have variable expressivity. Members of the extended family often are shocked and feel a profound sense of guilt that they are the one who has carried the gene alteration that caused their loved one to have a genetic condition.

Careful self-assessment of feelings is essential for the nurse. The nurse must continually advocate for clients and support their decisions even if the decisions contradict the nurse's own ideals and morals. Coping with genetic revelations and making genetic-related treatment decisions are difficult activities for everyone. The nurse must remember that clients will need resources and support, and also help in gathering information about reproductive options.

Evaluation

Expected outcomes of delivering nursing care with a genetic focus include:

- The client will make informed and voluntary decisions related to genetic health issues.
- The client will accurately identify:
  - Basic genetic concepts and simple inheritance risk probabilities
  - What to expect from a genetic referral.
  - The influence of genetic factors in health promotion and health maintenance.
  - Differences between medical and genetic tests.
  - Social, legal and ethical issues related to genetic testing.

VISIONS FOR THE FUTURE

Nurses are often the primary caregivers to whom clients turn for information, guidance, and clarification of ideas. This nursing role is essential not only in providing direct nursing care but as a member of the community. As more information about the genetic revolution becomes available to consumers—in areas such as pharmacogenomics, gene transfer, ethics, genetic engineering, and stem cell research—the role of nurses remains not only vital but grows enormously. Nurses should remain educated, informed, knowledgeable, and ready to discuss trends and changes with clients and their families.

MEDIALINK Case Study: Genetic Resources

EXPLORE MEDIALINK

Prentice Hall Nursing MediaLink DVD-ROM

Audio Glossary
NCLEX-RN® Review

Animation

Human Genome Project

COMPANION WEBSITE www.prenhall.com/lemone

Audio Glossary
NCLEX-RN® Review
Care Plan Activity: Genetic Implications of Adult Health Nursing Care
Case Study: Genetic Implications
MediaLink Application: Create and Analyze a Family Pedigree
Links to Resources

CHAPTER HIGHLIGHTS

- Nurses are responsible for basic genetic knowledge and delivering the expected standard of genetic nursing care.
- When cell division does not occur as expected, chromosomal alterations on the autosomes or sex chromosomes can result.
- Chromosomal alterations can be seen in a human karyotype.
- Protein-directing genes are very important to life and functioning as a human being because proteins are very specialized and perform a variety of functions within the cell.
- Different forms of genes are alleles.
- An individual may be identified as heterozygous or homozygous for a single gene.
- Some gene alterations cause disease and some are protective from disease.
- Mitochondrial gene alterations are inherited from the mother and are primarily involved in high-energy organs such as skeletal muscles, brain, and heart muscle.
- Multifactorial inheritance does not follow Mendelian inheritance patterns.
- Genetic healthcare providers present the individual and their family with information to promote informed decisions.
- Many types of genetic tests are available and they differ from routine medical tests.
- All genetic tests have special considerations related to social, financial, ethical, and legal implications.
- Basic genetic nursing care involves family risk assessment through a detailed family history, drawing a three-generation pedigree, and integrating genetic concepts into a physical assessment.
- Basic genetic nursing involves initiating a referral to a genetic specialist.
- Knowledge of the principles of inheritance allows the nurse to not only offer and reinforce genetic information to clients and their families but also to assist them in managing their care and in making reproductive decisions.
- Genetic concepts can be applied to health promotion and health maintenance.
- The nurse must be aware of the social, ethical, cultural, and spiritual issues related to the delivery of genetic nursing care.

TEST YOURSELF NCLEX-RN® REVIEW

1 The client is discussing the inheritance of an autosomal dominant trait. The client has the condition and the client's wife does not. They have one child without the condition. The nurse would be correct in explaining to the client that he is most likely which genotype?
   1. FF
   2. Ff
   3. ff
   4. X_fY

2 The male client diagnosed with Fabry disease is admitted to the unit. Which statement made by the client would indicate to the nurse that the client understands Mendelian inheritance concepts? "I have the disease because . . ." (Select all that apply.)
   1. my mother had Fabry and my father did not."
   2. my father's mother had Fabry disease."
   3. my grandmother's brother had Fabry disease."
   4. my father has Fabry disease."

3 The nurse is providing information regarding genetic testing to a couple who believe they are carriers of an autosomal recessive gene alteration. Which statement by the nurse is appropriate?
   1. "If both of you are carriers, all of your sons will be affected and all of your daughters will be carriers."
   2. "Chromosomal studies will reveal if you are actually a carrier."
   3. "Newborn screening will reveal if your child is affected."
   4. "During the genetic evaluation, you will be asked to provide at least a three-generation family history."

4 The nurse knows that which assessment data obtained during a family history may suggest a genetic condition or inherited susceptibility to a common disease? (Select all that apply.)
   1. breast cancer at age 33
   2. a sibling who died unexpectedly while playing basketball at age 66
   3. colon polyps in four third-degree relatives
   4. a brother's unexplained infertility

5 When analyzing a family pedigree, the nurse notes the pedigree demonstrates that successive generations contain affected individuals, both males and females are affected, and there is no father-to-offspring inheritance. What is the most likely pattern of inheritance?
   1. autosomal dominant
   2. autosomal recessive
   3. x-linked recessive
   4. multifactorial
   5. mitochondrial

6 When the nurse is developing a teaching plan, which statement is a correct rationale regarding the health promotion and health maintenance benefits from an assessment of family history? (Select all that apply.)
   1. Clinical treatment options can be more focused.
   2. Prophylactic treatments can be started early.
   3. Specific diet, exercise regimen, and genotype can be determined.
   4. Single-gene alteration can be diagnosed.

7 The nurse is recording a family pedigree. Which would be correct to include in drawing the pedigree?
   1. Detailed information is important for all persons recorded on the pedigree.
   2. The maternal side of the family should be placed on the left of the page.
   3. The proband is marked with an arrow and a "P."
   4. Two generations should be recorded and labeled with Roman numerals.

8 The nurse would consider which assessment finding(s) as minor anomalies? (Select all that apply.)
   1. café au lait spots
   2. ear pits
   3. atrial septal defect (ASD)
   4. hypertelorism

9 Which are appropriate concepts for the nurse to include when developing a teaching plan for the client prior to genetic testing? (Select all that apply.)
   1. Predispositional genetic tests are medically indicated when the seriousness and mortality of the disease can be reduced with knowledge of the gene alteration.
   2. To meet quality assurance, laboratories should hold a CLIA88 certification.
   3. A mutation panel contains the most common gene alterations but it may not include all of the disease-causing mutations.
   4. Family members affected by genetic test results have a legal right to the test results.

10 The client asks the nurse if a genetic referral is necessary. Which information would be appropriate for the nurse to provide? Most likely genetic specialists will: (Select all that apply.)
   1. provide direction for important decision making.
   2. complete chromosomal studies.
   3. ask to see photographs of relatives.
   4. provide information about the natural history of the condition.

See Test Yourself answers in Appendix C.

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