





unit 3

Pregnancy





# chapter 10

## Fetal Development and Genetics

### Key TERMS

autosomal dominant inherited disorders  
autosomal recessive inherited disorders  
blastocysts  
chromosomes  
cri du chat syndrome  
embryonic stage  
fertilization  
fetal circulation  
fetal stage  
fragile X syndrome  
genes  
genetic counseling  
genetics  
karyotype  
monosomies  
morula  
multifactorial inherited disorders  
mutation  
placenta  
polyploidy  
preembryonic stage  
trisomies  
trophoblasts  
umbilical cord  
X-linked inherited disorders  
zona pellucida  
zygote

### Learning OBJECTIVES

*After studying the chapter content, the student should be able to accomplish the following:*

1. Define the key terms.
2. Describe the process of fertilization, implantation, and cell differentiation.
3. Explain the functions of the placenta, umbilical cord, and amniotic fluid.
4. Outline normal fetal development from conception through birth.
5. Discuss the education/counseling needs of patients undergoing genetic testing.
6. Delineate the role of the nurse involved in genetic-related activities.



## WOW

*Being a nurse without awe is like food without spice. Nurses only*

*have to witness the miracle of life to find their lost awe.*

Human reproduction is one of the most intimate spheres of an individual's life. For conception to occur, a healthy ovum from the woman has to be released from the ovary; pass into a normal, open fallopian tube; and start being transported downward. Sperm from the male must be deposited into the vagina and be able to swim approximately seven inches to meet the ovum and penetrate its hard cover to begin the wondrous human life. All this activity takes place within a 5-hour time span, during which the sperm reaches the outer portion of the fallopian tube, where **fertilization** takes place (Gilbert & Harmon, 2003).

Nurses caring for the childbearing family need to have a basic understanding of conception and prenatal development to be able to identify problems/variations and to initiate appropriate interventions should any problems occur. This chapter presents an overview of fetal development, beginning with conception. It also discusses hereditary influences on fetal development and the nurse's role in **genetic counseling**.

## Fetal Development

Fetal development during pregnancy is measured in number of weeks after fertilization. The duration of pregnancy is about 40 weeks from the time of fertilization. This equates to 9 calendar months or approximately 266 to 280 calendar days. The three different stages of fetal development during pregnancy are

1. **Preembryonic stage:** fertilization through the second week
2. **Embryonic stage:** end of the second week through the eighth week
3. **Fetal stage:** end of the eighth week until birth

**Fetal circulation** is a significant aspect of fetal development that spans all three stages.

### Preembryonic Stage

The preembryonic stage begins with fertilization, also called *conception*. Fertilization is defined as the union of ovum and sperm, which starts the onset of pregnancy. Fertilization typically occurs around 2 weeks after the last normal menstrual period in a 28-day cycle (Dillon, 2003). Fertilization requires a timely interaction between the release of the mature ovum at ovulation and the ejaculation of enough healthy, mobile sperm to survive the hostile vaginal environment through which they must travel to meet the ovum. All things considered, the act of conception is difficult at best. To say merely that it occurs when

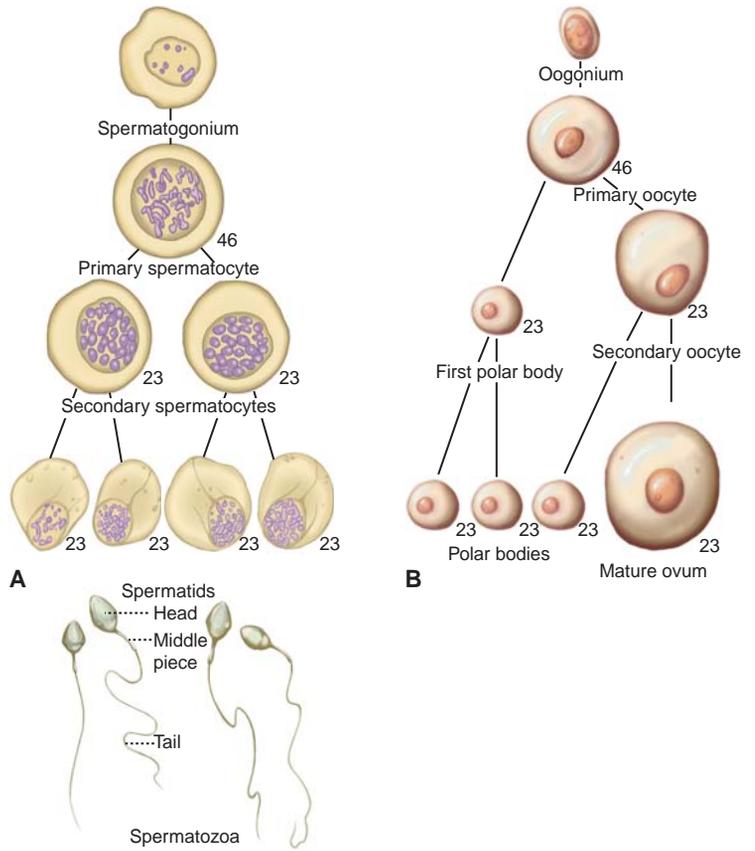
the sperm unites with the ovum is deceptively simple. Getting them both together at the right time involves an intricate interplay of hormonal preparation and an overwhelming number of natural barriers. A human being is truly an amazing outcome of this elaborate process.

Prior to fertilization, the ovum and the spermatozoon undergo the process of meiosis. The primary oocyte completes its first meiotic division before ovulation. The secondary oocyte begins its second meiotic division just before ovulation. Primary and secondary spermatocytes undergo meiotic division while still in the testes (Fig. 10-1).

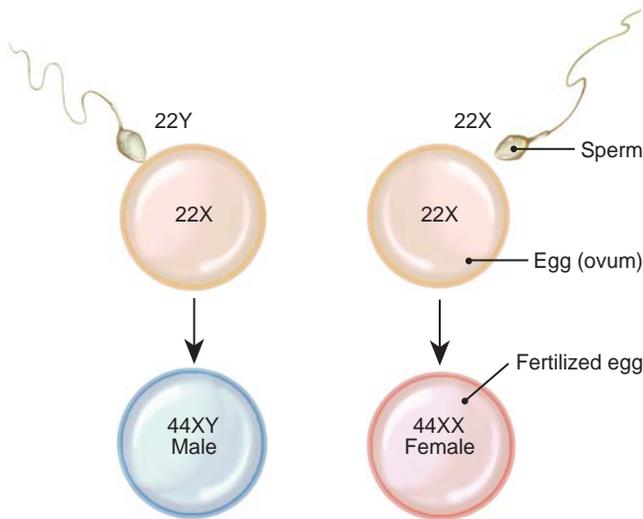
Although more than 200 million sperm/mL are contained in the ejaculated semen, only one is able to enter the female ovum to fertilize it. All others are blocked by the clear protein layer called the **zona pellucida**. The zona pellucida disappears in about 5 days. Once the sperm gets to the plasma membrane, the ovum resumes meiosis and forms a nucleus with half the number of **chromosomes** (23). When the nucleus from the ovum and the nucleus of the sperm make contact, they lose their respective nuclear membranes and combine their maternal and paternal chromosomes. Because each nucleus contains a haploid number of chromosomes (23), this union restores the diploid number (46). The resulting **zygote** begins the process of a new life. This genetic information from both ovum and sperm establishes the unique physical characteristics of the individual. Sex determination is also determined at fertilization and depends on whether the ovum is fertilized by a Y-bearing sperm or an X-bearing sperm. An XX zygote will become a female and an XY zygote will become a male (Fig. 10-2).

Fertilization takes place in the outer third of the ampulla of the fallopian tube. When the ovum is fertilized by the sperm (now called a *zygote*), a great deal of activity immediately takes place. Mitosis, or *cleavage*, takes place as the zygote is slowly transported into the uterine cavity as a result of tubal muscular movements (Fig. 10-3). After there are four cleavages, the 16 cells appear as a solid ball of cells or **morula**, meaning "little mulberry." The morula reaches the uterine cavity about 72 hours after fertilization (Sloane, 2002).

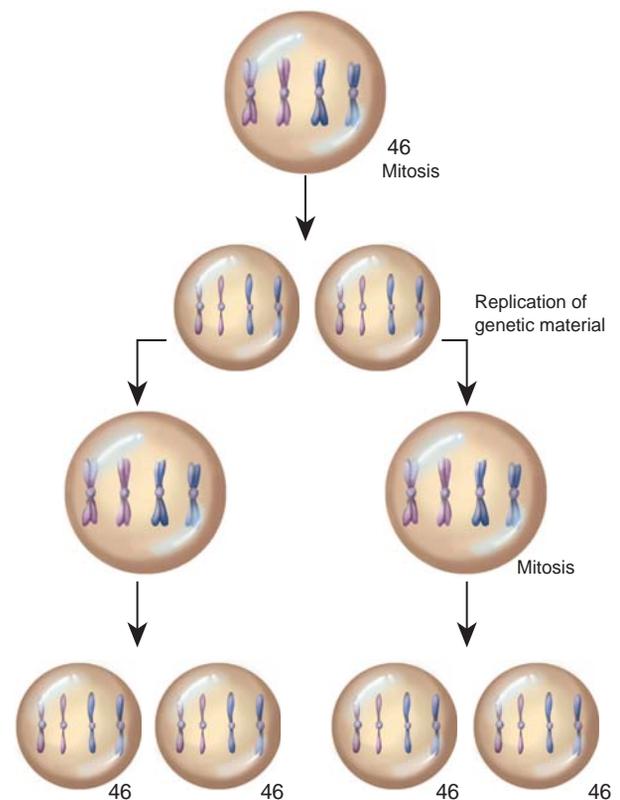
With additional cell division, the morula divides into specialized cells that will later form fetal structures. Within the morula, an off-center, fluid-filled space appears, transforming it into a hollow ball of cells called **blastocysts** (Fig. 10-4). The inner surface of the blastocysts will form the embryo and amnion. The outer layers of cells surrounding the blastocyst cavity are called **trophoblasts**. Eventually, the trophoblast develops into one of the embryonic membranes, the chorion, and helps to form the **placenta**.



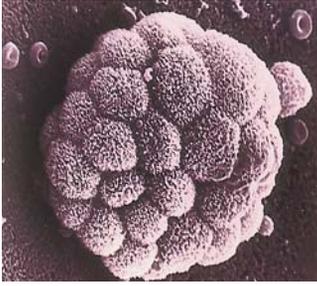
● **Figure 10-1** The formation of gametes by the process of meiosis is known as gametogenesis. **(A)** Spermatogenesis. One spermatogonium gives rise to four spermatozoa. **(B)** Oogenesis. From each oogonium, one mature ovum and three abortive cells are produced. The chromosomes are reduced to one-half the number characteristic for the general body cells of the species. In humans, the number in the body cells is 46, and that in the mature spermatozoon and secondary oocyte is 23.



● **Figure 10-2** Inheritance of gender. Each ovum contains 22 autosomes and an X chromosome. Each spermatozoon (sperm) contains 22 autosomes and either an X chromosome or a Y chromosome. The gender of the zygote is determined at the time of fertilization by the combination of the sex chromosomes of the sperm (either X or Y) and the ovum (X).



● **Figure 10-3** Mitosis of the stoma cells.



● Figure 10-4 Blastocyst.

By now the developing blastocyst needs to receive more food and oxygen to keep growing. The trophoblast attaches itself to the surface of the endometrium for further nourishment. Normal implantation occurs in the upper uterus (fundus). This location is best because it has a rich supply of blood and strong muscular fibers, which clamp down on blood vessels after the placenta separates from the inner wall of the uterus. Also, the lining is thickest here so the placenta cannot attach so strongly that it remains attached after birth (McKinney, James, Murray, & Ashwill, 2005). Figure 10-5 describes the process of fertilization and implantation.

Concurrent with the development of the trophoblast and implantation there is further differentiation of the

inner cell mass. Some of the cells become the embryo itself, and others give rise to the membranes that surround and protect it. Three embryonic layers of cells are formed as follows:

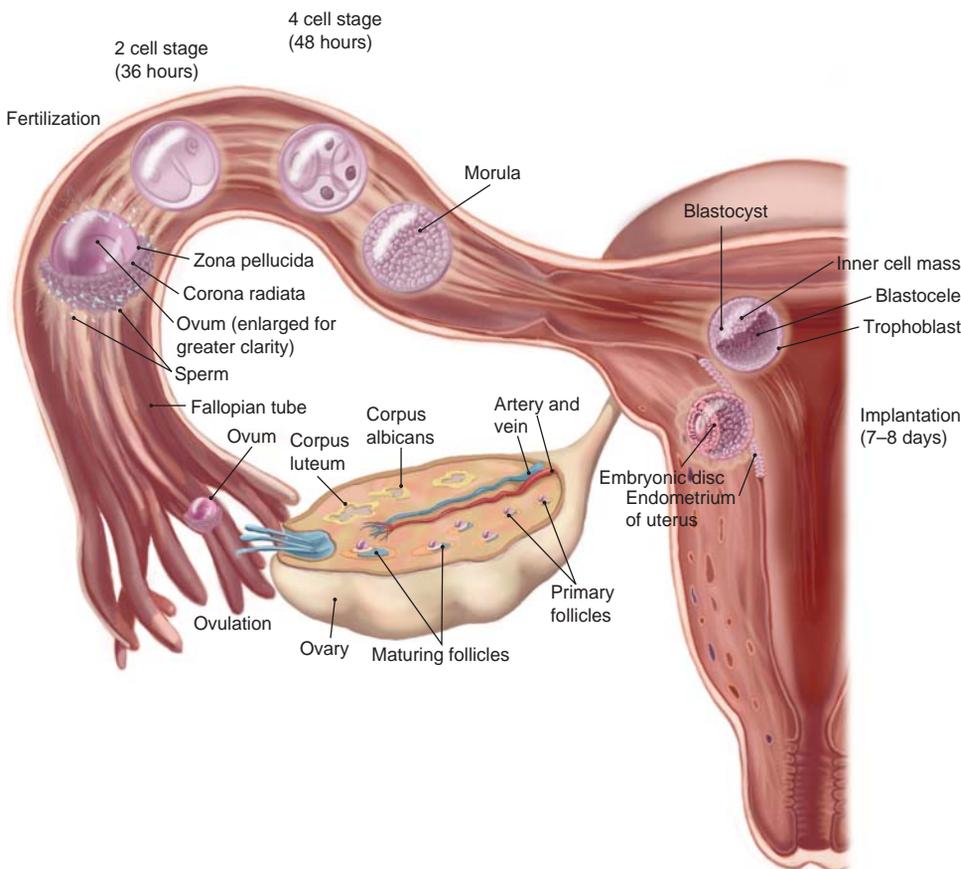
1. Ectoderm—forms the central nervous system, special senses, skin, and glands
2. Mesoderm—forms skeletal, urinary, circulatory, and reproductive organs
3. Endoderm—forms respiratory system, liver, pancreas, and digestive system

These three layers are formed at the same time as the embryonic membranes, and all tissues, organs, and organ systems develop from these three primary germ cell layers (London, Ladewig, Ball, & Bindler, 2003).

Despite all the dramatic activities that go on internally to create a human life, many women are not aware right away that a potential pregnancy has been initiated. Several weeks will pass before even one of the presumptive signs of pregnancy—missing the first menstrual period—will take place. Box 10-1 provides a summary of preembryonic development.

## Embryonic Stage

The embryonic stage of development begins at day 15 after conception and continues through week 8. Basic structures



● Figure 10-5 Fertilization and tubal transport of the zygote. From fertilization to implantation, the zygote travels through the fallopian tube, experiencing rapid mitotic division (cleavage). During the journey toward the uterus the zygote evolves through several stages, including morula and blastocyst.

## BOX 10-1

## SUMMARY OF PREEMBRYONIC DEVELOPMENT

- Fertilization takes place in ampulla of the fallopian tube.
- Union of sperm and ovum forms a *zygote* (46 chromosomes).
- Cleavage cell division continues to form a *morula* (mass of 16 cells).
- The inner cell mass is called *blastocyst*, which forms the embryo and amnion.
- The outer cell mass is called *trophoblast*, which forms the placenta and chorion.
- Implantation occurs 7 to 10 days after conception in the endometrium.

of all major body organs and the main external features are completed during this time period. See Table 10-1 and Figure 10-6 for a summary of embryonic development.

The embryonic membranes (Fig. 10-7) begin to form around the same time of implantation. The chorion consists of trophoblast cells and a mesodermal lining. It has finger-like projections called *chorionic villi* on its surface. The amnion originates from the ectoderm germ layer during the early stages of embryonic development. It is a thin protective membrane that contains amniotic fluid. As the embryo grows, the amnion expands until it touches the chorion. These two fetal membranes form the fluid-filled amniotic sac, or bag of waters, that protects the floating embryo (Littleton & Engebretson, 2005).

Amniotic fluid surrounds the embryo and increases in volume as the pregnancy progresses, reaching approximately a liter in volume by term. Amniotic fluid is derived from two sources: fluid transported from the maternal blood across the amnion and fetal urine. It is constantly changing in volume as the fetus swallows and voids into it. Sufficient amounts of amniotic fluid help maintain a constant body temperature for the fetus, permit symmetric growth and development, cushion the fetus from trauma, allow the **umbilical cord** to be relatively free of compression, and promote fetal movement to enhance musculoskeletal development. It is made up of 98% water and 2% organic matter. It is slightly alkaline and contains albumin, urea, uric acid, creatinine, bilirubin, lecithin, sphingomyelin, epithelial cells, vernix, and fine hair called *lanugo* (Lowdermilk & Perry, 2004).

The volume of amniotic fluid is important in determining fetal well-being. It gradually fluctuates throughout the course of the pregnancy. Alterations in normal amniotic fluid volume can relate to potential problems in the fetus. Having a deficiency in the amount of amniotic fluid (<500 mL at term) is called *oligohydramnios* and is associated with utero placental insufficiency and fetal renal abnormalities. Having an excessive amount of amniotic fluid (>2000 mL at term) is termed *hydram-*

*nios* and is associated with maternal disease such as diabetes, neural tube defects, chromosomal deviations, and malformations of the central nervous system and/or gastrointestinal tract that prevent normal swallowing of amniotic fluid by the fetus (Olds, London, Ladewig, & Davidson, 2004).

At the same time the placenta is developing (end of the second week), the umbilical cord is also formed from the amnion. It is the lifeline from the mother to the growing embryo. It contains one large vein and two small arteries. Wharton's jelly (a specialized connective tissue) surrounds these three blood vessels in the umbilical cord to prevent compression, which would choke off the blood supply and nutrients to the growing life inside. At term, the average umbilical cord is 22 inches long and about an inch in width (Olds et al., 2004).

The precursor cells of the placenta—the trophoblasts—first appear 4 days after fertilization as the outer layer of cells of the blastocyst. These early blastocyst trophoblasts differentiate into all the cells that form the placenta. When fully developed, the placenta serves as the interface between the mother and the developing fetus. As early as 3 days after conception, the trophoblasts make human chorionic gonadotropin (hCG), a hormone, which ensures that the endometrium will be receptive to the implanting embryo. During the next few weeks the placenta begins to make hormones that control the basic physiology of the mother in such a way that the fetus is supplied with the necessary nutrients and oxygen needed for successful growth. The placenta also protects the fetus from immune attack by the mother, removes waste products from the fetus, induces the mother to bring more food to the placenta, and, near the time of delivery, produces hormones that mature fetal organs in preparation for life outside the uterus (Kilman, 2003).

At no time during pregnancy is there any direct contact between the blood of the fetus and the blood of the mother, so there is no mixing of blood. There are always layers of fetal tissue that separate the maternal blood and the fetal blood. These fetal tissues are called the *placental barrier*. Materials can be interchanged only through diffusion. The maternal uterine arteries deliver the nutrients and the mother's uterine veins carry oxygen and the fetal waste products away. The structure of the placenta is usually completed by week 12.

The placenta is not only a transfer organ but a factory as well. It produces several hormones necessary for normal pregnancy:

- *Human chorionic gonadotropin* (or hCG)—preserves the corpus luteum and its progesterone production so that the endometrial lining of the uterus is maintained; is the basis for pregnancy tests
- *Human placental lactogen* (hPL)—modulates fetal and maternal metabolism, participates in the development of

(text continues on page 218)

**Table 10-1** Embryonic and Fetal Development

**Week 3**

Beginning development of brain, spinal cord, and heart  
 Beginning development of the gastrointestinal tract  
 Neural tube forms, which later becomes the spinal cord  
 Leg and arm buds appear and grow out from body

**Week 4**

Brain differentiates  
 Limb buds grow and develop more



4 weeks

**Week 5**

Heart now beats at a regular rhythm  
 Beginning structures of eyes and ears  
 Some cranial nerves are visible  
 Muscles innervated

**Week 6**

Beginning formation of lungs  
 Fetal circulation established  
 Liver produces RBCs  
 Further development of the brain  
 Primitive skeleton forms  
 Central nervous system forms  
 Brain waves detectable

**Week 7**

Straightening of trunk  
 Nipples and hair follicles form  
 Elbows and toes visible  
 Arms and legs move  
 Diaphragm formed  
 Mouth with lips and early tooth buds

**Week 8**

Rotation of intestines  
 Facial features continue to develop  
 Heart development complete  
 Resembles a human being  
 (Ratner, 2002)



8 weeks

**Weeks 9-12**

Sexual differentiation continues  
 Buds for all 20 temporary teeth laid down  
 Digestive system shows activity

Head comprises nearly half the fetus size  
 Face and neck are well formed  
 Urogenital tract completes development  
 Red blood cells are produced in the liver  
 Urine begins to be produced and excreted  
 Fetal gender can be determined by week 12  
 Limbs are long and thin; digits are well formed



12 weeks

**Weeks 13-16**

A fine hair develops on the head called *lanugo*  
 Fetal skin is almost transparent  
 Bones become harder  
 Fetus makes active movement  
 Sucking motions are made with the mouth  
 Amniotic fluid is swallowed  
 Fingernails and toenails present  
 Weight quadruples  
 Fetal movement (also known as *quickening*) detected by mother



16 weeks

**Weeks 17-20**

Rapid brain growth occurs  
 Fetal heart tones can be heard with stethoscope  
 Kidneys continue to secrete urine into amniotic fluid  
 Vernix caseosa, a white greasy film, covers the fetus  
 Eyebrows and head hair appear  
 Brown fat deposited to help maintain temperature  
 Nails are present on both fingers and toes  
 Muscles are well developed



20 weeks

(continued)

**Table 10-1** Embryonic and Fetal Development (continued)

**Weeks 21–24**

Eyebrows and eyelashes are well formed  
 Fetus has a hand grasp and startle reflex  
 Alveoli forming in lungs  
 Skin is translucent and red  
 Lungs begin to produce *surfactant*



25 weeks

**Weeks 29–32**

Rapid increase in the amount of body fat  
 Increased central nervous system control over body functions  
 Rhythmic breathing movements occur  
 Lungs are not fully mature  
 Fetus stores iron, calcium, and phosphorus



32 weeks

**Weeks 25–28**

Fetus reaches a length of 15 inches  
 Rapid brain development  
 Eyelids open and close  
 Nervous system controls some functions  
 Fingerprints are set  
 Blood formation shifts from spleen to bone marrow  
 Fetus usually assumes head-down position



28 weeks

**Weeks 33–38**

Testes are in scrotum of male fetus  
 Lanugo begins to disappear  
 Increase in body fat  
 Fingernails reach the end of fingertips  
 Small breast buds are present on both sexes  
 Mother supplies fetus with antibodies against disease  
 Fetus is considered full term at 38 weeks  
 Fetus fills uterus (Bailey, 2003)



37 weeks



**A**

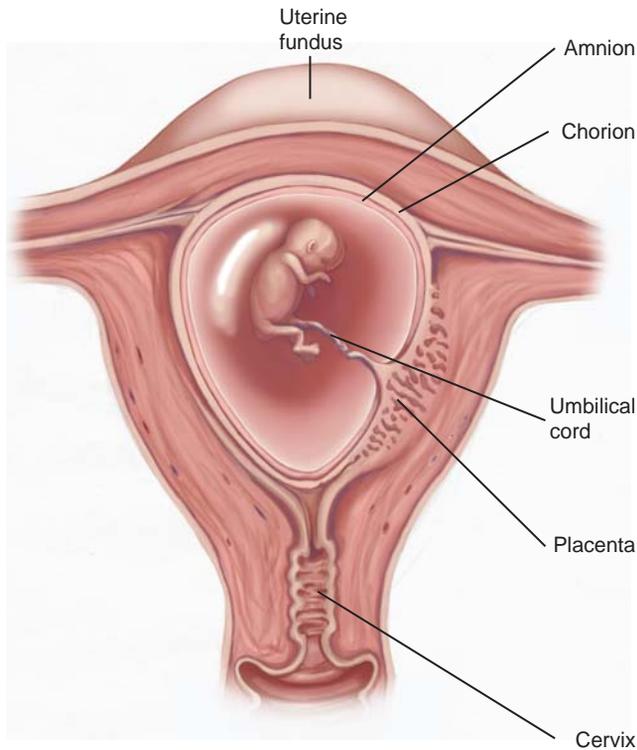


**B**



**C**

● **Figure 10-6** Embryonic development. (A) 4-week embryo; (B) 5-week embryo; (C) 6-week embryo.



● **Figure 10-7** The embryo is floating in amniotic fluid surrounded by the protective fetal membranes (amnion and chorion).

maternal breasts for lactation, and decreases maternal insulin sensitivity to increase its availability for fetal nutrition

- **Estrogen** (estriol)—causes enlargement of a woman’s breasts, uterus, and external genitalia; stimulates myometrial contractility
- **Progesterone** (progesterin)—maintains the endometrium, decreases the contractility of the uterus, stimulates maternal metabolism and breast development, provides nourishment for the early conceptus
- **Relaxin**—acts synergistically with progesterone to maintain pregnancy, causes relaxation of the pelvic ligaments, softens the cervix in preparation for birth (Condon, 2004)

The placenta should not be thought of as a barrier between mother and conceptus, but rather as a pass-through in which almost everything the mother ingests (food, alcohol, drugs) “passes through” to the developing conceptus. This is why it is so important to advise pregnant women not to take unauthorized substances (drugs, alcohol, tobacco), because they can be harmful to the conceptus.

During the embryonic stage, rapid growth of the conceptus takes place as all organs and structures are forming. It is during this critical period of differentiation when the growing embryo is most susceptible to damage from external sources, including teratogens (substances that cause birth defects such as alcohol and drugs), infections (such

as rubella or cytomegalovirus), radiation, and nutritional deficiencies.

## Fetal Stage

The average pregnancy lasts 280 days from the first day of the last menstrual period. The fetal stage is from the end of the eighth week until birth. It is the longest period of prenatal development. It is during this period that the conceptus is sufficiently developed to be called a fetus. Although all major systems are present in their basic form, dramatic growth and refinement of all organ systems take place during the fetal period (Table 10-1). Figure 10-8 depicts a 12- to 15-week fetus.

## Fetal Circulation

The circulation through the fetus during uterine life is different than ours. Fetal circulation encompasses the circulation of blood from the placenta to and through the fetus, and back to the placenta. Fetal circulation is one of the first organ systems needed to be able to function properly to sustain the fetus. Before it develops, nutrients and oxygen diffuse through the extraembryonic coelom and the yolk sac from the placenta. As the embryo increases in size, its nutrient needs increase and the amount of tissue easily reached by diffusion increases. Thus, the circulation must develop quickly and accurately (Everett & Hammill, 2002).

The circulatory system of the fetus functions much differently from that of a newborn. The most significant difference is that oxygen is received from the placenta during fetal life and via the lungs after birth. In addition, the fetal liver does not have the metabolic functions that it will have after birth because the mother’s body performs these functions. Three shunts are present in fetal life:

1. **Ductus venosus**—connects the umbilical vein to the inferior vena cava
2. **Ductus arteriosus**—connects the main pulmonary artery to the aorta
3. **Foramen ovale**—anatomic opening between the right and left atrium



● **Figure 10-8** Fetal development: 12- to 15-week fetus.

The whole function of fetal circulation is to carry highly oxygenated blood to vital areas (e.g., heart, brain) while first shunting it away from less vital ones (e.g., lungs, liver). The placenta essentially takes over the functions of the lungs and liver during fetal life, and therefore large volumes of oxygenated blood are not needed. Fetal circulation can be traced as follows: The oxygenated blood is carried from the placenta to the fetus via the umbilical vein. About half of this blood passes through the hepatic capillaries and the rest flows through the *ductus venosus* into the inferior vena cava. Blood from the vena cava is mostly deflected through the *foramen ovale* into the left atrium, then to the left ventricle, into the ascending aorta, and on to the head and upper body. This allows the fetal coronary circulation and brain to receive the blood with the highest level of oxygenation.

Deoxygenated blood from the superior vena cava flows into the right atrium, right ventricle, and then into the pulmonary artery. Because of high pulmonary vascular resistance, only a small percentage (5–10%) of the blood in the pulmonary artery flows to the lungs, the majority of it being shunted through the patent *ductus arteriosus* and then down the descending aorta (O’Toole, 2003). The fetal lungs are essentially nonfunctional and filled with fluid. The presence of this fluid makes the lungs resistant to the flow of blood into them and they receive only enough blood as required for proper nourishment. Finally, two umbilical arteries carry the unoxygenated blood from the descending aorta back to the placenta.

At birth, a dramatic change in the fetal circulatory pattern occurs. The foramen ovale, ductus arteriosus, ductus venosus, and the umbilical vessels are no longer needed. When the newborn takes a first breath at birth, the lungs inflate, which tends to draw blood into them from the right ventricle. The increase in blood flow into and out of the lungs increases pressure in the left atrium. This causes a one-way flap on the left side of the foramen ovale, called the *septum primum*, to press against the opening, effectively separating the two atria. This also increases blood flow to the lungs, because blood entering the right atrium can no longer bypass the right ventricle, which pumps it into the pulmonary artery and on to the lungs. The ductus venosus closes with the clamping of the umbilical cord and inhibition of blood flow through the umbilical vein. The ductus arteriosus constricts partly in response to higher arterial oxygen levels that occur after the first few breaths. This closure prevents blood from the aorta from entering the pulmonary artery (Sabella, 2003). Thus, the newborn is left with the adult pattern of circulation, in which deoxygenated blood enters the right atrium and is pumped through the right atrium to the pulmonary arteries. In the lungs it is oxygenated to be returned to the left atrium of the heart via the pulmonary veins. Oxygenated blood can then be pumped to all parts of the body by the left ventricle. See Figure 10-9 for a summary of the fetal circulation pattern.

## Genetics

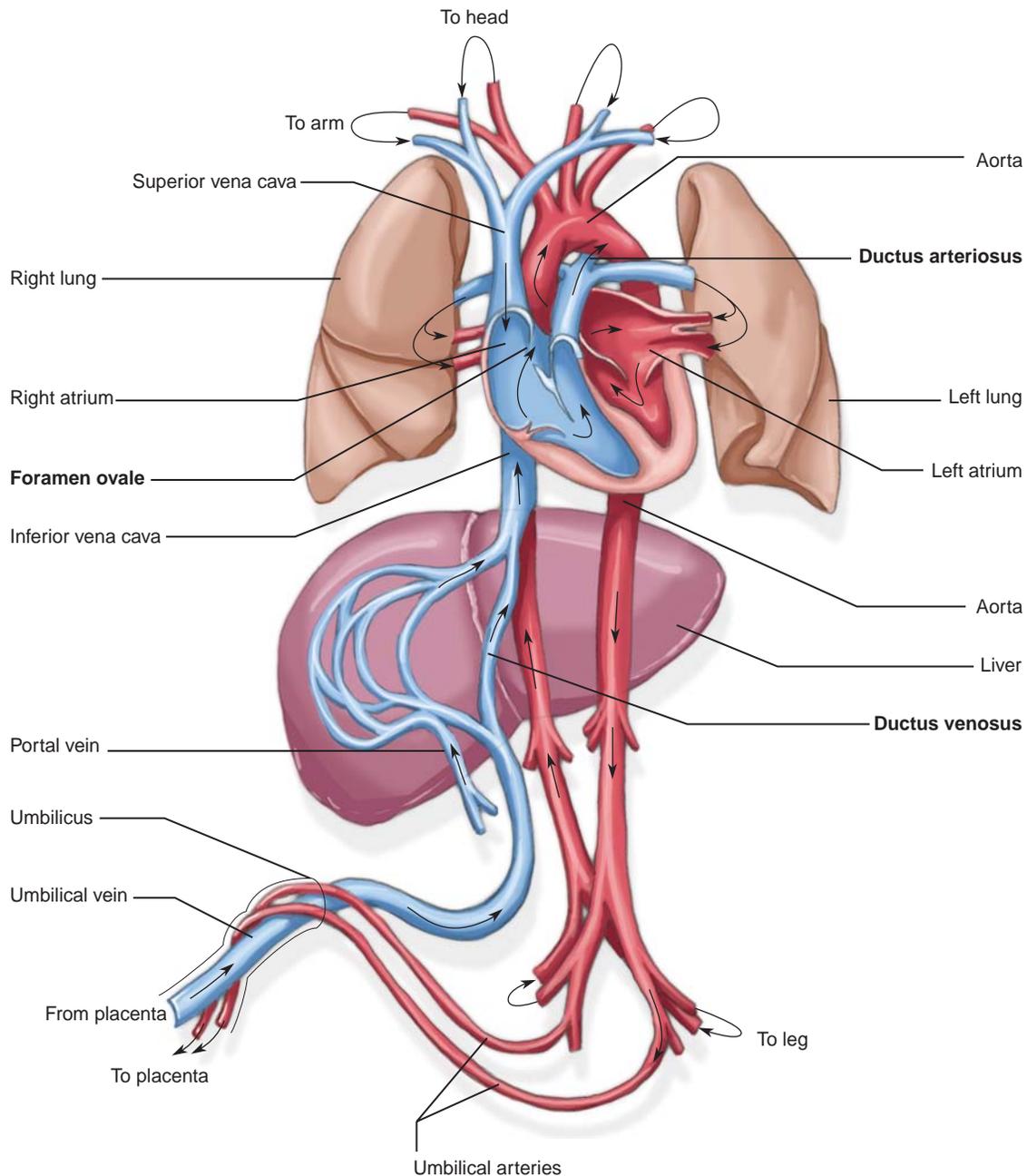
**Genetics** is the study of the patterns of inheritance of specific traits (O’Toole, 2003). Traditionally, genetics has been associated with childbearing decision making and caring for children with genetic disorders. Recent genetic and technologic advances are helping to expand our understanding of how genetic changes affect human diseases such as diabetes, cancer, Alzheimer disease, and other multifactorial diseases that are prevalent in adults (Greco & Mahon, 2002). Genetic science has the potential to revolutionize health care with regard to national screening programs, predisposition testing, detecting genetic disorders, and pharmacogenetics. Today, nurses are required to have basic skills and knowledge in genetics so they can take on new roles and help patients understand the personal and societal effects of genetic information (Burton, 2003).

### Advances in Genetic Science

Recent advances in genetic knowledge and technology have affected all areas of health. These advances have increased the number of health interventions that can be undertaken with regard to genetic disorders. For example, genetic diagnosis is now possible before conception and earlier in pregnancy. Genetic testing can now identify presymptomatic conditions in children and adults. Gene therapy can be used to replace or repair defective or missing **genes** with normal ones. Gene therapy has been used for a variety of disorders, including cystic fibrosis, melanoma, HIV, and hepatitis (Lewis, Heitkemper, & Dirksen, 2004). It may also treat many chronic illnesses. The potential exists for creation of increased intelligence and size through genetic intervention. Genetic agents may replace drugs, general surgery may be replaced by gene surgery, and genetic intervention may replace radiation (Terzioglu & Dinc, 2004).

The information gained from the Human Genome Project, an international 13-year effort begun in 1990 by the Department of Energy and National Institutes of Health, has brought new advances in genetic testing (Lea & Williams, 2002). These advances will have a profound effect on the diagnosis, treatment, and prevention of diseases. The goal of this project was to map the human genome (complete set of genetic instructions in the nucleus of each human cell) by 2005. A substantially complete version of this project was announced in 2002. Two key findings from the project were found: (1) All human beings are 99.9% identical at the DNA level and (2) approximately 30,000 genes make up the human genome (International Human Genome Sequencing Consortium, 2004). An individual’s genome represents their genetic blueprint, which determines genotype (the gene pairs inherited from parents) and phenotype (physical characteristics and manifestations) (Edgar, 2004).

Research from the Human Genome Project has provided a better understanding of the genetic contribution to disease. It is now recognized that most common human



● **Figure 10-9** Fetal circulation. Arrows indicate the path of blood. The umbilical vein carries oxygen-rich blood from the placenta to the liver and through the ductus venosus. From there it is carried to the inferior vena cava to the right atrium of the heart. Some of the blood is shunted through the foramen ovale to the left side of the heart where it is routed to the brain and upper extremities. The rest of the blood travels down to the right ventricle and through the pulmonary artery. A small portion of the blood travels to the non-functioning lungs, while the remaining blood is shunted through the ductus arteriosus into the aorta to supply the rest of the body.

diseases such as myocardial infarction, cancer, mental illness, diabetes, and Alzheimer disease are a result of complex interactions between a number of factors, including the influence of one or more genes and a variety of environmental exposures (Greco & Mahon, 2002).

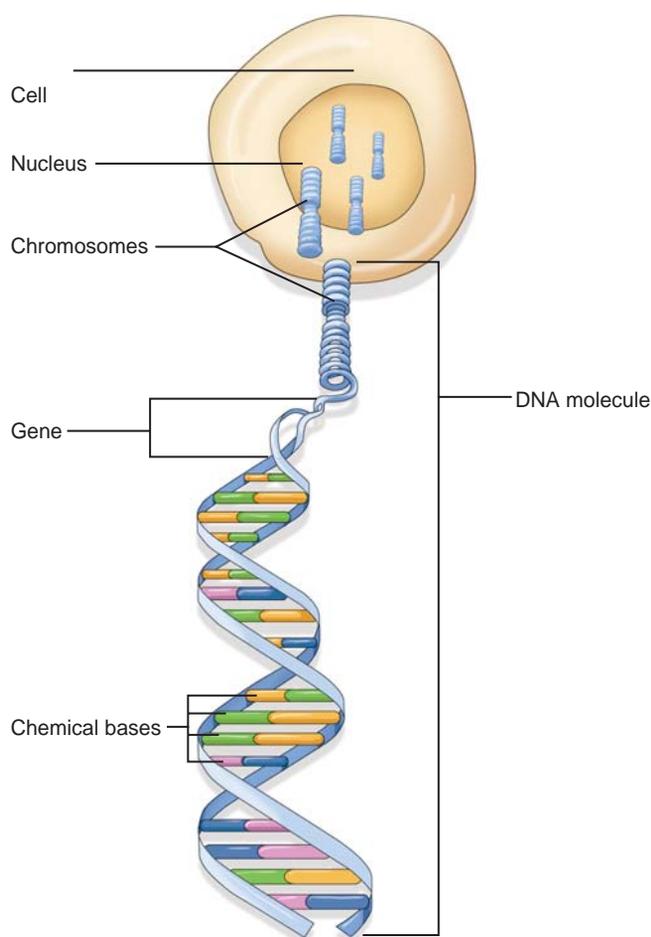
The potential benefits of these discoveries are vast, but so is the potential for misuse. These benefits challenge all healthcare professionals to consider the many

ethical, legal, and social ramifications of genetics knowledge in human lives. In the near future, individual risk profiling based on an individual's unique genetic makeup will be used to tailor prevention, treatment, and ongoing management of health conditions. This profiling will raise issues associated with patient privacy and confidentially related to workplace discrimination and access to health insurance. Issues of autonomy are equally problem-

atic as society considers how to address injustices that will inevitably surface when disease risk can be determined years in advance of its occurrence. Nurses will certainly play an important role in developing new policies and charting a course in this bold new world of genetics. Nurses, however, must know the basics of genetics to do this.

## Basics of Genetics

The nucleus within the cell is the controlling factor in all cellular activities because it contains chromosomes, which are made up of genes—the “how-to” units of instruction for building. Each person has a unique genetic constitution, or genotype, made up of 30,000 to 40,000 genes. An individual *gene* is a unit of heredity. Each gene has a segment of DNA with a specific set of instructions for making proteins needed by body cells for proper functioning. Genes control the types of proteins made and the rate at which they are produced (Sloane, 2002) (Fig. 10-10). Any change in gene structure or location leads to a **mutation**,



● **Figure 10-10** DNA is made up of four chemical bases. Tightly coiled strands of DNA are packaged in units called chromosomes, housed in the cell’s nucleus. Working subunits of DNA are known as genes. (From the National Institute of Health and National Cancer Institute. [1995]. *Understanding gene testing* [NIH Pub. No. 96-3905]. Washington, DC: U.S. Department of Human Services.)

which may alter the type and amount of protein produced (Fig. 10-11). Genes never act in isolation; they always interact with other genes and the environment. They are arranged in a specific linear formation along a chromosome. Genes carry instructions for either dominant or recessive traits, which determine the color of eyes and hair, body build, and the growth and development of every physical and biochemical system (Lea, 2003).

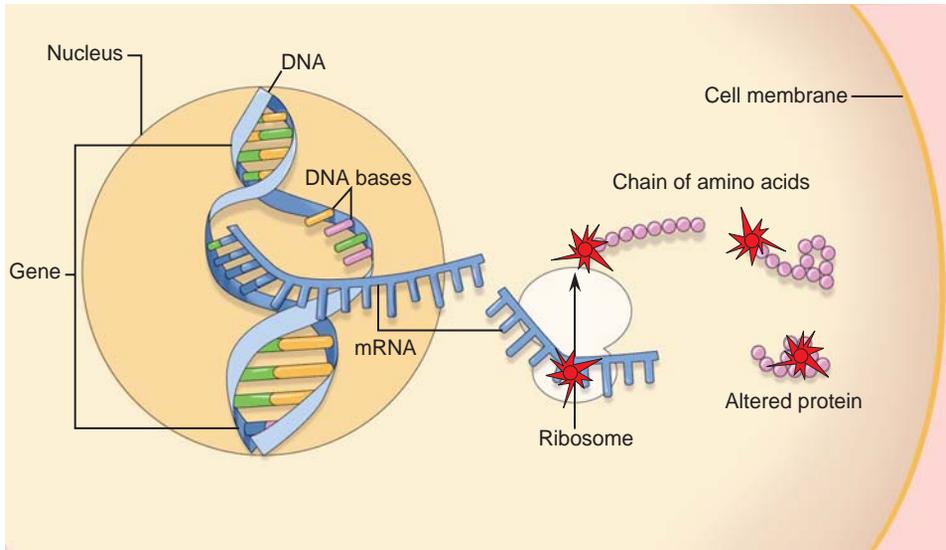
A chromosome is a filament-like nuclear structure consisting of chromatin that stores genetic information as base sequences in DNA, and with a number that is constant in each species. Each chromosome is composed of varying numbers of genes. A total of 22 chromosome pairs are autosomes, and the 23rd pair comprises the sex chromosomes. Humans have 46 paired chromosomes that are found in all cells of the body—except the ovum and sperm cells, which have just 23 chromosomes. In males and females, 22 of the chromosome pairs are the same (autosomes). The last pair is the sex chromosomes—two X chromosomes in females, and an X chromosome and a Y chromosome in males. Offspring receive one chromosome of each of the 23 pairs from each parent.

DNA stores genetic information and encodes the instructions for synthesizing specific proteins needed to maintain life. The structure of DNA is double stranded and is identified as a double helix. The side pieces of the double helix are made up of a sugar, deoxyribose, and a phosphate, occurring in alternating groups. The cross-connections or rungs of the ladder are attached to the sides and are made up of four nitrogenous bases: adenine, cytosine, thymine, and guanine. The sequence of the base pairs as they form each rung of the ladder is referred to as the *genetic code* (Sloane, 2002).

Regulation and expression of the thousands of human genes is very complex and is the result of many intricate interactions within each cell. Alterations in gene structure, function, transcription, translation, and protein synthesis can influence an individual’s health (Lea, 2002). Gene mutations are a permanent change in the sequence of DNA. Some mutations have no significant affect, whereas others can have a tremendous impact on the health status of the individual. Several genetic disorders can result from these mutations, such as cystic fibrosis, sickle-cell disease, phenylketonuria, or hemophilia.

The pictorial analysis of the number, form, and size of an individual’s chromosomes is termed **karyotype**. White blood cells and fetal cells in amniotic fluid are commonly analyzed. The chromosomes are numbered from the largest to the smallest, 1 to 22, and the sex chromosomes are designated by the letter X or Y. A female karyotype is designated as 46,XX and a male karyotype is designated as 46,XY. See Figure 10-12 for an example of a karyotyping pattern.

Just as there are maternal and paternal copies of each chromosome, there are also maternal and paternal copies of each gene. The sequence of the maternal and paternal gene pair may be *homozygous* (identical) or *heterozygous*



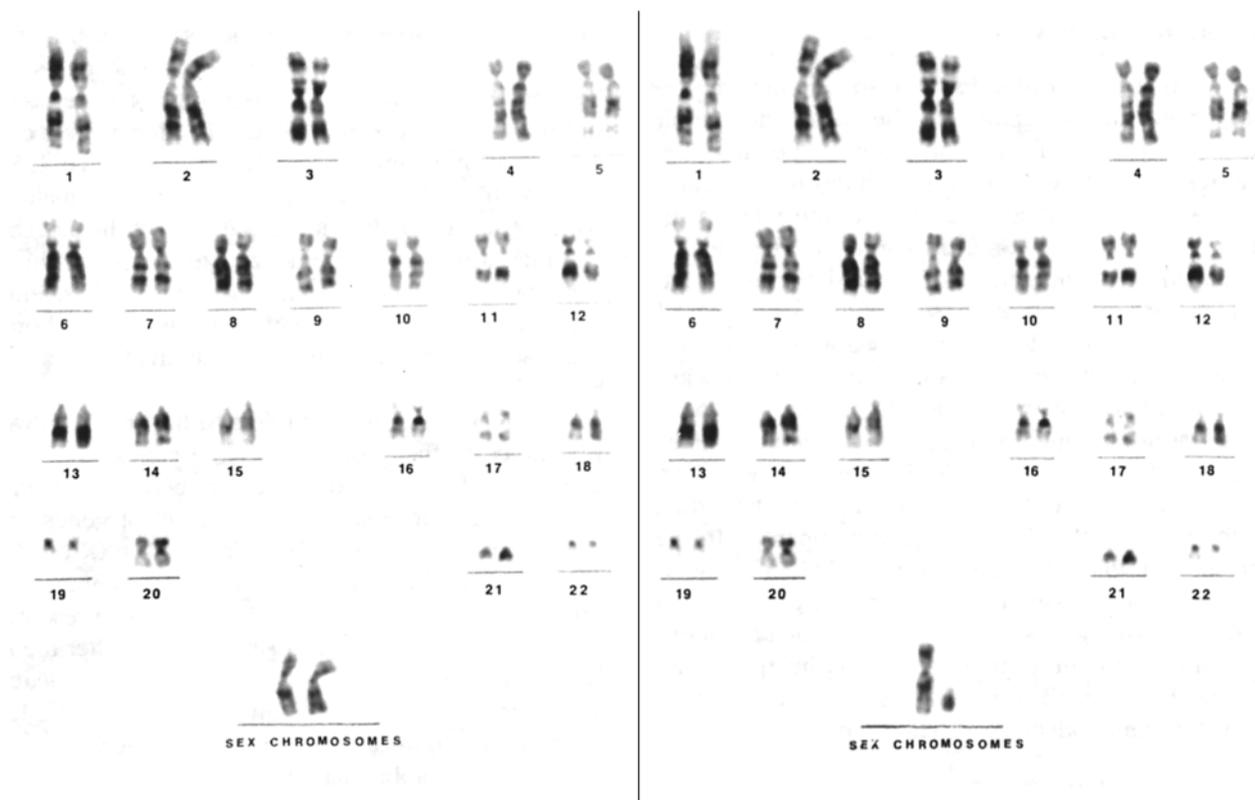
● **Figure 10-11** When a gene contains a mutation, the protein encoded by that gene will be abnormal. Some protein changes are insignificant, others are disabling. (From the National Institutes of Health and National Cancer Institute. [1995]. *Understanding gene testing* [NIH Pub. No. 96-3905]. Washington, DC: U.S. Department of Human Services.)

(different). Homozygous means both alleles (a different form of the same gene copy) for a trait are the same. For example, WW stands for homozygous dominant; ww stands for homozygous recessive. Heterozygous indicates members of the allelic pair are different, such as Ww. For example, an offspring inherits blood type AB. The type A allele (gene copy) came from one parent and the type B allele from the other parent. This would therefore be considered heterozygous for the gene pair. If the offspring

inherited blood type O, this would be considered a homozygous gene pairing because both parents gave the same gene pair (B) (Black & Hawks, 2005).

### Chromosomal Abnormalities

The mechanisms underlying cell division are so complex that an occasional error is not unexpected. Errors resulting in chromosomal abnormalities can occur during mitosis



● **Figure 10-12** Karyotype pattern. (A) Normal female karyotype. (B) Normal male karyotype.

or meiosis. These errors can occur in either the autosomes or sex chromosomes. About 1 in 160 live-born infants is born with a chromosomal abnormality (March of Dimes, 2005). These often cause major defects because they involve added or missing genes. There are two major kinds of autosomal errors: changes in the number of chromosomes or changes in the structure of the chromosomes.

### Numeric Abnormalities

- Entire single added chromosome (trisomy)
- Entire single chromosome missing (monosomy)
- One or more added sets of chromosomes (polyploidy)

### Structural Abnormalities

- Part of a chromosome missing or added
- Rearrangements of material within the chromosomes
- Two chromosomes that adhere to each other
- Fragility of a specific site on the X chromosome (Murray & McKinney, 2006)

### Numeric Abnormalities

Numeric abnormalities of chromosomes are most commonly seen as **trisomies**, **monosomies**, and **polyploidy**. All involve added or missing single chromosomes, or multiple sets of chromosomes.

### Trisomies

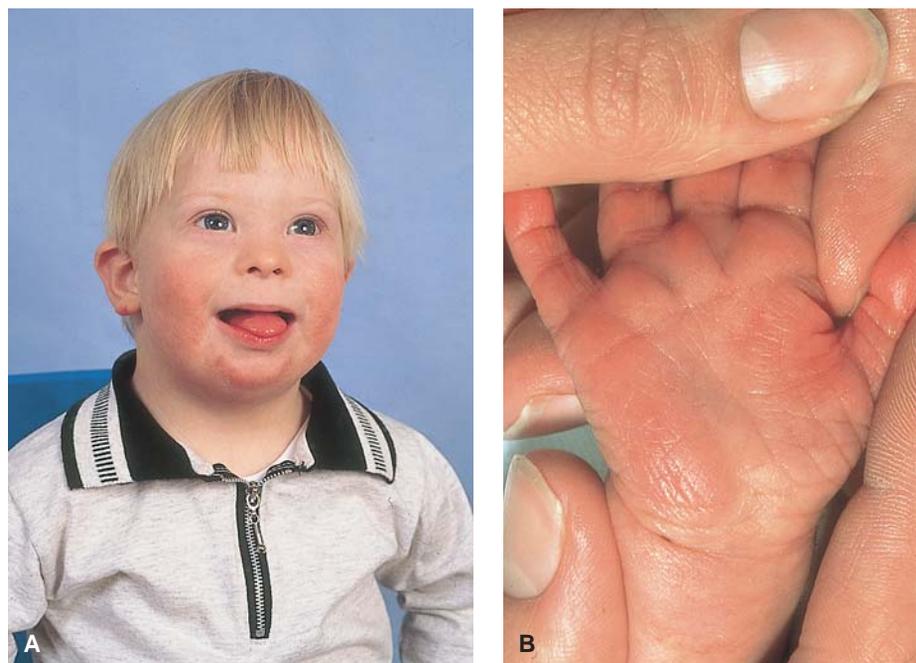
Trisomies are the product of the union of a normal ovum or sperm with a gamete that contains an extra chromosome. This means there will be 47 chromosomes instead of the normal 46. Down syndrome is an example of a trisomy. Individuals with Down syndrome have three copies of chromosome 21. Down syndrome affects 1 in

800 to 1000 live-born babies. The risk of this and other trisomies increases with maternal age. The risk of having a baby with Down syndrome is about 1 in 1250 for a woman at age 25, 1 in 1000 at 30, 1 in 400 at 35, and 1 in 100 at age 40 (March of Dimes, 2004). Individuals with Down syndrome have characteristic features that are usually identified at birth (Fig. 10-13). These common characteristics include

- Small, low-set ears
- Hyperflexibility
- Muscle hypotonia
- Deep crease across palm (termed *simian crease*)
- Flat facial profile
- Small white, crescent-shaped spots on irises
- Open mouth with protruding tongue
- Broad, short fingers (Littleton & Engebretson, 2005)

The outlook for children with Down syndrome is much brighter now than it was years ago. Most children with Down syndrome have mental retardation in the mild to moderate range. With early intervention and special education, many learn to read and write, and participate in diverse childhood activities (March of Dimes, 2004). Despite modern medical technology, the individual with Down syndrome has a shortened life expectancy, with 22% dying in the first decade and 50% dying by the age of 60, mostly from cardiac and malignant conditions (Littleton & Engebretson, 2005).

Another chromosomal disorder involving an extra chromosome is Klinefelter syndrome, which occurs only in males. About 1 in 400 males born have Klinefelter syndrome. There is an extra X chromosome (XXY) present. The extra genetic material causes abnormal development



● **Figure 10-13** (A) Typical facial features of the child with Down syndrome. (B) A simian line, a horizontal crease seen in children with Down syndrome.

of the testicles, resulting in decreased production of sperm and male sex hormones. Clinical manifestations may include

- Mild mental retardation
- Small testicles
- Infertility
- Long arms and legs
- Enlarged breast tissue (gynecomastia)
- Scant facial and body hair
- Decreased sex drive (libido) (Mayo Clinic, 2002)

No treatment can correct this genetic abnormality, but testosterone replacement therapy can improve symptoms resulting from a deficiency. Most males with Klinefelter syndrome (XXY) are diagnosed after their 14th year of life after experiencing a late onset of puberty. Infertility is common and life expectancy is normal (Verklan & Walden, 2004).

Two other common trisomies are trisomy 18 and trisomy 13. Trisomy 18 and trisomy 13 are, respectively, the second and third most commonly diagnosed autosomal trisomies in live-born infants. Although these conditions are associated with a high degree of infant mortality, 5 to 10% of children with these conditions survive beyond the first year of life (Rasmussen, Wong, Yang, May, & Friedman, 2003). Trisomy 18 or Edward syndrome occurs in 1 of every 5000 newborns, with advanced maternal age as a causative factor. Prenatally, there are several findings apparent on ultrasound, which includes intrauterine growth restriction (IUGR), hydramnios or oligohydramnios, cardiac malformations, single umbilical artery, and decrease in fetal movement. Additionally, trisomy 18 has been associated with an decrease in maternal serum levels of maternal serum alpha-fetoprotein (MSAFP) and hCG. Most affected newborns are female, with a 4:1 ratio to males. Affected newborns have 47 chromosomes (three at chromosome 18) and are characterized by severe mental retardation, growth deficiency of cranium (microcephaly), low-set ears, small for gestational age, seizures, drooping eyelids, webbing of fingers, congenital heart defects, rocker-bottom feet, and severe hypotonia (Sterk, 2004). Infants with trisomy 18 have multiple anomalies that are severe, and life expectancy is greatly reduced beyond a few months.

Trisomy 13 or Patau syndrome affects 1 of 12,000 newborns with 47 chromosomes (three of chromosome 13) present. Maternal age is also thought to be a causative factor in this genetic disorder (Lea, 2003). The common abnormalities associated with trisomy 13 are microcephaly, cardiac defects, central nervous system anomalies, polydactyly (Fig. 10-14), severe mental retardation, severe hypotonia, and seizures (Lowdermilk & Perry, 2004). Life expectancy is only a few months for most infants with trisomy 13. Care is supportive in nature for these infants.



● Figure 10-14 An infant with trisomy 13 has supernumerary digits (polydactyly).

### Monosomies

Monosomies exist when each body cell has a missing chromosome, with a total number of 45. The only monosomy compatible with life is Turner syndrome, or monosomy X. It affects about 1 in 2000 newborn girls (March of Dimes, 2005). Clinical manifestations include low posterior hair line and webbing of the neck, short stature, broad skeletal abnormalities, shield-like chest with widely spaced nipples, puffy feet, no toenails, underdeveloped secondary sex characteristics, and infertility (Postellon, 2005). Only about a third are diagnosed as newborns with this syndrome, with the remaining two thirds being diagnosed in early adolescence when they experience primary amenorrhea. Most females with Turner syndrome are of normal intelligence and usually live essentially normal lives (Sterk, 2004).

### Polyploidy

Polyploidy causes an increase in the number of haploid sets (23) of chromosomes in a cell. Triploidy refers to three whole sets of chromosomes in a single cell (in humans, a total of 69 chromosomes per cell); tetraploidy refers to four whole sets of chromosomes in a single cell (in humans, a total of 92 chromosomes per cell). Polyploidy usually results in an early spontaneous abortion and is incompatible with life.

### Structural Abnormalities

Structural abnormalities involve only part of the chromosome and generally occur as deletions or translocations. Part of the chromosome may be missing or added, or DNA within the chromosome may be rearranged. They include the **cri du chat syndrome**, **fragile X syndrome**, and many others.

### Cri du Chat Syndrome

Cri du chat syndrome, or *cry of the cat*, is caused by a missing piece of chromosome 5. It was named cri du chat based on the distinctive cry in newborns associated with a laryngeal defect, which sounds like a mewling cat. Incidence of the disorder is thought to be approximately 1 in 50,000 live births (Chen, 2005). It is characterized by a cat-like, high-

pitched cry in infancy; severe mental retardation; microcephaly; low birth weight and slow growth; hypotonia; failure to thrive; wide-set eyes; small jaw; low-set ears; and various organ malformations. No specific treatment is available for this syndrome. With contemporary interventions, the chance of survival to adulthood is possible. Currently, death occurs in 6 to 8% of the overall population affected with the syndrome. Pneumonia, aspiration pneumonia, and congenital heart defects are the common causes of death (Chen, 2005). Parents should be referred for genetic counseling (Brooks, 2003).

### Fragile X Syndrome

Fragile X syndrome, also termed *Martin-Bell syndrome*, is an X-linked chromosomal abnormality. The X chromosome demonstrates breaks and gaps. The syndrome is usually diagnosed by molecular DNA studies. Conservative estimates report that fragile X syndrome affects approximately 1 in 4000 males and 1 in 8000 females. The rate of the female carrier state has been estimated to be as high as 1 in 250 (Jewell, 2004). Typically, a female becomes the carrier and will be mildly affected. The male who receives the X chromosome that has a fragile site will exhibit the full effects of the syndrome. Transmission occurs through carrier mothers and not through unaffected carrier fathers (McKinney et al., 2005). It is characterized by mental retardation, hyperactivity, short attention span, hand flapping, strabismus, hypotonia, speech delay, inflexible behavior, autistic-like behavior, poor eye contact, tactile defensiveness, double jointedness, and perseverative speech (continued repetition of words or phrases). Fragile X syndrome is the most common form of male retardation (Jewell, 2005). Aside from the morbidity associated with mental retardation and cognitive/behavioral/neuropsychological problems, the life span of an individual with fragile X syndrome is unaffected by the disorder. There is no cure for this disorder. Referrals to speech, occupational, and physical therapy are needed, as well as special education and counseling (Hockenberry, 2005).

### Inheritance Patterns

Genetic disorders can be categorized into autosomal dominant, autosomal recessive, X linked, or multifactorial. If the mutation occurs on the autosome, the genetic disorder is termed *autosomal*. If the defect is on the X chromosome, the genetic disorder is called *X linked*. Inherited characteristics are passed from parent to child by the genes in each chromosome. These traits are classified as dominant (strong) or recessive (weak).

### Autosomal Dominant Inherited Disorders

**Autosomal dominant inherited disorders** occur when an abnormal gene pair (heterozygous) from one parent is capable of causing disease even though the match-

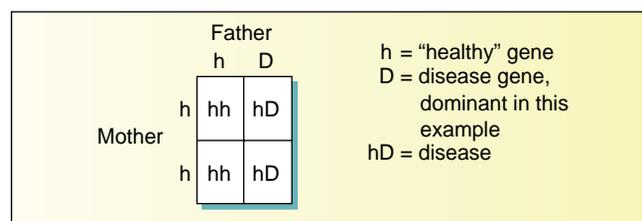
ing gene from the other parent is normal. The abnormal gene dominates the outcome of the gene pair (Fig. 10-15). The following are characteristics of autosomal dominant inheritance:

- Individuals are affected in succeeding generations.
- Affected individuals will have an affected parent.
- Children will have a 50% chance of being affected.
- Male and female family members are equally affected.
- Severity of the inherited disorder varies among family members.
- Having the gene mutation does not mean an individual will have the disease (Lewis et al., 2004).

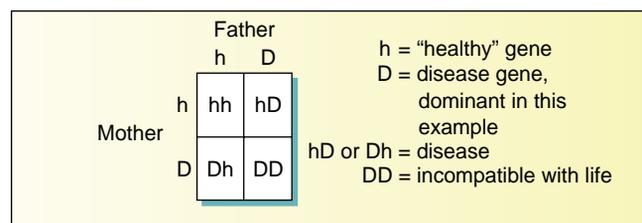
Examples of autosomal dominant inherited disorders are familial hypercholesterolemia, neurofibromatosis, breast and ovarian cancer related to BRCA genes, Huntington disease, and hereditary nonpolyposis colorectal cancer (Lea, 2002).

### Autosomal Recessive Inherited Disorders

**Autosomal recessive inherited disorders** are caused by mutations of two gene pairs (homozygous) on a chromosome. Recessive inheritance occurs when both genes of a pair are abnormal, which then produce disease. If only one gene in the pair is abnormal, the disease is not manifested or is only mildly manifested. However, a person with a single defective gene is called a *carrier* (meaning the disease can be passed on to the children; Fig. 10-16). Autosomal recessive inherited disorders are generally seen among particular ethnic groups (e.g., Tay-Sachs disease, which is highest among Jewish people of central and eastern European descent) and tend to occur more often in children whose parents are related by blood (e.g., first cousins). These types of conditions have a horizontal, rather than vertical, inheritance pattern, with relatives in a

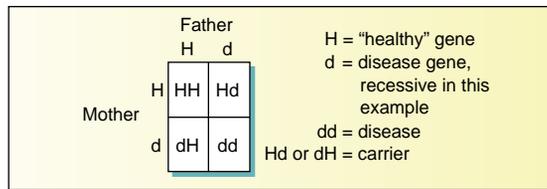


A

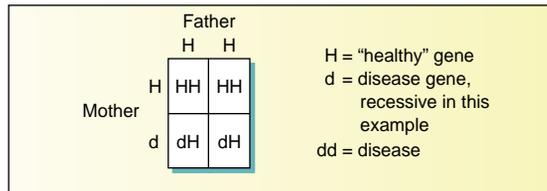


B

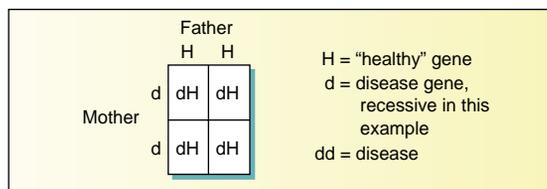
● Figure 10-15 Autosomal dominant inheritance.



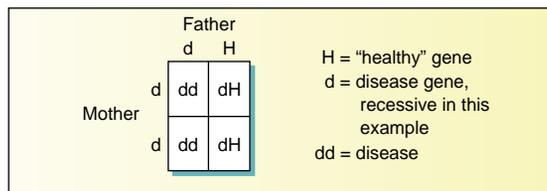
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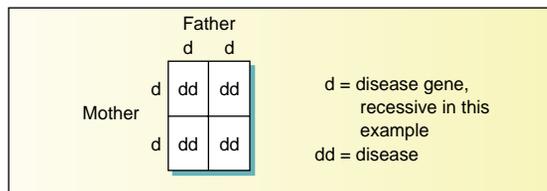
B



C



D



E

● Figure 10-16 Autosomal recessive inheritance.

single generation tending to have the condition (Littleton & Engebretson, 2005). The following are characteristics of autosomal recessive inheritance:

- Usually there is a negative family history of the disorder.
- Males and females are equally affected.
- Affected offspring have unaffected parents who are heterozygous.
- Each pregnancy carries a 25% risk of transmitting the condition.
- Offspring have a 50% chance of becoming a trait carrier (Lea, 2002).

Examples of autosomal recessive disorders include sickle-cell disease, thalassemia, phenylketonuria, Tay–Sachs disease, and cystic fibrosis.

### X-Linked Inherited Disorders

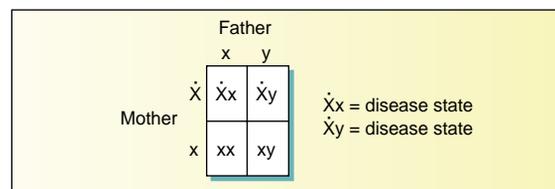
**X-linked inherited disorders** can follow dominant or recessive patterns; however, in each case, the gene mutation is on the X chromosome. Usually only men are affected by these disorders because women who carry the mutated gene on one X chromosome have another X chromosome to compensate for the mutation. However, women who carry the mutated gene can transmit the mutated gene to their offspring (Fig. 10-17) (Lewis et al., 2004). The following are characteristics of X-linked inheritance:

- Affected individuals are usually males.
- Females are affected by the condition if it is a dominant X-linked disorder.
- Most affected individuals will have unaffected parents.
- Daughters of an affected male are usually carriers.
- Female carriers have a 50% chance of transmitting the disorder to their sons.
- Sons of an affected male are usually unaffected (Spahis, 2002).

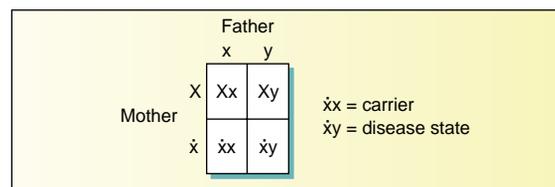
Examples of X-linked disorders are hemophilia, Duchenne dystrophy (childhood muscular dystrophy), and color blindness (red–green).

### Multifactorial Inherited Disorders

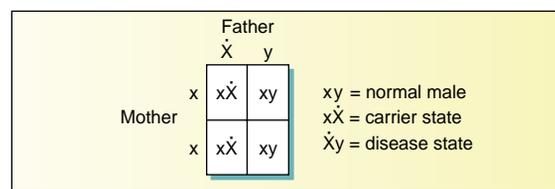
**Multifactorial inherited disorders**, or polygenic disorders, are caused by a combination of polygenic (several genes) and environmental factors. Not all conditions are transmitted via a single gene. In multifactorial conditions,



A



B



C

● Figure 10-17 X-linked inheritance.

more than one gene is involved and environmental factors are also involved. Environmental factors might include health status, age of the parents, and/or exposure to pollution. These disorders tend to run in families, but do not show the same inherited characteristics as the single-gene mutation conditions. The cause of many birth defects and common adult-onset health conditions, such as arthritis, diabetes, and cancer, is multifactorial and complex. The following are characteristics of multifactorial inheritance:

- There is an additive effect by having more family members affected.
- Fewer genes are involved if environmental influences are great.
- There is a tendency for gender bias in occurrence rates for some disorders such as pyloric stenosis in males and congenital hip dysplasia in females.
- It is difficult to determine or predict the risk factors of disorders to offspring.
- The disorders vary from mild to severe (Spahis, 2002).

Examples of multifactorial inherited disorders are cleft lip, soft palate, congenital heart disease, neural tube defects, pyloric stenosis, hypertension, diabetes, some heart diseases, cancer, and mental illness (Lowdermilk & Perry, 2004).

## Nursing Management

The nurse is likely to interact with the patient and her family in a variety of ways related to genetics—taking a family history, scheduling genetic testing, explaining the purposes of all screening and diagnostic tests, answering questions, and addressing concerns raised by family members.

## Nursing Assessment

Nurses in any practice setting can obtain a patient's genetic history during the initial encounter. The purpose is to gather patient and family information that may provide clues regarding whether the patient has a genetic trait, inherited condition, or inherited predisposition (Lea, 2002). At a basic level, all nurses should be able to take a family medical history to assist in identifying those at risk for hereditary conditions, and make a referral to a genetic specialist when appropriate.

At least three generations should be spanned when taking a family history during the initial patient encounter. Box 10-2 presents focused assessment questions to be asked of each family member. Based on the information gathered from the genetic history, the nurse must decide whether a referral is necessary to a genetic specialist or whether further evaluation is needed. Prenatal testing to assess for genetic risks and defects might be used to identify genetic disorders. These tools are described in Table 10-2.

## Genetic Counseling

Genetic counseling provides detailed information on the occurrence or risk of recurrence of a genetic disease in

### BOX 10-2

#### FOCUSED HEALTH ASSESSMENT: GENETIC HISTORY

- What was the cause and age of death for deceased family members?
- Does any consanguinity exist between relatives?
- Do any serious illnesses or chronic conditions exist?
  - If so, what was the age of onset?
- Do any female family members have a history of miscarriages, stillbirths, or diabetes?
- Do any female members have a history of alcohol or drug use during pregnancy?
- What were the ages of female members during child-bearing, especially if older than 35?
- Do any family members have mental retardation or developmental delays?
- Do any family members have a known or suspected metabolic disorder such as PKU?
- Do any family members have an affective disorder such as bipolar disorder?
- Have any close relatives been diagnosed with any type of cancer?
- What is your ethnic background (explore as related to certain disorders)?
- Do any family members have a known or suspected chromosomal disorder?
- Do any family members have a progressive neurologic disorder?

Source: Lea, 2003.

that family (Greco & Mahon, 2002). Genetic counselors help families make informed decisions about their future based on the assessment of their current risk profile.

When all the previous information has been processed and analyzed, the genetic counselor discusses with the family all the implications of the findings, presents options, and outlines possible outcomes. Genetic counseling is a nondirective process in that family members are given the facts to make their own decisions. Paramount in this whole process is the fact that family members are the ones who have to live with their decisions, and therefore healthcare professionals must respect and support them throughout this period.

Nursing responsibilities related to genetic counseling include

- Interviewing and active listening skills to identify genetic concern
- Knowing basic genetic terminology and inheritance patterns
- Explaining basic concepts of probability and disorder susceptibility
- Safeguarding privacy and confidentiality of patients' genetic information
- Providing complete informed consent to facilitate decisions about genetic testing

Table 10-2 Prenatal Tests to Assess Risk for Genetic Disorders?

Test	Description	Indication	Timing
Family pedigree	A visual representation of a family history	To identify an inheritance pattern that needs further evaluation	Ideally prior to conception; however, may be mapped anytime during pregnancy
Alpha-fetoprotein	A sample of the woman's blood is drawn to evaluate plasma protein that is produced by the fetal liver, yolk sac, and GI tract, and crosses from the amniotic fluid into the maternal blood.	Increased levels might indicate a neural tube defect, Turner syndrome, tetralogy of Fallot, multiple gestation, omphalocele, gastroschisis, or hydrocephaly. Decreased levels might indicate Down syndrome or trisomy 18.	Typically performed between 15 and 18 weeks' gestation
Amniocentesis	Amniotic fluid aspirated from the amniotic sac; safety concerns include infection, pregnancy loss, and fetal needle injuries	To perform chromosome analysis, alpha-fetoprotein, DNA markers, viral studies, karyotyping; and identify inborn errors of metabolism	Usually performed between 15 and 20 weeks' gestation to allow for adequate amniotic fluid volume to accumulate; results take 2 to 4 weeks
Chorionic villus sampling	Removal of small tissue specimen from the fetal portion of the placenta, which reflects the fetal genetic makeup; main complications include severe transverse limb defects and spontaneous pregnancy loss	To detect fetal karyotype, sickle-cell anemia, phenylketonuria, Down syndrome, Duchenne muscular dystrophy, and numerous other genetic disorders	Typically performed between 10 and 12 weeks' gestation, with results available in less than a week
Percutaneous umbilical blood sampling	Insertion of a needle directly into a fetal umbilical vessel under ultrasound guidance; two potential complications: fetal hemorrhage and risk of infection	Used for prenatal diagnosis of inherited blood disorders such as hemophilia A, karyotyping, detection of fetal infection, determination of acid-base status, and assessment and treatment of isoimmunization	Generally performed after 16 weeks' gestation

(continued)

**Table 10-2** Prenatal Tests to Assess Risk for Genetic Disorders? (continued)

Test	Description	Indication	Timing
Fetal nuchal translucency (FNT)	An intravaginal ultrasound that measures fluid collection in the subcutaneous space between the skin and the cervical spine of the fetus	To identify fetal anomalies; abnormal fluid collection can be associated with genetic disorders (trisomies 13, 18, and 21), Turner syndrome, cardiac deformities, and/or physical anomalies. When the FNT is greater than 2.5 mm, the measurement is considered abnormal.	Performed between 10 and 14 weeks' gestation
Level II ultrasound/fetal scan	Use of high-frequency sound waves to visualize the fetus	Enables evaluation of structural changes to be identified early	Typically performed after 18 weeks' gestation
Triple marker test	Serum screening test using the levels of three maternal serum markers—MSAFP, unconjugated estriol, and hCG—in combination with maternal age to calculate risk	To identify risk for Down syndrome, neural tube defects, and other chromosomal disorders. Elevated hCG combined with lower than normal estriol and MSAFP levels indicate increased risk for Down syndrome or other trisomy condition.	Performed between 16 and 18 weeks' gestation

Sources: Agarwal, 2003; Lowdermilk & Perry, 2004; Littleton & Engebretson, 2005; Mattson & Smith, 2004; Terzioglu & Dinc, 2004; Wald, Rodeck, Hackshaw, Walters, Chitty, & Mackinson, 2003; Youngkin & Davis, 2004.

- Discussing costs of genetic services, benefits, and risks of using health insurance for payment of genetic services; and potential risks of discrimination
- Recognizing and defining ethical, legal, and social issues
- Providing accurate information about risks and benefits of genetic testing
- Providing culturally appropriate media to convey genetic information
- Monitoring patients' emotional reactions after receiving genetic analysis
- Providing information on appropriate local support groups
- Knowing your own limitations and making appropriate referrals (Cook, 2003)

### Consider THIS!

As I waited for the genetic counselor to come into the room, my mind was filled with numerous fears and questions. What does an inconclusive amniocentesis really

mean? What if this pregnancy produced an abnormal baby? How would I cope with a special child in my life? If only I had gone to the midwife sooner when I thought I was pregnant, but still in denial. Why did I wait so long to admit this pregnancy and get prenatal care? If only I had started to take my folic acid pills when prescribed. Why didn't I research my family's history to know of any hidden genetic conditions? What about my sister with a Down syndrome child? What must I have been thinking? I guess I could play the "what-if" game forever and never come up with answers. It was too late to do anything about this pregnancy because I was in my last trimester. I started to pray silently when the counselor opened the door. . . .

**Thoughts:** This woman is reviewing the last several months, looking for answers to her greatest fears. Inconclusive screenings can introduce emotional torment for many women as they wait for validating results. Are these common thoughts and fears for many women facing potential genetic disorders? What supportive interventions might the nurse offer?

One of the most important aspects of genetic counseling in which the nurse is involved is follow-up counseling after the couple has been to the specialist. The nurse is in an ideal position to help families review what has been discussed during the genetic counseling sessions and to answer any additional questions they might have. The nurse can provide helpful information, support families in their decisions, and make appropriate referrals within their community.

## Summary

Nurses have entered a new era in which the reality of genetic advances and knowledge affects the practice of every nurse. Understanding the contribution of genetic factors to the development of disease is fundamental to the practice of every nurse. The future of genetic technology is here with genetic testing, gene therapy, pharmacogenetics, genetic selection, and other challenges to nursing practice. Nurses need to be ready by educating themselves about conception, fetal development, and genetics to assist their patients in their decision-making process for their future health and welfare.

Genetic advances pose new and unique ethical dilemmas to the issues of privacy, confidentiality, access to and justice in health care, and informed health decisions. For nurses, this means ensuring the privacy and confidentiality of genetic information derived from assessments, genetic tests, and other interventions. Nurses will be instrumental in providing patients and their families with the information to make knowledgeable decisions about their reproductive health and their personal lives. Nurses will need additional education in genetics so they can assume their potential roles in the ever-growing field of genetics.

## KEY CONCEPTS

- Fertilization takes place in the outer third of the ampulla of the fallopian tube.
- Three embryonic layers of cells are formed as follows:
  - Ectoderm—forms central nervous system, special senses, skin, and glands
  - Mesoderm—forms skeletal, urinary, circulatory, and reproductive systems
  - Endoderm—forms respiratory system, liver, pancreas, and digestive system
- Amniotic fluid surrounds the embryo and increases in volume as the pregnancy progresses, reaching approximately a liter in volume by term.
- At no time during pregnancy is there any direct connection between the blood of the fetus and the blood of the mother, so there is no mixing of blood.
- A specialized connective tissue known as Wharton's jelly surrounds the three blood vessels in the umbilical cord to prevent compression, thus choking off the blood supply and nutrients to the growing life inside.
- The placenta protects the fetus from immune attack by the mother, removes waste products from the fetus, induces the mother to bring more food to the placenta, and, near the time of delivery, produces hormones that mature fetal organs in preparation for life outside the uterus.
- The purpose of fetal circulation is to carry highly oxygenated blood to vital areas (heart and brain) while first shunting it away from less vital ones (lungs and liver).
- Humans have 46 paired chromosomes that are found in all cells of the body, except the ovum and sperm cells, which have just 23 chromosomes.
- Genetic science has the potential to revolutionize health care with regard to national screening programs, predisposition testing, detecting genetic disorders, and pharmacogenetics.
- Research from the Human Genome Project has provided a better understanding of the genetic contribution to disease.
- Each person has a unique genetic constitution, or genotype, made up of approximately 30,000 to 40,000 genes.
- Humans have 46 paired chromosomes that are found in all cells of the body, except the ovum and sperm cells, which have just 23 chromosomes.
- Genetic disorders can be categorized as autosomal dominant, autosomal recessive, X-linked recessive, or multifactorial.
- Genetic counselors help families make informed decisions about their future based on the assessment of their current risk profile.

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## Web Resources

- American Society of Human Genetics, [www.faseb.org/genetics/ashg/ashgmenu.htm](http://www.faseb.org/genetics/ashg/ashgmenu.htm)
- CDC: Office of Genetics, [www.cdc.gov/genetics/activities/ogdp.htm](http://www.cdc.gov/genetics/activities/ogdp.htm)
- 5p-Society (Cat Cry Syndrome), [www.fivepminus.org/](http://www.fivepminus.org/)
- Gene Clinics, [www.geneclinics.org](http://www.geneclinics.org)
- Gene Tests, [www.genetests.org](http://www.genetests.org)
- Genetic Alliance, [www.geneticalliance.org](http://www.geneticalliance.org)
- Human Genome Project of the US Department of Energy, [www.ornl.gov/hgmis](http://www.ornl.gov/hgmis)
- International Society of Nurses in Genetics, [www.nursing.creighton.edu/isong](http://www.nursing.creighton.edu/isong)
- Klinefelter Syndrome and Associates, <http://genetic.org/ks/>
- National Coalition for Health Professional Education in Genetics, [www.nchpeg.org](http://www.nchpeg.org)
- National Down Syndrome Society, [www.ndss.org/](http://www.ndss.org/)
- National Human Genome Research Institute, [www.nhgri.gov](http://www.nhgri.gov)
- Turner Syndrome Society of the United States, [www.turner-syndrome-us.org](http://www.turner-syndrome-us.org)
- Virtual Library on Genetics, [www.ornl.gov/TechResources/Human\\_Genome/genetics.html](http://www.ornl.gov/TechResources/Human_Genome/genetics.html)
- Visible Embryo, [www.visembryo.ucsf.edu](http://www.visembryo.ucsf.edu)

## Chapter WORKSHEET

### ● MULTIPLE CHOICE QUESTIONS

1. Fertilization usually occurs in the
  - a. Fundus of the uterus
  - b. Endometrium of the uterus
  - c. Upper portion of fallopian tube
  - d. Follicular tissue of the ovary
2. Pregnancy tests detect the presence of which hormone in the maternal blood or urine?
  - a. hPL
  - b. hCG
  - c. FSH
  - d. TSH
3. Which of the following prenatal tests is used to detect neural tube defects such as spina bifida or anencephaly?
  - a. Alpha-fetoprotein
  - b. Fetoscopy
  - c. CT scan
  - d. Coombs test
4. Most health conditions are believed to result from which of the following?
  - a. Unrelated to human genetics
  - b. Have no underlying cause of disease
  - c. Caused by poor lifestyle choices
  - d. Have both genetic and environmental factors
5. The first step in establishing the genetic risk for a condition is to
  - a. Observe the patient and family over time
  - b. Conduct extensive psychological testing
  - c. Obtain a thorough family health history
  - d. Complete an extensive exclusionary list
6. Increased risk of having offspring with chromosomal abnormalities is greater in women who are
  - a. Expecting twins or triples
  - b. Emotionally distressed over the pregnancy
  - c. Experiencing their first pregnancy
  - d. 35 years or older
7. Which of the following is characteristic of Down syndrome?
  - a. The infant has 46 paired chromosomes
  - b. The male infant has only 23 chromosomes
  - c. The female has two extra YY chromosomes
  - d. The infant has an extra chromosome number 21

### ● CRITICAL THINKING EXERCISE

1. Mr. and Mrs. Martin wish to start a family, but they can't agree on something important. Mr. Martin wants his wife to be tested for cystic fibrosis (CF) to see if she is a carrier. Mr. Martin had a brother with CF and watched his parents struggle with the hardship and the expense of caring for him for years, and he doesn't want to experience it in his own life. Mr. Martin has found out he is a CF carrier. Mrs. Martin doesn't want to have the test because she figures that once a baby is in their arms, they will be glad, no matter what.
  - a. What information/education is needed for this couple to consider before deciding whether to have the test?
  - b. How can you assist this couple in their decision-making process?
  - c. What is your role in this situation if you don't agree with their decision?

## ● STUDY ACTIVITIES

1. Obtain/rent the video entitled *Miracle of Life* and view it for the spectacular photography showing conception and fetal development. What are your impressions? Is the title of this video realistic?
2. Arrange a visit to a local hospital obstetric ultrasound department. Request permission to observe several obstetric ultrasounds and discuss with the technician what the findings may indicate (after the patient has left).
3. Select one of the Helpful Informational Resource Web sites to explore the topic of genetics. Critique the information presented. Was it understandable to a lay person? What specifically did you learn?
4. Draw your own family pedigree representing your family history that identifies their inheritance patterns. Share it with your family to validate its accuracy. What did you discover about your family's past health?
5. Select one of the various prenatal screening tests (alpha-fetoprotein, amniocentesis, chorionic villus sampling, or fetal nuchal translucency) and research it in depth. Role-play with another nursing student how you would explain its purpose, procedure, and potential findings to an expectant couple at risk for a potential fetal abnormality.

Share your findings with your classmates during a discussion group.

