Learning Objectives

Upon completion of the chapter, you will be able to:

- **1.** Characterize the process of fertilization, implantation, and cell differentiation.
- **2.** Examine the functions of the placenta, umbilical cord, and amniotic fluid.
- **3.** Outline normal fetal development from conception through birth.
- **4.** Compare the various inheritance patterns, including nontraditional patterns of inheritance.
- **5.** Analyze examples of ethical and legal issues surrounding genetic testing.
- **6.** Research the role of the nurse in genetic counseling and genetic-related activities.

KEY TERMS

allele

blastocyst

embryonic stage

fertilization

fetal stage

genes

genetic counseling

genetics

genome

genomics

genotype

heterozygous

homozygous

karyotype

mosaicism

monosomies

morula

mutation

phenotype

placenta

polyploidy

preembryonic stage

teratogen

trisomies

trophoblast

umbilical cord

zona pellucida

zygote

Robert and Kate Shafer have just received the good news that Kate's pregnancy test was positive. It had been a long and anxious 3 years of trying to start a family. Although both are elated about the prospect of becoming parents, they are also concerned about the possibility of a genetic problem because Kate is 38 years old. What might be their first step in looking into their genetic concern? As a nurse, what might raise concerns for you?

WOW: *Words of Wisdom*

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. Conception occurs when a healthy ovum from the woman is released from the ovary, passes into an open fallopian tube, and starts its journey downward. Sperm from the male is deposited into the vagina and swims approximately 7 inches to meet the ovum at the outermost portion of the fallopian tube, the area where fertilization takes place. This process occurs in about an hour (Bennett & Williamson, 2010). When one spermatozoon penetrates the ovum's thick outer membrane, a new living cell is formed that is unlike the cells of either parent. Soon, the two nuclei will fuse, bringing together about 25,000 genes to guide human development.

Nurses caring for the childbearing family need to have a basic understanding of conception and prenatal development so they can identify problems or variations and can 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. An average human pregnancy lasts for about 280 days or 40 weeks from the date of the last menstrual period (LMP). Traditionally, it has been calculated as 10 lunar months, or, in terms of the modern calendar, 9 months. Fertilization of the egg by the sperm, however, usually occurs (considering an average menstrual cycle of 28 days) 14 days after the last period. Thus, the average actual duration of a human pregnancy (gestation period) is 280 days − 14 days = 266 days.

The three stages of fetal development during pregnancy are the:

- 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 the union of ovum and sperm, which is the starting point of pregnancy. Development during this stage takes place in an organized fashion that is cephalocaudal, proximal to distal, and general to specific. Fertilization typically occurs around 2 weeks after the last normal menstrual period in a 28-day cycle (Carcio & Secor, 2010). 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 overly simple because this union requires an intricate interplay between hormonal preparation and overcoming 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. Gametogenesis is the process by which gametes (ovum or sperm cells) are produced to initiate the development of a new individual. The gametes must have a haploid number of chromosomes (23) so when they come together to form the zygote, the normal human diploid number of chromosomes (46) is established (**Fig. 10.1**).

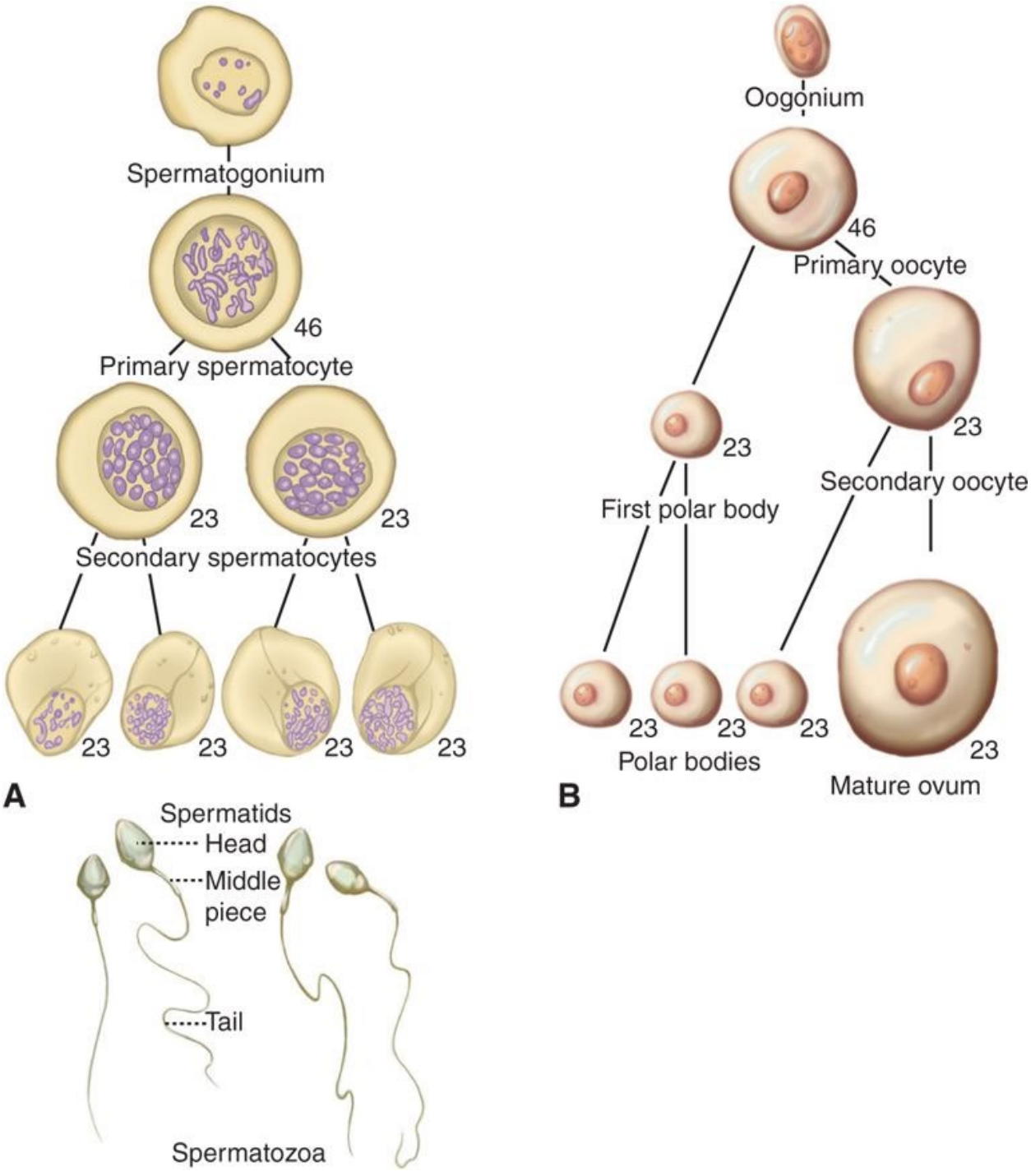


FIGURE 10.1

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.

Although each milliliter of ejaculated semen contains more than 200 million sperm, only one is able to enter the 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 reaches 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. The 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**

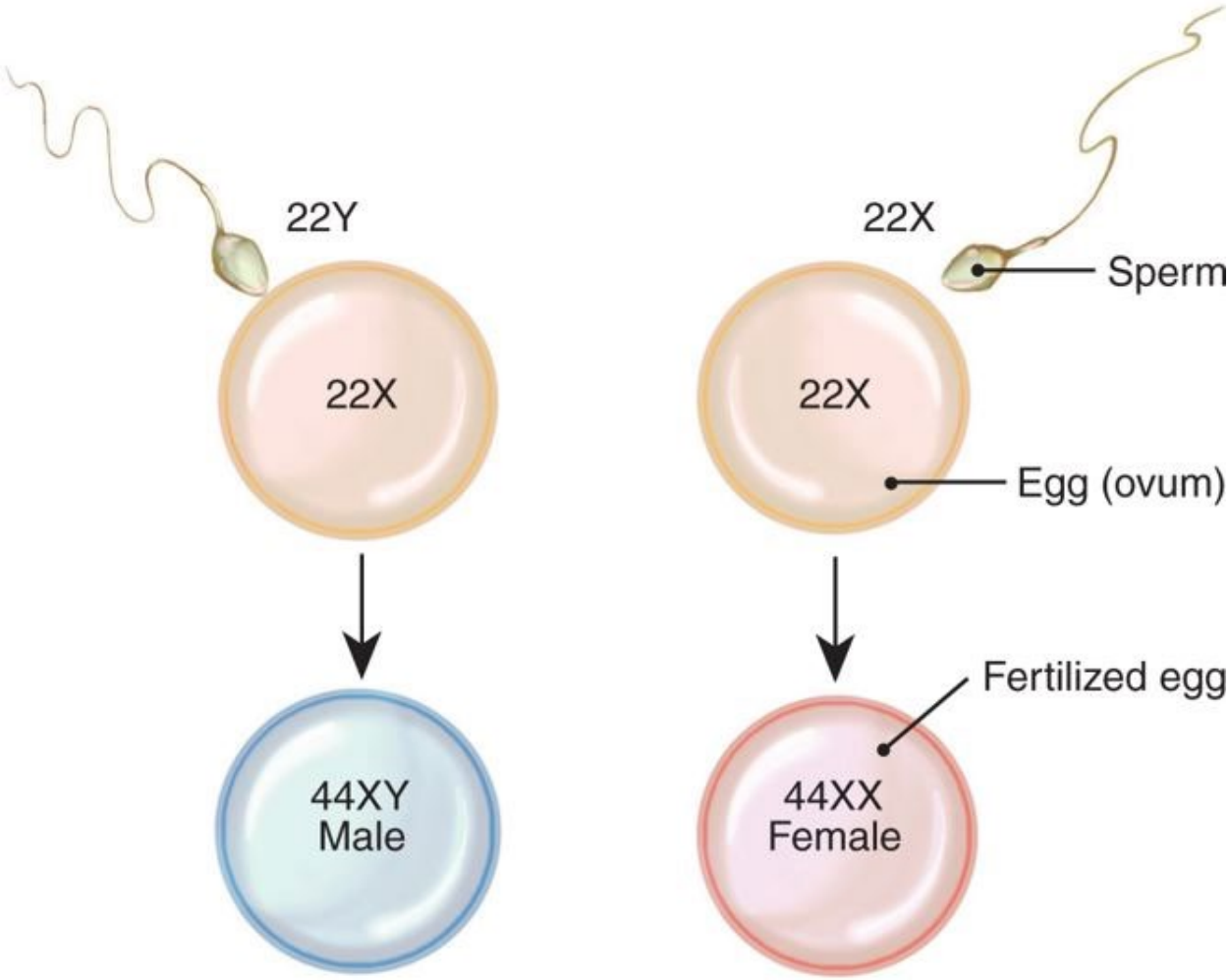


FIGURE 10.2

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).

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,* occurs as the zygote is slowly transported into the uterine cavity by tubal muscular movements (**Fig. 10.3**). After a series of 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 (Stables & Rankin, 2010).

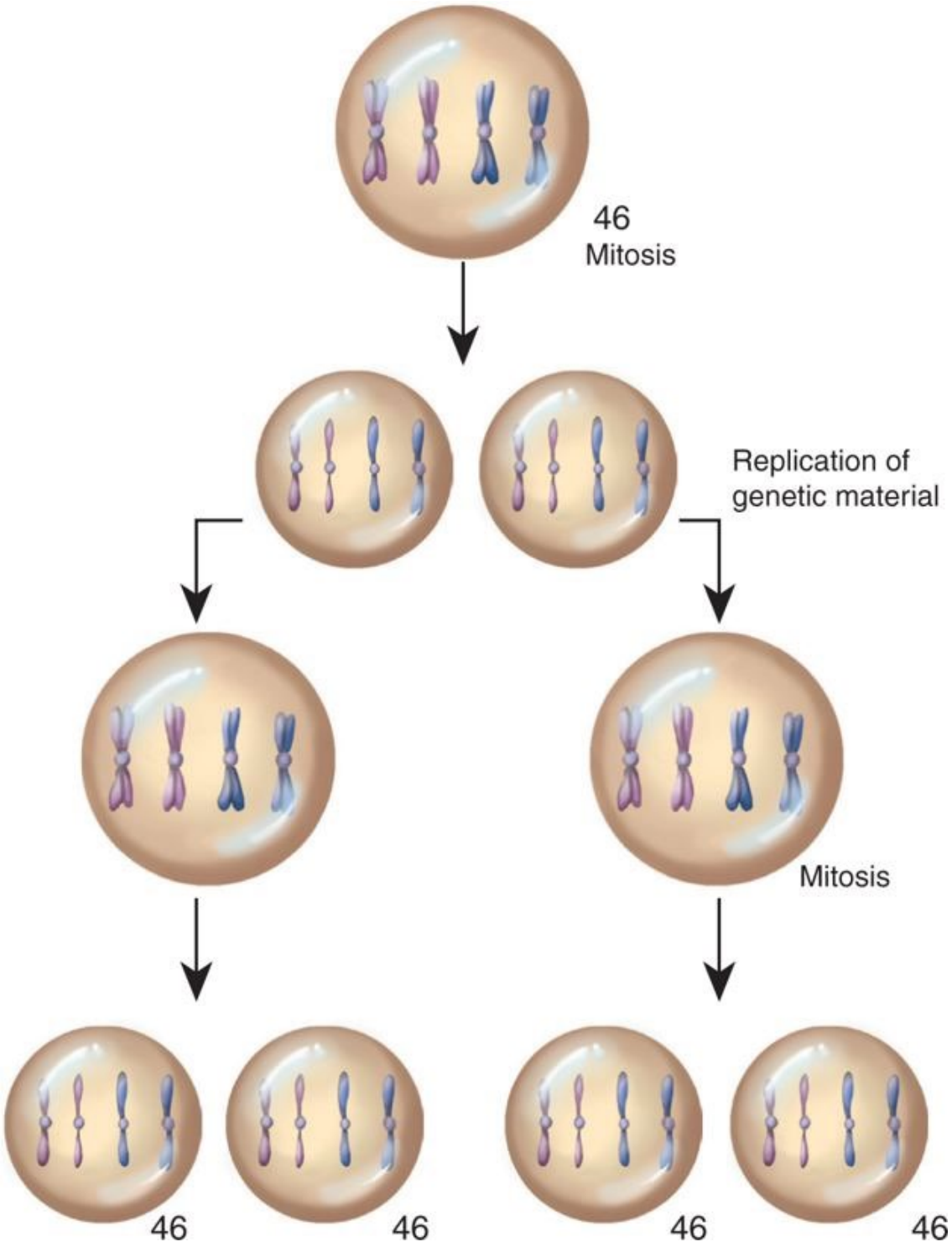


FIGURE 10.3

Mitosis of the stoma cells.

Multiple fetuses can also occur at the time of fertilization when more than one ovum is fertilized. Identical twins (also called monozygotic twins) occur when one fertilized egg splits and develops into two (or occasionally more) fetuses. The fetuses usually share one placenta. Identical twins have the same genes, so they generally look alike and are the same sex. Fraternal twins (also called dizygotic twins) develop when two separate eggs are fertilized by two different sperm. Each twin usually has its own placenta. Fraternal twins (like other siblings) share about 50% of their genes, so they can be different sexes. They generally do not look any more alike than brothers or sisters born from different pregnancies. Fraternal twins are more common than identical twins (see **Chapter 19** for further details).

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 a **blastocyst** (**Fig. 10.4**). The inner surface of the blastocyst will form the embryo and amnion. The outer layer of cells surrounding the blastocyst cavity is called a **trophoblast**. Eventually, the trophoblast develops into one of the embryonic membranes, the chorion, and helps to form the placenta.

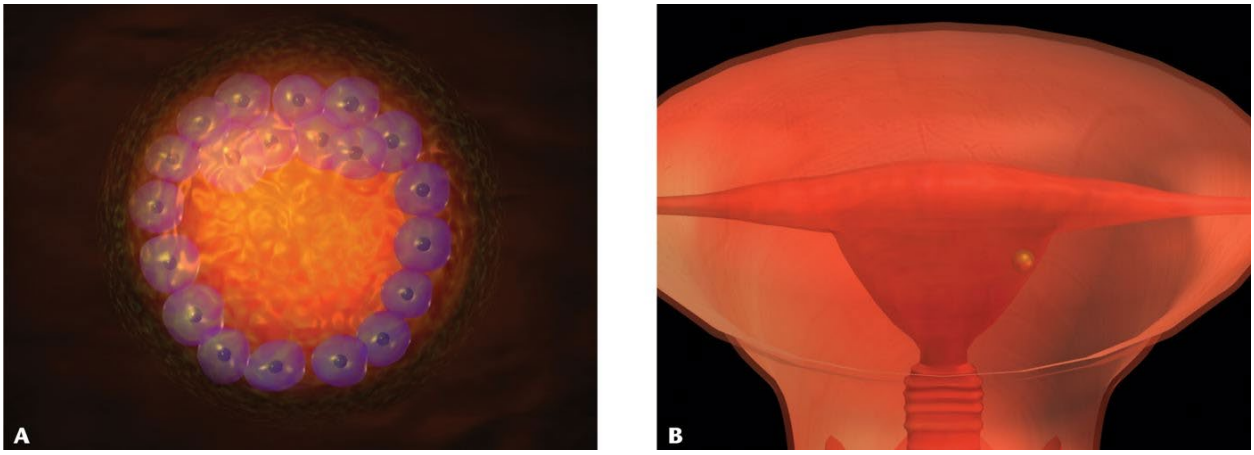


FIGURE 10.4

(**A**) Fertilized human egg (zygote) having reached the blastocyst stage. Zygote contains 20 to 30 eggs and a fluid-filled blastocele is beginning to form. (**B**) Implantation. Stylized image showing a frontal view of a uterus with a blastocyst about to implant into endometrium of the uterus. (Images from LifeART image copyright (c) 2011 Lippincott Williams & Wilkins. All rights reserved.)

At this time, the developing blastocyst needs more food and oxygen to keep growing. The trophoblast attaches itself to the surface of the endometrium for further nourishment. Normally, implantation occurs in the upper uterus (fundus), where a rich blood supply is available. This area also contains strong muscular fibers, which clamp down on blood vessels after the placenta separates from the inner wall of the uterus. Additionally, the lining is thickest here so the placenta cannot attach so strongly that it remains attached after birth (Moore, Persaud, & Torchia, 2011). **Figure 10.5** shows the process of fertilization and implantation.

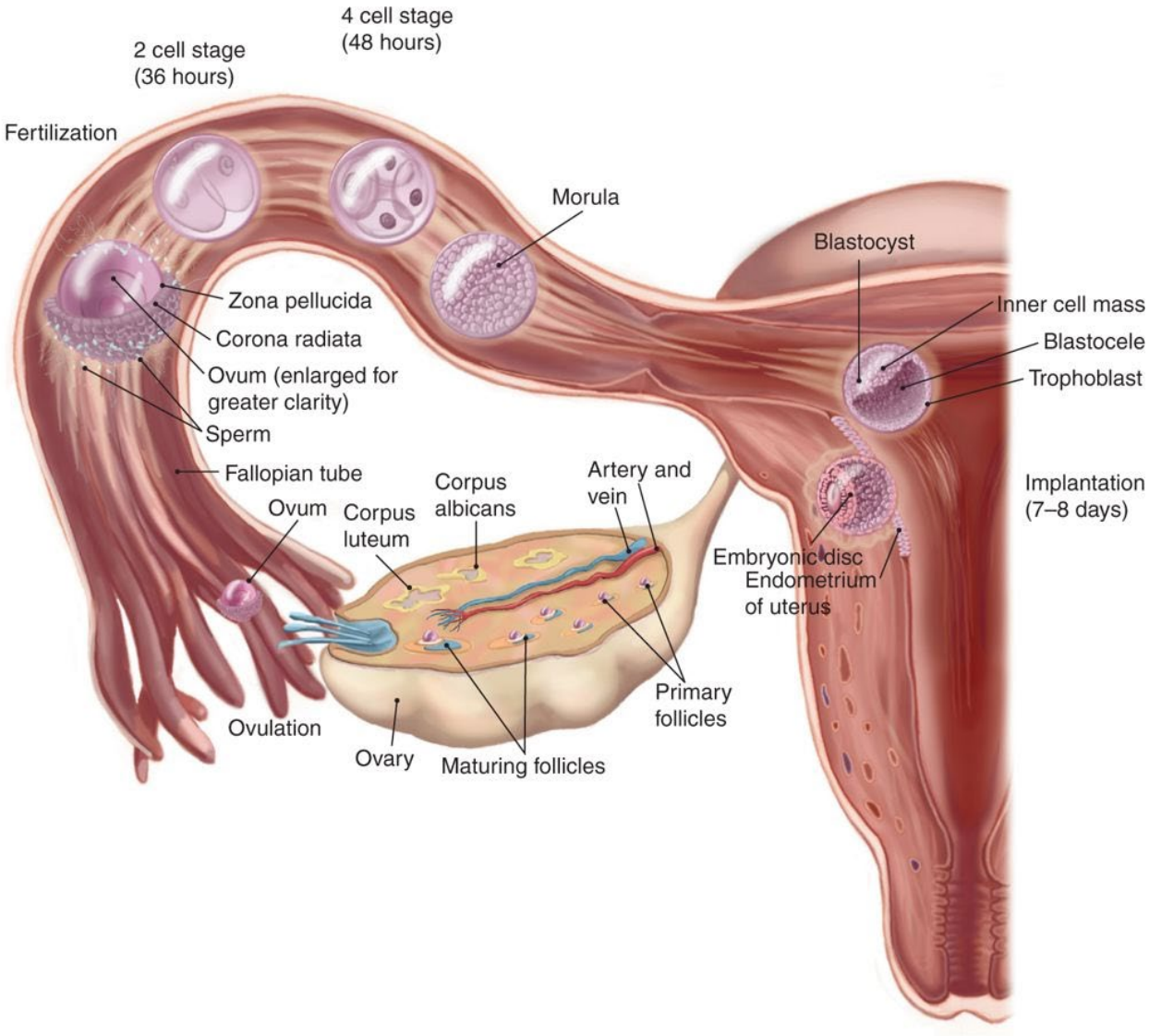


FIGURE 10.5

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.

Concurrent with the development of the trophoblast and implantation, further differentiation of the inner cell mass occurs. Some of the cells become the embryo itself, and others give rise to the membranes that surround and protect it. The three embryonic layers of cells formed are:

- 1. *Ectoderm:* forms the central nervous system, special senses, skin, and glands
- 2. *Mesoderm:* forms the skeletal, urinary, circulatory, and reproductive organs
- 3. *Endoderm:* forms the 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 (Carcio & Secor, 2010). **Box 10.1** summarizes preembryonic development.

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 a *blastocyst,* which forms the embryo and amnion.
- The outer cell mass is called a *trophoblast,* which forms the placenta and chorion.
- Implantation occurs 7 to 10 days after conception in the endometrium.

Despite the intense and dramatic activities going on internally to create a human life, many women are unaware that pregnancy has begun. Several weeks will pass before even one of the presumptive signs of pregnancy—missing the first menstrual period—will take place.

Embryonic Stage

The embryonic stage of development begins at day 15 after conception and continues through week 8. Basic structures of all major body organs and the main external features are completed during this time period, including internal organs. Table 10.1 and Fig. 10.6 summarize embryonic development.

TABLE 10.1: EMBRYONIC AND FETAL DEVELOPMENT

WEEK 3

Beginning development of brain, spinal cord, and heart

Beginning development of the gastrointestinal tract

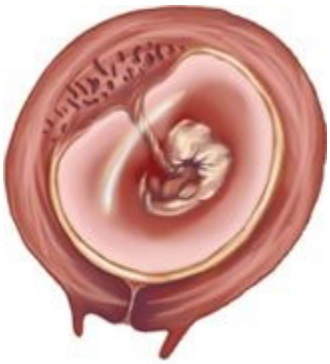
Neural tube forms, which later becomes the spinal cord

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

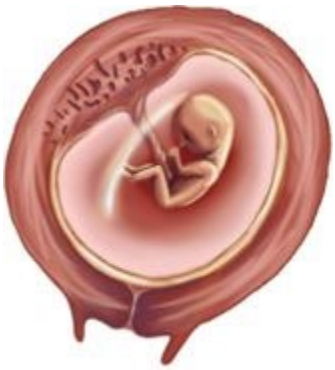
WEEK 8

Rotation of intestines

Facial features continue to develop

Heart development complete

Resembles a human being



8 weeks

WEEKS 9–12

Sexual differentiation continues

Buds for all 20 temporary teeth laid down

Digestive system shows activity

Head makes up nearly half the fetus size

Face and neck are well formed

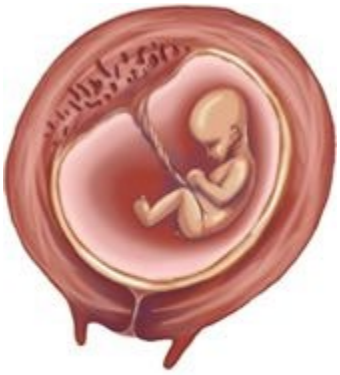
Urogenital tract completes development

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 called *lanugo* develops on the head

Fetal skin is almost transparent

Bones become harder

Fetus makes active movement

Sucking motions are made with the mouth

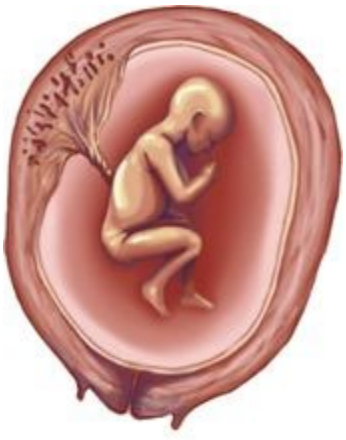
Amniotic fluid is swallowed

External genitalia are recognizable

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

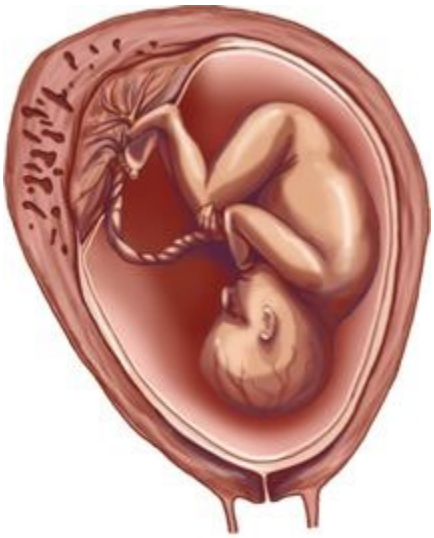
Vernix caseosa, a white greasy film, covers the fetus

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

WEEKS 21–24

Eyebrows and eyelashes are well formed

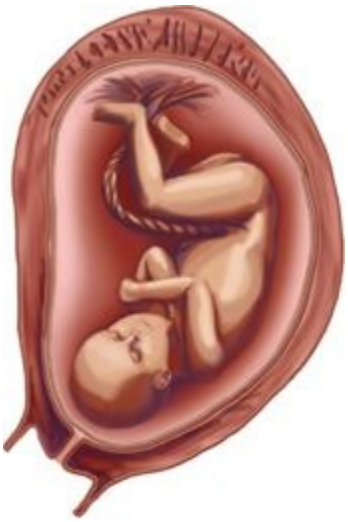
Fetus has a hand grasp and startle reflex

Alveoli forming in lungs

Skin is translucent and red

Eyelids remain sealed

Lungs begin to produce *surfactant*



25 weeks

WEEKS 25–28

Fetus reaches a length of 15 inches

Rapid brain development

Eyelids open and close

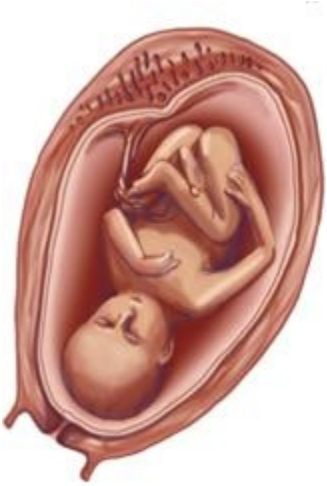
Nervous system controls some functions

Fingerprints are set

Subcutaneous fat is visible under the skin

Blood formation shifts from spleen to bone marrow

Fetus usually assumes head-down position



28 weeks

WEEKS 29–32

Rapid increase in the amount of body fat

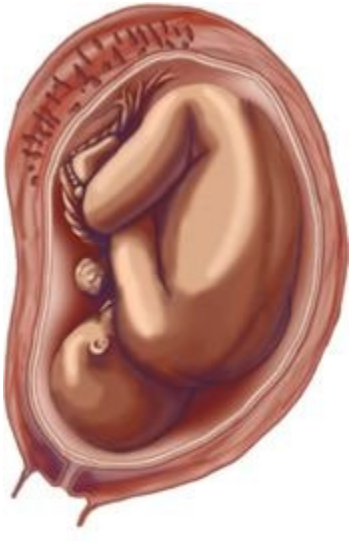
Increased central nervous system control over body functions

Rhythmic breathing movements occur

Lungs are not fully mature

Pupillary light reflex is present

Fetus stores iron, calcium, and phosphorus



32 weeks

WEEKS 33–38

Testes are in scrotum of male fetus

Lanugo begins to disappear

Has strong hand grasp reflex

Increase in body fat

Earlobes formed and firm

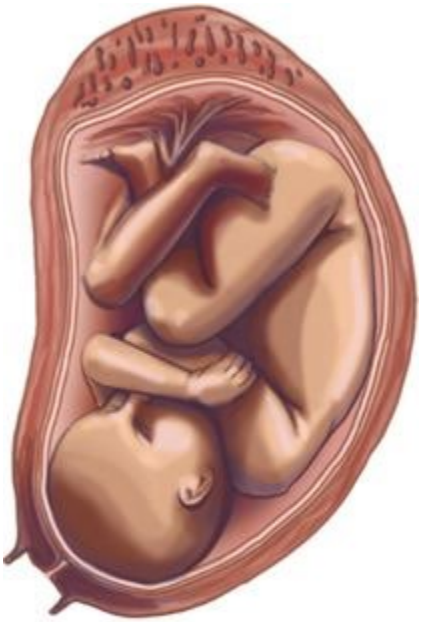
Fingernails reach the end of fingertips

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



37 weeks

Adapted from March of Dimes. (2012d). *How your baby grows*. Retrieved from **http://www.marchofdimes.com/pregnancy/yourbody_babygrowth.html**; Mattson, S., & Smith, J. E. (2010). *Core curriculum for maternal-newborn nursing* (4th ed.), St. Louis, MO: Saunders Elsevier; and Moore, K. L., Persaud, T. V. N., & Torchia, M. G. (2011). *The developing human* (9th ed.), Philadelphia, PA: Saunders Elsevier.

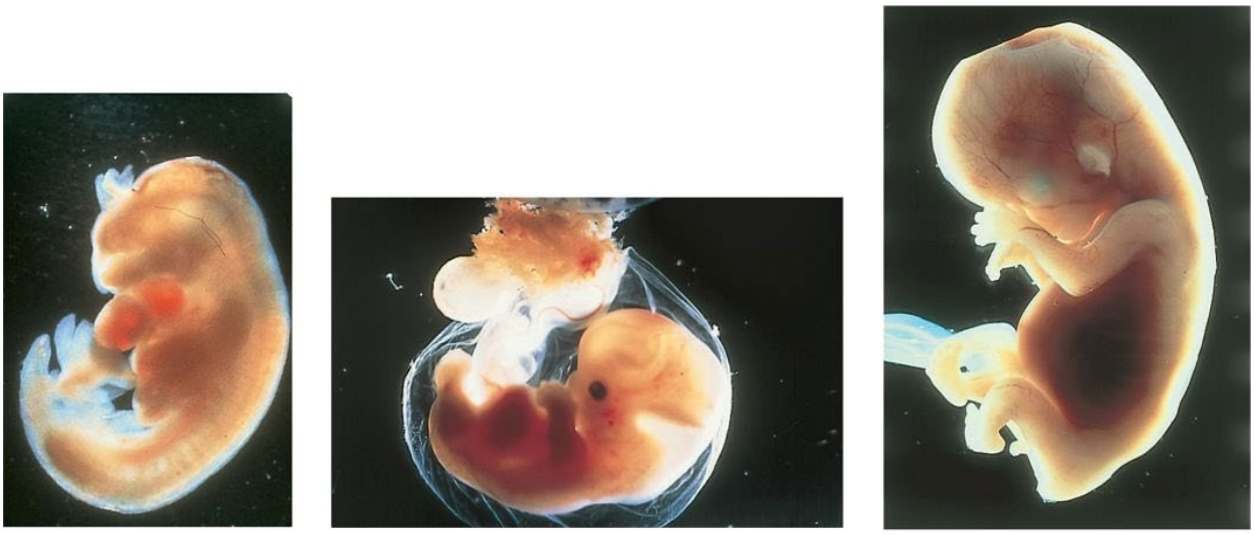


FIGURE 10.6

Embryonic development. (**A**) Four-week embryo. (**B**) Five-week embryo. (**C**) Six-week embryo.

The embryonic membranes (**Fig. 10.7**) begin to form around the time of implantation. The chorion consists of trophoblast cells and a mesodermal lining. The chorion 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. Alongside of the amnion, a yoke sac develops as a second cavity about day 8 or 9 after conception. The yoke sac aids in transferring maternal nutrients and oxygen to the embryo during the second and third weeks of gestation when development of the uteroplacental circulation is under way. As the pregnancy progresses, the yoke sac atrophies and is incorporated into the umbilical cord. 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 (Polin, Fox, & Abman, 2011).

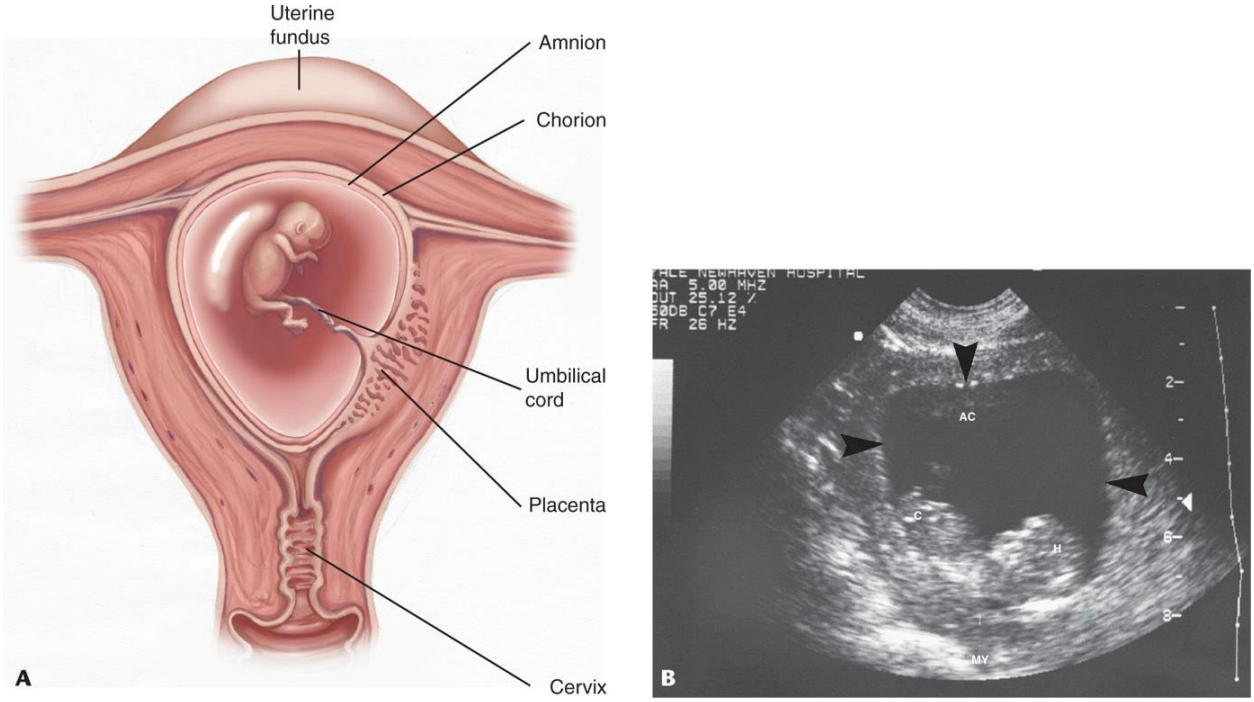


FIGURE 10.7

(**A**) The embryo is floating in amniotic fluid, surrounded by the protective fetal membranes (amnion and chorion). (**B**) Longitudinal sonogram of a pregnant uterus at 11 weeks showing the intrauterine gestational sac (*black arrowheads*) and the amniotic cavity (AC) filled with amniotic fluid; the fetus is seen in longitudinal section with the head (H) and coccyx (C) well displayed. The myometrium (MY) of the uterus can be identified. *(*Figure B is courtesy of L Scoutt.*)*

Amniotic fluid surrounds the embryo and increases in volume as the pregnancy progresses, reaching approximately a liter at term. Amniotic fluid is derived from two sources: fluid transported from the maternal blood across the amnion and fetal urine. Its volume changes constantly as the fetus swallows and voids. 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 from compression, and promote fetal movement to enhance musculoskeletal development. Amniotic fluid is composed 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. Amniotic fluid is essential for fetal growth and development, especially fetal lung development. It is dynamic, constantly changing as the fluid moves back and forth across the placental membrane (Polin et al., 2011).

The volume of amniotic fluid is important in determining fetal well-being. It gradually fluctuates throughout the pregnancy. The rate of change of amniotic fluid volume depends on the gestational age. During the fetal stage, the increase is 10 mL/week, and it increases to 50 to 60 mL/week at 19 to 25 weeks' gestation. The volume is at a maximum at 34 weeks' gestation before it undergoes a gradual decrease by term. Alterations in amniotic fluid volume can be associated with problems in the fetus. Too little amniotic

fluid (<500 mL at term), termed *oligohydramnios,* is associated with uteroplacental insufficiency and fetal renal abnormalities. Too much amniotic fluid (>2,000 mL at term), termed *hydramnios,* is associated with maternal 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 (Blackburn, 2012).

While 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 cut off fetal blood and nutrient supply. At term, the average umbilical cord is 22 inches long and about an inch wide (Stables & Randkin, 2010).

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 of the cells that form the **placenta**. When fully developed, the placenta serves as the interface between the mother and the developing fetus. Most commonly, the placenta develops at the uterine fundus. As early as 3 days after conception, the trophoblasts make human chorionic gonadotropin (hCG), a hormone that 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 nutrients and oxygen needed for 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 ready fetal organs for life outside the uterus (Pasca & Penn, 2010).

Theoretically, at no time during pregnancy does the mother's blood mix with fetal blood because there is no direct contact between their bloods; layers of fetal tissue always 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 to the placenta, which in turn provides nutrients to the developing fetus; the mother's uterine veins carry 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:

- *hCG:* preserves the corpus luteum and its progesterone production so that the endometrial lining of the uterus is maintained; this is the basis for pregnancy tests.
- *Human placental lactogen (hPL) or human chorionic somatomammotropin (hCS):* modulates fetal and maternal metabolism, participates in the development of maternal breasts for lactation, and decreases maternal glucose utilization, which increases glucose availability to the fetus.
- *Estrogen (estriol):* causes enlargement of a woman's breasts, uterus, and external genitalia; stimulates myometrial contractility.
- *Progesterone (progestin):* maintains the endometrium, decreases the contractility of the uterus, stimulates maternal metabolism and breast development, provides nourishment for the early conceptus (the products of conception after fertilization in the early stages of growth and differentiation).

- *Relaxin:* acts synergistically with progesterone to maintain pregnancy, causes relaxation of the pelvic ligaments, softens the cervix in preparation for birth (Lloyd, 2010).

The placenta acts as a pass-through between the mother and fetus, not a barrier. 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 use drugs, alcohol, and tobacco, because they can be harmful to the conceptus.

During the embryonic stage, the conceptus grows rapidly as all organs and structures are forming. During this critical period of differentiation 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 the time from the end of the eighth week until birth. It is the longest period of prenatal development. During this stage, the embryo is mature enough 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 (see **Table 10.1**). **Figure 10.8** depicts a 12- to 15-week-old fetus.

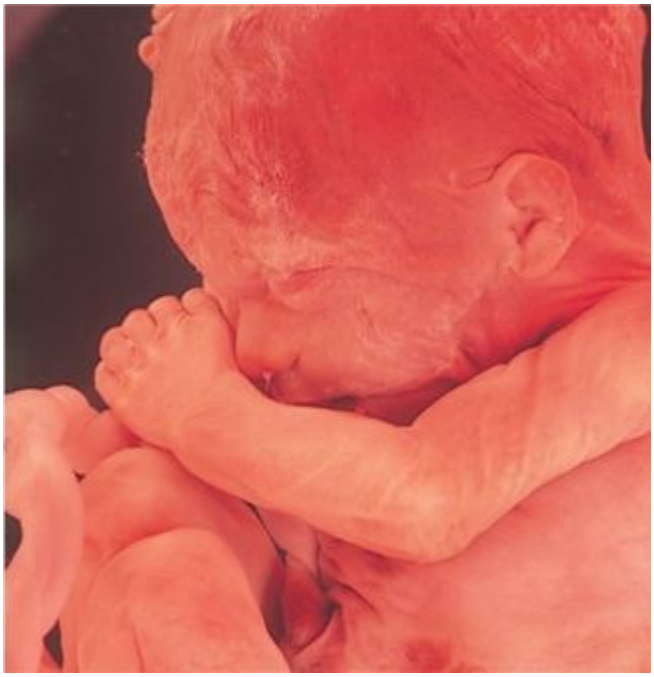


FIGURE 10.8

Fetal development: 12- to 15-week fetus.

Fetal Circulation

The circulation through the fetus during uterine life differs from that of a child or an adult. In the extrauterine world, oxygenation occurs in the lungs and oxygenated blood returns via the pulmonary veins to the left side of the heart to be ejected by the left ventricle into the systemic circulation. In contrast, fetal circulation oxygenation occurs in the placenta, and the fetal lungs are nonfunctional as far as the transfer of oxygen and carbon dioxide is concerned. For oxygenated blood derived from the placenta to reach the fetus's systemic circulation, it has to travel through a series of shunts to accomplish this.

Thus, fetal circulation involves the circulation of blood from the placenta to and through the fetus, and back to the placenta. A properly functioning fetal circulation system is essential 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 grows, its nutrient needs increase and the amount of tissue easily reached by diffusion increases. Thus, the circulation must develop quickly and accurately (Polin et al., 2011).

Three shunts also are present during 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

Take Note!

Fetal circulation functions to carry highly oxygenated blood to vital areas (e.g., heart, brain) while first shunting it away from less important ones (e.g., lungs, liver). The placenta essentially takes over the functions of the lungs and liver during fetal life. As a result, large volumes of oxygenated blood are not needed.

The blood with the highest oxygen content is delivered to the fetal heart, head, neck, and upper limbs, while the blood with the lowest oxygen content is shunted toward the placenta.

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 the brain to receive the blood with the highest level of oxygenation.

Deoxygenated blood from the superior vena cava flows into the right atrium, the right ventricle, and then the pulmonary artery. Because of high pulmonary vascular resistance,

only a small percentage (5% to 10%) of the blood in the pulmonary artery flows to the lungs; the majority is shunted through the patent ductus arteriosus and then to the descending aorta (Stables & Rankin, 2010). The fetal lungs are essentially nonfunctional because they are filled with fluid, making them resistant to incoming blood flow. They receive only enough blood 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 umbilical vessels are no longer needed. With the newborn's first breath, the lungs inflate, which leads to an increase in blood flow to the lungs from the right ventricle. This increase raises the pressure in the left atrium, causing a one-way flap on the left side of the foramen ovale, called the septum primum, to press against the opening, creating a functional separation between the two atria. Blood flow to the lungs increases because blood entering the right atrium can no longer by-pass the right ventricle. As a result, the right ventricle pumps blood into the pulmonary artery and on to the lungs. Typically the foramen ovale is functionally closed within 1 to 2 hours after birth. It is physiologically closed by 1 month by deposits of fibrin that seal the shunt. Permanent closure occurs by the sixth month of life.

The ductus venosus, which links the inferior vena cava with the umbilical vein, usually closes with the clamping of the umbilical cord and inhibition of blood flow through the umbilical vein. This fetal structure closes by the end of the first week. The ductus arteriosus constricts partly in response to the higher arterial oxygen levels that occur after the first few breaths. This closure prevents blood from the aorta from entering the pulmonary artery. Functional closure of the ductus arteriosus in a term infant usually occurs within the first 72 hours after birth. Permanent closure occurs at 3 to 4 weeks of age (Blackburn, 2012). Frequently a functional or innocent murmur is auscultated by the nursery nurse when there are delayed fetal shunt closures, but they usually are not associated with a heart lesion (Verklan & Walden, 2010). All of these changes at birth leave the newborn with the typical adult pattern of circulation. **Figure 10.9** shows fetal circulation.

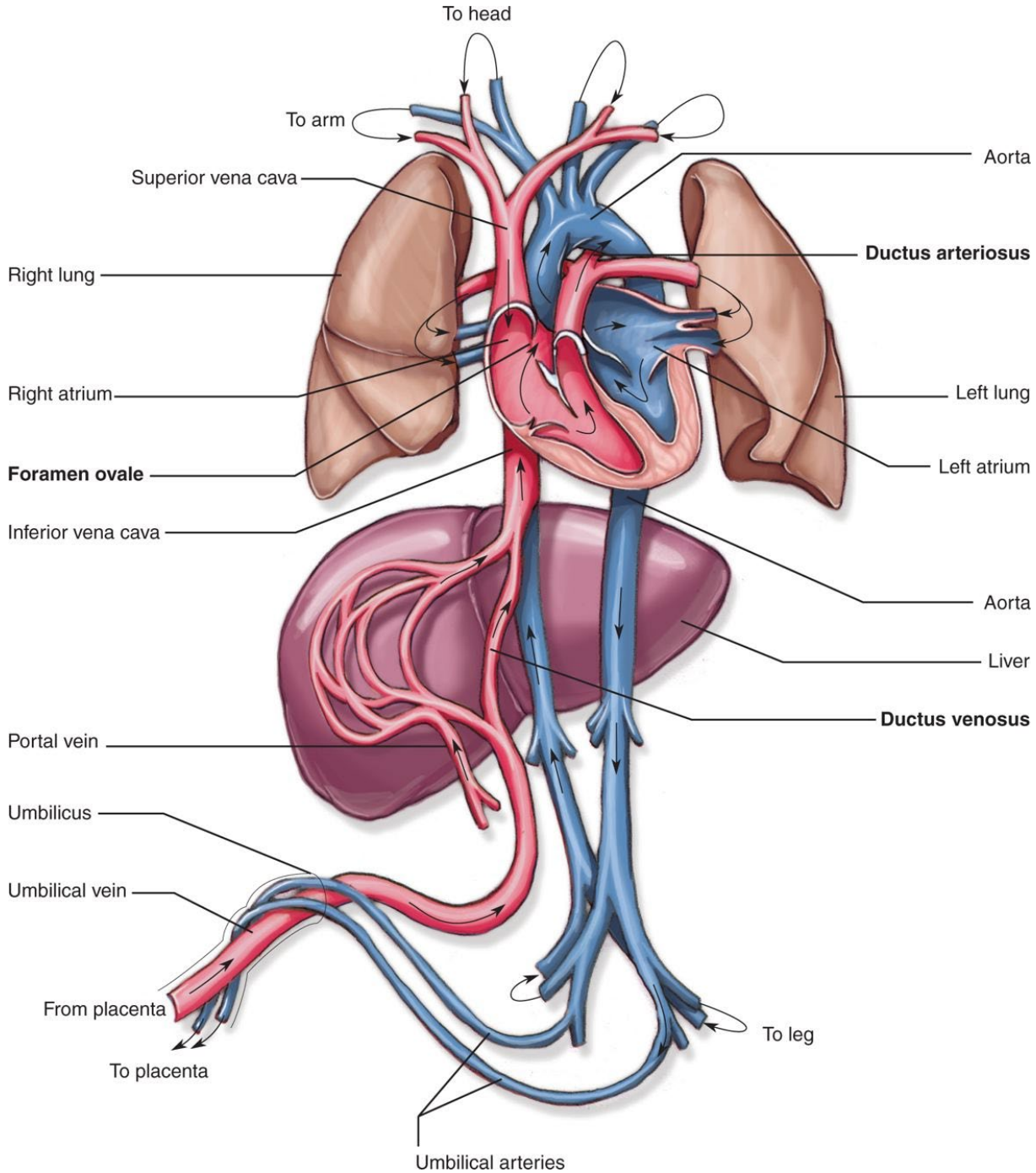


FIGURE 10.9

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 nonfunctioning lungs, while the remaining blood is shunted through the ductus arteriosus into the aorta to supply the rest of the body.

GENETICS

Genetics is the study of individual genes and their role in heritance (Gregg & Simpson, 2010). **Genomics**, a relatively new science, is the study of all genes and includes interactions among genes as well as interactions between genes and the environment. Genomics plays a role in complex conditions such as heart disease and diabetes. Another emerging area of research is that of pharmacogenomics, the study of genetic and genomic influences on pharmacodynamics and pharmacotherapeutics (J. Lewis, 2011).

According to the Centers for Disease Control and Prevention (2011), birth defects and genetic disorders occur in 1 in 33 infants born in the United States and cause 1 in 5 infant deaths. Traditionally, genetics has been associated with making decisions about childbearing and caring for children with genetic disorders. Currently, genetic and technological advances are expanding our understanding of how genetic changes affect human diseases such as diabetes, cancer, Alzheimer's disease, and other multifactorial diseases that are prevalent in adults (Jorde, Carey, & Bamshad, 2010). Newborn screening is perhaps the most widely used application of genetics in perinatal and neonatal care. Our ability to diagnose genetic conditions is more advanced than our ability to cure or treat the disorders. However, accurate diagnosis has led to improved treatment and outcomes for those affected with these disorders.

Take Note!

Genetic science has the potential to revolutionize health care with regard to national screening programs, predisposition testing, detection of genetic disorders, and pharmacogenetics.

Genetics has been a part of perinatal care for decades. Ultrasounds and maternal serum screening have become routine elements of prenatal care. Preconception carrier screening for conditions such as Tay-Sachs disease has been in place among high-risk populations such as Ashkenazi Jews. Amniocentesis and chorionic villus sampling are diagnostic tests that may confirm a genetic anomaly in a developing fetus. Fetal nuchal translucency, as seen on ultrasound, is suggestive of the presence of trisomy 21 or Down syndrome (R. Lewis, 2011).

Today, nurses are required to have basic skills and knowledge in genetics, genetic testing, and genetic counseling so they can assume new roles and provide information and support to women and their families. Roles for maternity nurses in genetic health care have expanded significantly as genetics education and counseling have become a standard of care. Today, nurses may provide preconception counseling for women at risk for the transmission of a genetic disorder. In addition, they may provide prenatal care for women with genetically linked disorders that require specialized care or may participate in screening infants for birth defects and genetic disorders (Beery & Workman, 2011). Nurses

at all levels should participate in risk assessments for genetic conditions and disorders, explaining genetic risk and genetic testing, and supporting informed health decisions and opportunities for early intervention (American Nurses Association, 2009).

It is very clear that genomics will have a profound effect on health and illness at all levels. In the future, as the era of personalized health care moves forward, nurses will be responsible for ensuring that the scientific principles, ethical standards, and professional accountability of genetics and genomics practice are integrated into nursing practice. Nurses are increasingly doing this as they gain the necessary knowledge and skills. The strength of the nursing voice in genetics and genomic research will be the link to the bedside and the commitment to ensure that new knowledge is translated into evidence-based client care (J. Lewis, 2011).

Advances in Genetics

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 very early in pregnancy (see **Evidence-Based Practice 10.1**). 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, diabetes, HIV, and hepatitis (Tamura et al., 2010). The potential exists for creation of increased intelligence and size through genetic intervention. Recent research using gene therapy shows promise for the generation of insulin-producing cells to cure diabetes (Calne, Gan, & Lee, 2010). In the future, genetic agents may replace drugs, general surgery may be replaced by gene surgery, and genetic intervention may replace radiation. Recent successful trials on the treatment of ocular diseases and inherited immune deficiencies are particularly encouraging and have raised hopes that human gene therapy as a standard treatment option will finally become a reality. Continuous progress suggests that a wide range of diseases will be treated with gene therapy in the future (Herzog, Cao, & Srivastava, 2010).

EVIDENCE-BASED PRACTICE 10.1: DIFFERENT COMMUNICATION STRATEGIES FOR DISCLOSING RESULTS OF DIAGNOSTIC PRENATAL TESTING

Most parents want reassurance that their baby is all right genetically. Any screening program aimed at reassuring pregnant women that their unborn baby is healthy will cause anxiety while waiting for the test results. This study addressed three objectives: (1) to determine if revealing amniocentesis or chorionic villous sampling (CVS) results on a fixed date alters maternal anxiety during the waiting period, compared with a policy of revealing the result "when available" (i.e., variable date); (2) to evaluate whether issuing early results from a rapid molecular test alters maternal anxiety during the waiting period; and (3) to evaluate whether different methods of communication (telephone, fax, e-mail, face to face) have any impact on the parents' satisfaction and anxiety levels.

STUDY

Two studies (involving 286 women) from amniocentesis (but none from CVS) compared the impact of communicating results of rapid testing with waiting for definitive karyotyping. Unfortunately, it was not possible to perform a pooled analysis because one study reported only median (interquartile range) data, presumably because the data were not normally distributed. One study reported a statistically significant reduction in the average anxiety during the waiting period for women who had had a rapid test compared with those who had not (mean difference −2.30; 95% confidence intervals −3.08 to −1.52). The other study compared median (interquartile range) for the trait- and state-anxiety scores and found no difference between the two groups.

Findings

This study found no conclusive evidence that, while waiting for the full karyotype following amniocentesis, issuing results from a rapid analysis reduces maternal anxiety. The limited evidence from the two trials included in this review does not help resolve the dilemma about whether full karyotyping should be abandoned in favor of limited rapid testing for women undergoing Down syndrome screening. This choice will rest on clinical arguments and cost effectiveness rather than impact on anxiety. There is also no evidence to support the view that issuing amniocentesis results as soon as they are available is more user friendly than using a predefined fixed date. Studies evaluating the effect of different strategies for disclosing results on women anxiety for CVS are needed.

Nursing Implications

Although the study failed to find evidence to support the objectives, it does point out the anxiety that couples endure while they are waiting for their prenatal test results. This knowledge should help nurses to support couples during their waiting period for their prenatal screening results. Nurses need to maintain a supportive attitude toward the family's questions, need for clarification, and patience as they wait. Encouraging the couple to express their anxieties can be therapeutic and the nurse can be an active listener for them. Advances in newborn screening are expected to continue to expand in the future, and it will be important for nurses to be aware of the biochemical and genetic bases for these screening tests to be able to offer an explanation to couples awaiting results and/or reassurances.

Adapted from Mujezinovic, F., Prosnik, A., & Alfirevic, Z. (2010). Different communication strategies for disclosing results of diagnostic prenatal testing. *Cochrane Database of Systematic Reviews, 2010*(11). doi:10.1002/14651858.CD007750.pub2.

The Human Genome Project (HGP) was an international 13-year effort to produce a comprehensive sequence of the human genome. It was started in 1990 by the Department of Energy and the National Institutes of Health and was completed in May 2003. The goals of the

Human Genome Project were to map, sequence, and determine the function of all human genes, which led to advances in the field of genetics and genetic testing (Alberts, 2011). An individual's **genome** represents his or her genetic blueprint, which determines **genotype** (the gene pairs inherited from parents; the specific genetic makeup) and **phenotype** (observed outward characteristics of an individual) (Lea, Skirton, Read, & Williams, 2011).

A primary goal of the HGP was to translate the findings into new strategies for the prevention, diagnosis, and treatment of genetic diseases and disorders. Two key findings from the project were that all human beings are 99.9% identical at the DNA level, and approximately 30,000 genes make up the human genome (International Human Genome Sequencing Consortium,

2009). Refer to for more information about the HGP.

Current and potential applications for the HGP in health care include rapid and more specific diagnosis of disease, with hundreds of genetic tests available in research or clinical practice; earlier detection of genetic predisposition to disease; less emphasis on treating the symptoms of a disease and more emphasis on looking at the fundamental causes of the disease; new classes of drugs; avoidance of environmental conditions that may trigger disease; and augmentation or replacement of defective genes through gene therapy. This new genetic knowledge and technology, along with the commercialization of this knowledge, will change both professional and parental understanding of genetic disorders.

The potential benefits of these discoveries are vast, but so is the potential for misuse. These advances challenge all health care professionals to consider the many ethical, legal, and social ramifications of genetics 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 client privacy and confidentiality related to workplace discrimination and access to health insurance. Issues of autonomy are equally problematic as society considers how to address the injustices that will inevitably surface when disease risk can be determined years before the disease occurs. Nurses will play an important role in developing policies and providing direction and support in this arena, and to do so they will need a basic understanding of genetics, including inheritance and inheritance patterns. (For more information on the ethical, social, and legal issues surrounding

human genetic research and advances, refer to .)

Inheritance

The nucleus within the cell is the controlling factor in all cellular activities because it contains chromosomes, long continuous strands of deoxyribonucleic acid (DNA) that carry genetic information. Each chromosome is made up of **genes**. Genes are individual units of heredity of all traits and are organized into long segments of DNA that occupy a specific location on a chromosome and determine a particular characteristic in an organism.

DNA stores genetic information and encodes the instructions for synthesizing specific proteins needed to maintain life. DNA is double stranded and takes the form of 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 (Rand, 2010) (**Fig. 10.10**).

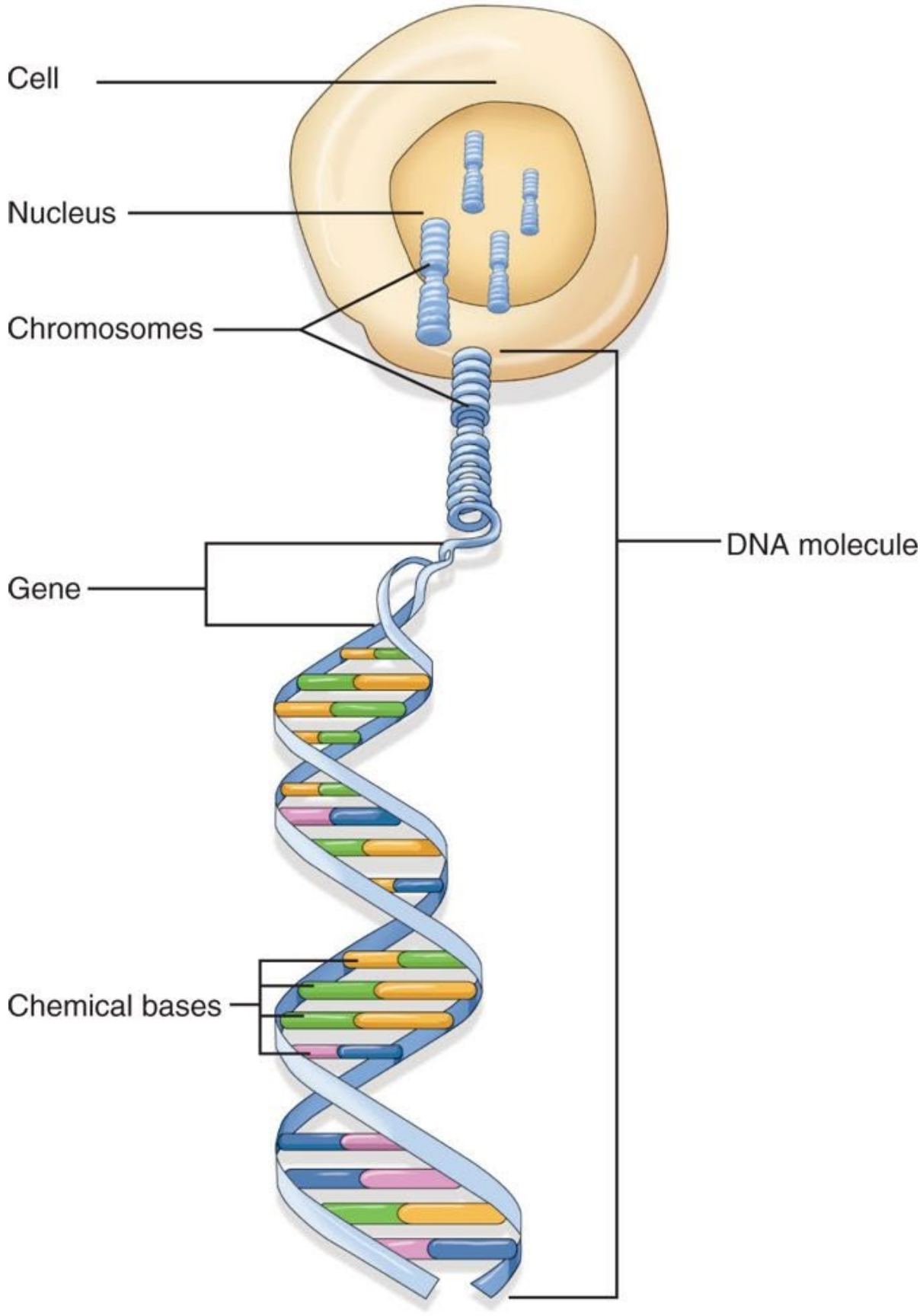

Cell
Nucleus
Chromosomes
DNA molecule
Gene
Chemical bases

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 Institutes of Health and National Cancer Institute. [1995]. *Understanding gene testing* [NIH Pub. No. 96-3905]. Washington, DC: U.S. Department of Human Services.)

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 (Jorde et al., 2010). Any change in gene structure or location leads to a **mutation**, 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 and lined up in a specific linear formation along a chromosome.

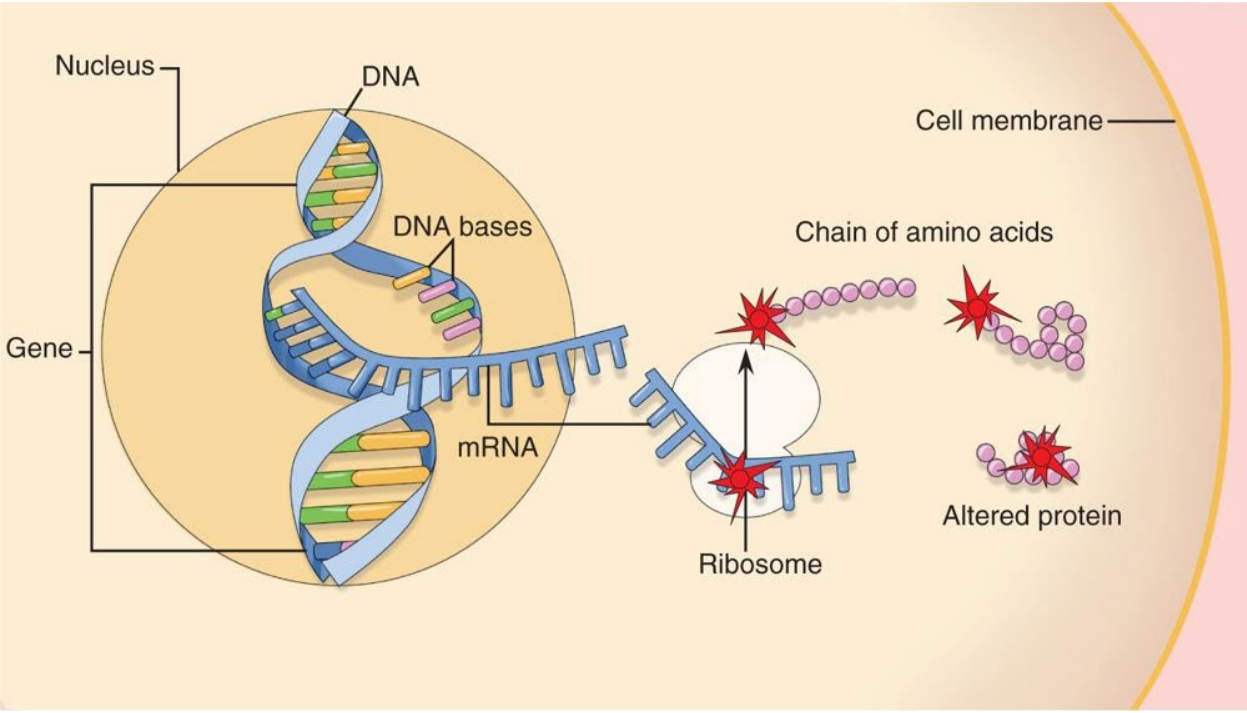


FIGURE 10.11

When a gene contains a mutation, the protein encoded by that gene will be abnormal. Some protein changes are insignificant, while 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.)

The genotype, the specific genetic makeup of an individual, usually in the form of DNA, is the internally coded inheritable information. It refers to the particular **allele**, which is one of two or more alternative versions of a gene at a given position or locus on a chromosome

that imparts the same characteristic of that gene. For instance, each human has a gene that controls height, but there are variations of these genes, called alleles, that code for a specific height. As another example, a gene that controls eye color may have an allele that can produce blue eyes or an allele that produces brown eyes. The genotype, together with environmental variation that influences the individual, determines the phenotype, or the observed, outward characteristics of an individual. A human inherits two genes, one from each parent. Therefore, one allele comes from the mother and one from the father. These alleles may be the same for the characteristic (**homozygous**) or different (**heterozygous**). For example, WW stands for homozygous dominant; ww stands for homozygous recessive. Heterozygous would be indicated as Ww. If the two alleles differ, such as Ww, the dominant one will usually be expressed in the phenotype of the individual.

Human beings typically have 46 chromosomes. This includes 22 pairs of non–sex chromosomes or autosomes and 1 pair of 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.

Regulation and expression of the thousands of human genes are very complex processes and are 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 (Gregg & Simpson, 2010). Gene mutations are a permanent change in the sequence of DNA.

Gene mutations can be inherited, spontaneous, or acquired. Inherited gene mutations are passed on from parent to child in the egg and sperm, and are passed on to all cells in that child's body when the body cells reproduce. Cystic fibrosis is an example of an inherited mutation. A spontaneous mutation can occur in individual eggs or sperm at the time of conception. A person who has the new spontaneous mutation has the risk of passing it on to his or her offspring. An example of a spontaneous mutation would be Marfan syndrome. Acquired mutations occur in body cells other than egg or sperm. They involve changes in DNA that take place after conception, during a person's lifetime. Acquired mutations are passed on when they reproduce to daughter cells (Beery & Workman, 2011). Some mutations have no significant effect, whereas others can have a tremendous impact on the health of the individual. Several genetic disorders such as cancer, sickle cell disease, phenylketonuria, and hemophilia, can result from these mutations.

The pictorial analysis of the number, form, and size of an individual's chromosomes is termed the **karyotype**. This analysis commonly uses white blood cells and fetal cells in amniotic fluid. 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. **Figure 10.12** illustrates an example of a karyotyping pattern.

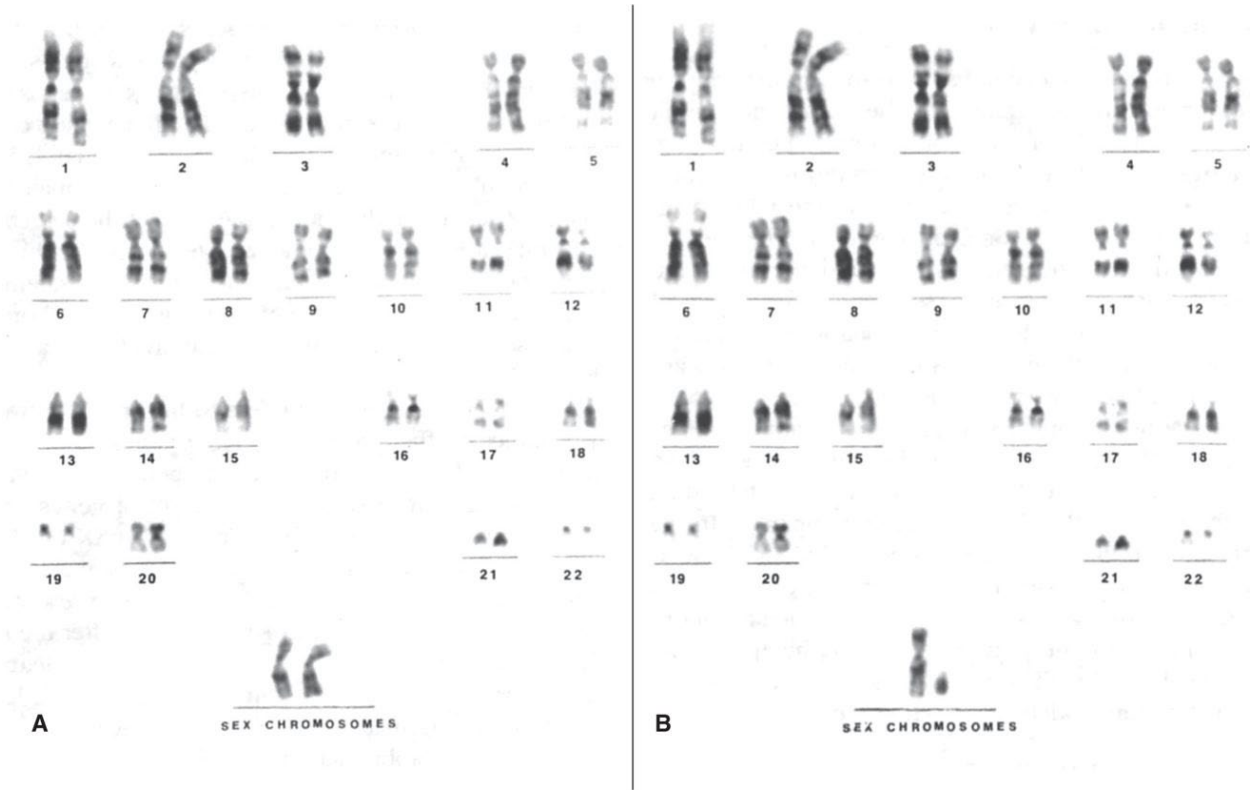


FIGURE 10.12

Karyotype pattern. (**A**) Normal female karyotype. (**B**) Normal male karyotype.

Patterns of Inheritance for Genetic Disorders

Patterns of inheritance demonstrate how a genetic disorder can be passed on to offspring. A genetic disorder is a disease caused by an abnormality in an individual's genetic material or genome. Diagnosis of a genetic disorder is usually based on clinical signs and symptoms or on laboratory confirmation of the presence of an altered gene associated with the disorder. Accurate diagnosis can be aided by recognition of the pattern of inheritance within a family. The pattern of inheritance is also vital to understand when teaching and counseling families about the risks in future pregnancies. Some genetic disorders occur in multiple family members, while others may occur in only a single family member. A genetic disorder is caused by completely or partially altered genetic material, whereas a familial disorder is more common in relatives of the affected individual but may be caused by environmental influences and not genetic alterations.

Monogenic Disorders

The principles of genetic disease inheritance of single-gene disorders are the same principles that govern the inheritance of other traits, such as eye and hair color. These are known as Mendel's

laws of inheritance, named for the genetic work of Gregor Mendel, an Austrian naturalist. These patterns occur due to a single gene being defective and are referred to as monogenic or sometimes Mendelian disorders. If the defect occurs on the autosome, the genetic disorder is termed *autosomal*; if the defect is on the X chromosome, the genetic disorder is termed *X linked*. The defect also can be classified as dominant or recessive. Monogenic disorders include autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive patterns.

AUTOSOMAL DOMINANT INHERITANCE DISORDERS

Autosomal dominant inherited disorders occur when a single gene in the heterozygous state is capable of producing the phenotype. In other words, the abnormal or mutant gene overshadows the normal gene and the individual will demonstrate signs and symptoms of the disorder. The affected person generally has an affected parent, and an affected person has a 50% chance of passing the abnormal gene to each of his or her children (**Fig. 10.13**). Affected individuals are present in every generation. Males and family members who are phenotypically normal (do not show signs or symptoms of the disorder) do not transmit the condition to their offspring. Females and males are equally affected and a male can pass the disorder on to his son. This male-to-male transmission is important in distinguishing autosomal dominant inheritance from X-linked inheritance. There are varying degrees of presentation among individuals in a family. Therefore, a parent with a mild form could have a child with a more severe form. Common types of genetic disorders that follow the autosomal dominant pattern of inheritance include neurofibromatosis (genetic disorders affecting the development and growth of neural cells and tissues), Huntington's disease (a genetic disorder affecting the nervous system characterized by abnormal involuntary movements and progressive dementia), achondroplasia (a genetic disorder resulting in disordered growth and abnormal body proportion), and polycystic kidney disease (a genetic disorder involving the growth of multiple, bilateral, grape-like clusters of fluid-filled cysts in the kidneys that eventually compress and replace functioning renal tissue).

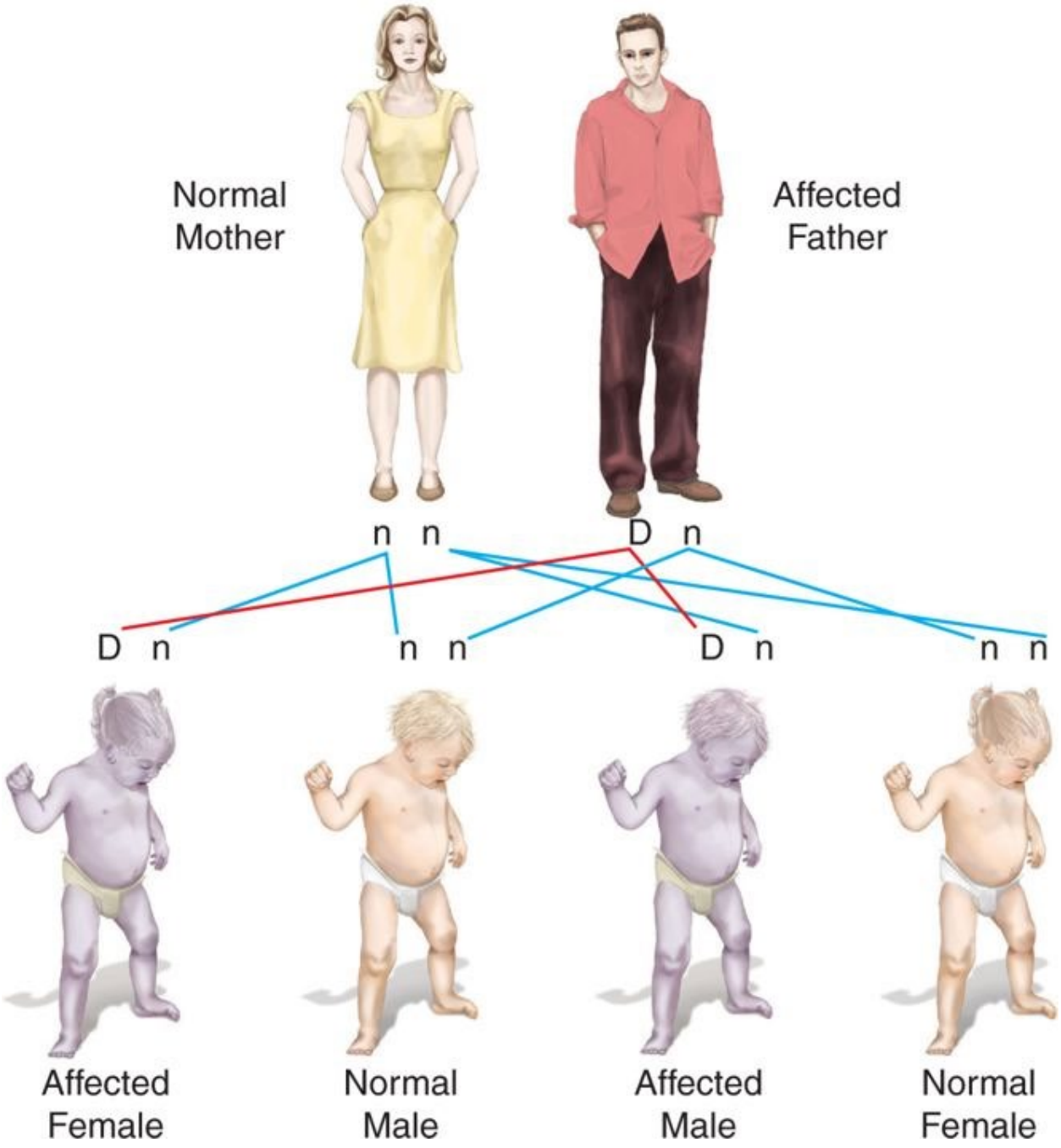


FIGURE 10.14

Autosomal recessive inheritance.

X-LINKED INHERITANCE DISORDERS

X-linked inheritance disorders are those associated with altered genes present on the X chromosome. They differ from autosomal disorders. If a male inherits an X-linked altered gene, he will express the condition. Because a male has only one X chromosome, all the genes on his X chromosome will be expressed (the Y chromosome carries no normal allele to compensate for the altered gene). Because females inherit two X chromosomes, they can be either heterozygous or homozygous for any allele. Therefore, X-linked disorders in females are expressed similarly to autosomal disorders.

Most X-linked disorders demonstrate a recessive pattern of inheritance. Males are more affected than females. A male has only one X chromosome and all the genes on his X chromosome will be expressed, whereas a female will usually need both X chromosomes to carry the disease. There is no male-to-male transmission (since no X chromosome from the male is transmitted to male offspring), but any man who is affected will have carrier daughters. If a woman is a carrier, there is a 50% chance that her sons will be affected and a 50% chance that her daughters will be carriers (**Fig. 10.15**). Common types of genetic disorders that follow X-linked recessive inheritance patterns include hemophilia (a genetic disorder involving a deficiency of one of the coagulation factors in the blood), color blindness, and Duchenne muscular dystrophy (a disorder involving progressive muscular weakness and wasting).

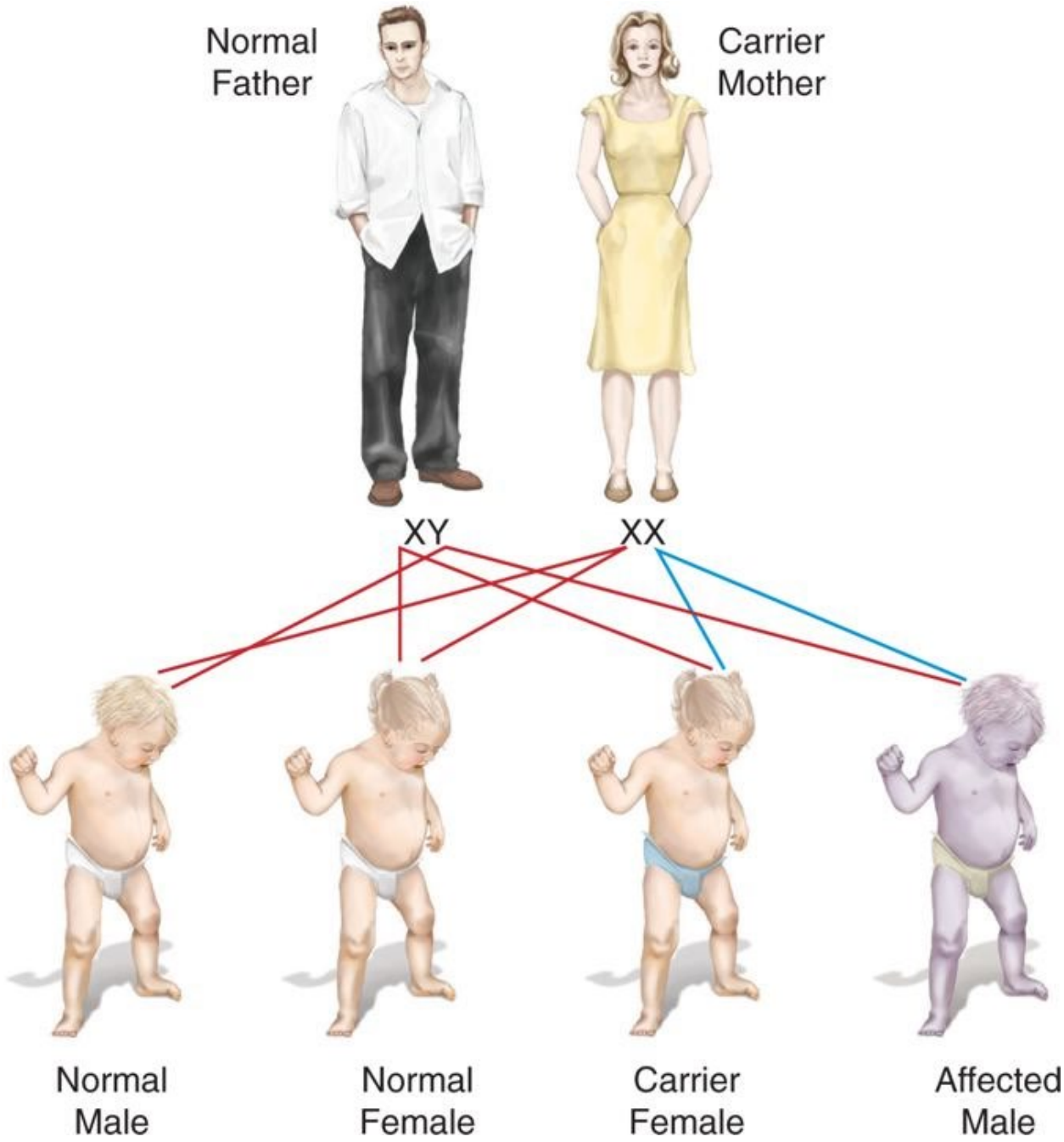


FIGURE 10.15

X-linked recessive inheritance.

X-linked dominant inheritance is present if heterozygous female carriers demonstrate signs and symptoms of the disorder. All of the daughters and none of the sons of an affected male have the condition, while both male and female offspring of an affected woman have a 50%

chance of inheriting and presenting with the condition (Fig. 10.16). X-linked dominant disorders are rare. The most common is hypophosphatemic (vitamin D–resistant) rickets (a disorder involving a softening or weakening of the bones).

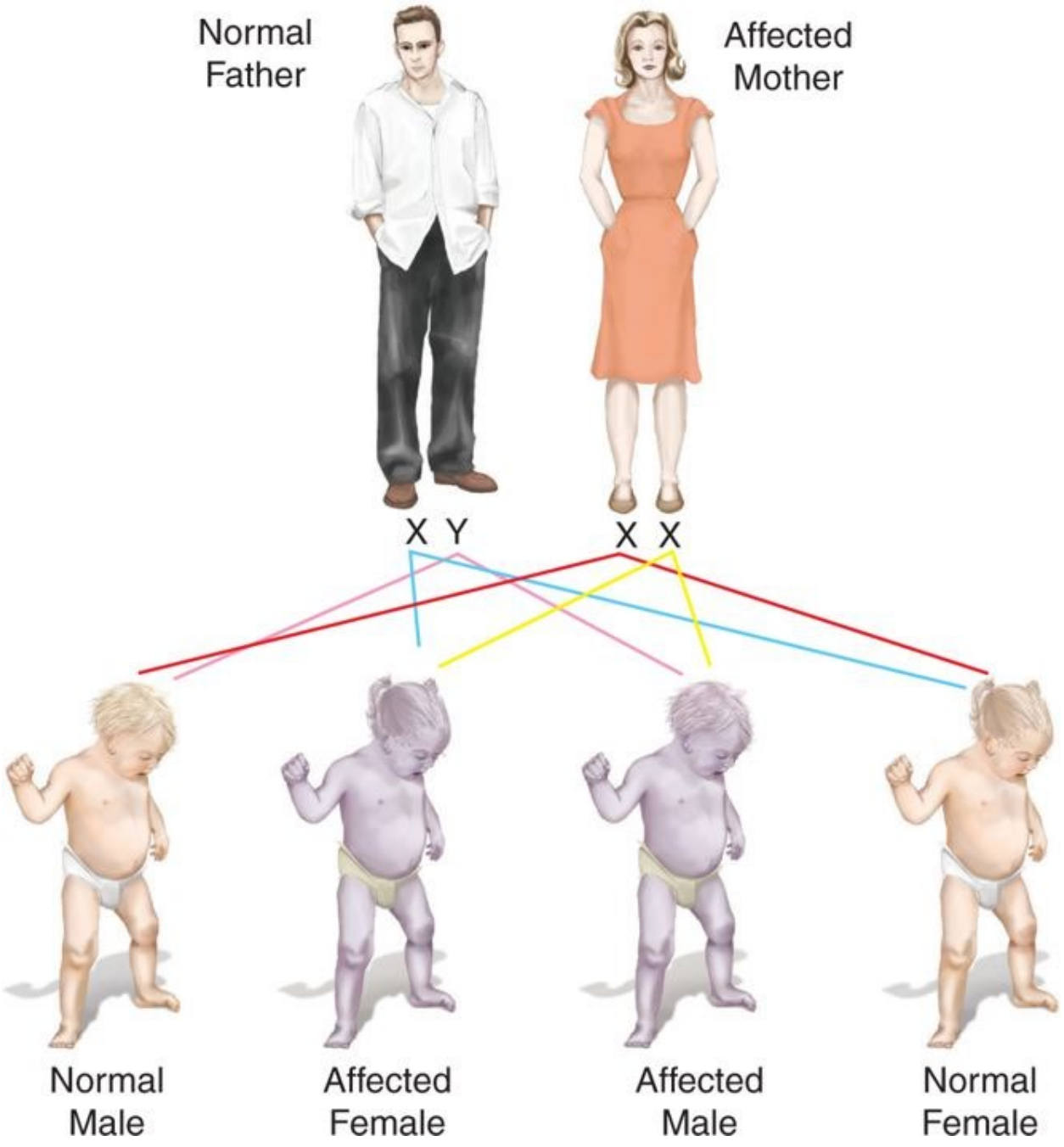


FIGURE 10.16

X-linked dominant inheritance.

Multifactorial Inheritance Disorders

Multifactorial inheritance disorders are thought to be caused by multiple genetic (polygenic) and environmental factors. Many of the common congenital malformations, such as cleft lip, cleft palate, spina bifida, pyloric stenosis, clubfoot, developmental hip dysplasia, and cardiac defects, are attributed to multifactorial inheritance. A combination of genes from both parents, along with unknown environmental factors, produces the trait or condition. An individual may inherit a predisposition to a particular anomaly or disease. The anomalies or diseases vary in severity, and often a sex bias is present. For example, pyloric stenosis is seen more often in males, while developmental hip dysplasia is much more likely to occur in females. Multifactorial conditions tend to run in families, but the pattern of inheritance is not as predictable as with single-gene disorders. The chance of recurrence is also lower than in single-gene disorders, but the degree of risk is related to the number of genes in common with the affected individual. The closer the degree of relationship, the more genes an individual has in common with the affected family member, resulting in a higher chance that the individual's offspring will have a similar defect. In multifactorial inheritance the likelihood that both identical twins will be affected is not 100%, indicating that there are nongenetic factors involved.

Nontraditional Inheritance Patterns

Molecular studies have revealed that some genetic disorders are inherited in ways that do not follow the typical patterns of dominant, recessive, X-linked, or multifactorial inheritance. Examples of nontraditional inheritance patterns include mitochondrial inheritance and genomic imprinting. As the science of molecular genetics advances and more is learned about inheritance patterns, other nontraditional patterns of inheritance may be discovered or found to be relatively common.

Chromosomal Abnormalities

In some cases of genetic disorders, the abnormality occurs due to problems with the chromosomes. Chromosomal abnormalities do not follow straightforward patterns of inheritance. Sperm and egg cells each have 23 unpaired chromosomes. When they unite during pregnancy they form a fertilized egg with 46 chromosomes. Sometimes before pregnancy begins, an error can occur during the process of cell division, leaving an egg or sperm with too many or too few chromosomes. If this egg or sperm cell joins with a normal egg or sperm cell, the resulting embryo has a chromosomal abnormality. Chromosomal abnormalities can also occur due to an error in the structure of the chromosome. Small pieces of the chromosome may be deleted, duplicated, inverted, misplaced, or exchanged with part of another chromosome. Most chromosomal abnormalities occur due to an error in the egg or sperm. Therefore, the abnormality is present in every cell of the body. However, some abnormalities can happen after fertilization during mitotic cell division and result in **mosaicism**. Mosaicism or mosaic form refers to a situation in which the chromosomal abnormalities do not show up in every cell and only some cells or tissues carry

the abnormality. In mosaic forms of the disorder, the symptoms are usually less severe than if all the cells were abnormal.

About 1 in 150 live-born infants is born with a chromosomal abnormality (March of Dimes, 2012b). These often cause major defects because they involve added or missing genes. Congenital anomalies and intellectual disability are often associated with chromosomal abnormalities. These abnormalities occur on autosomal as well as sex chromosomes and can result from changes in the number of chromosomes or changes in the structure of the chromosomes.

Numerical Abnormalities

Chromosomal abnormalities of number often result from nondisjunction, or failure of the chromosome pair to separate during cell division, meiosis, or mitosis. Few chromosomal numerical abnormalities are compatible with full-term development and most result in spontaneous abortion. One type of chromosomal number abnormality is **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.

Some numerical abnormalities do support development to term because the chromosome on which the abnormality is present carries relatively few genes (such as chromosome 13, 18, 21, or X). Two common abnormalities of chromosome number are monosomies or trisomies. In **monosomies** there is only one copy of a particular chromosome instead of the usual pair (an entire single chromosome is missing). In these cases, all fetuses spontaneously abort in early pregnancy. Survival is seen only in mosaic forms of these disorders. In **trisomies**, there are three of a particular chromosome instead of the usual two (an entire single chromosome is added). Trisomies may be present in every cell or may present in the mosaic form. The most common trisomies include trisomy 21 (Down syndrome), trisomy 18, and trisomy 13.

TRISOMY 21

Down syndrome is an example of a trisomy. The cause of Down syndrome is one of three types of abnormal cell division involving chromosome 21. All three abnormalities result in extra genetic material from chromosome 21, which is responsible for the characteristic features and developmental problems of Down syndrome. The three genetic variations that can cause Down syndrome include trisomy 21, where the infant has three copies of chromosome 21—instead of the usual two copies—in all of his or her cells; mosaic, where infants have some cells with an extra copy of chromosome 21; and translocation, where part of chromosome 21 becomes attached (translocated) to another chromosome, before or at conception. More than 90% of the cases of Down syndrome are caused by trisomy 21 (March of Dimes, 2012c) (**Fig. 10.17**).

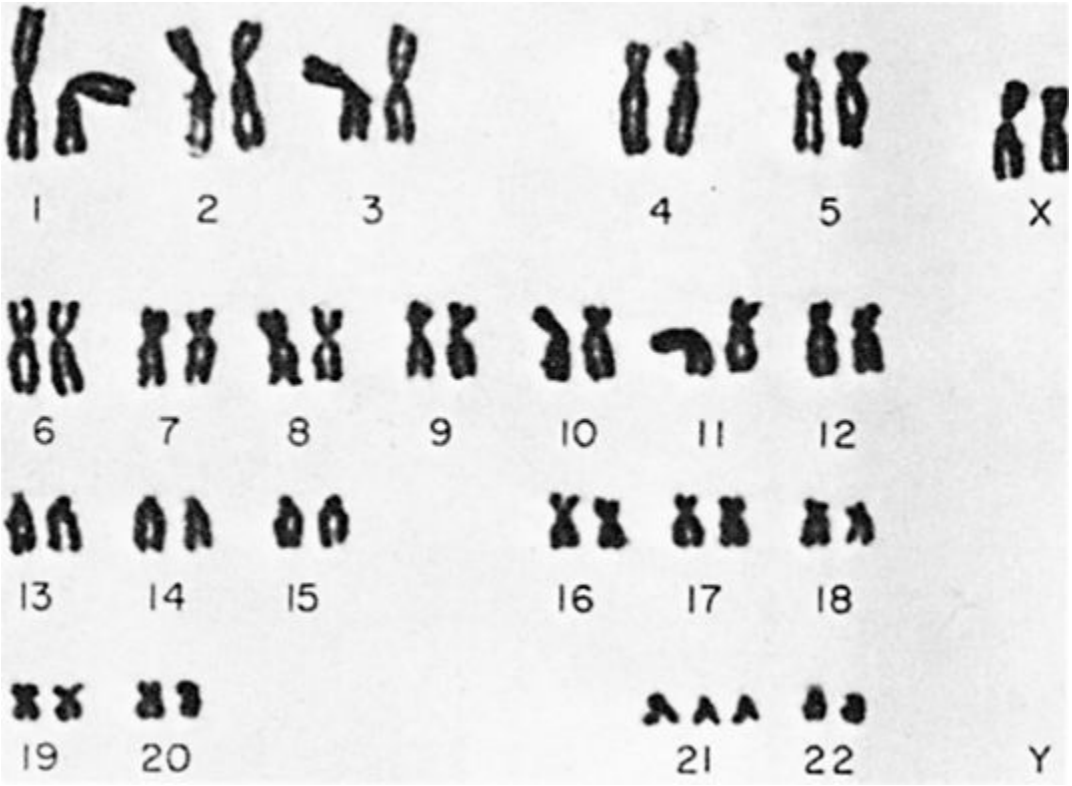


FIGURE 10.17

Karyotype of a child with Down syndrome.

Down syndrome affects 1 in 800 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 1,250 for a woman at age 25, 1 in 1,000 at 30, 1 in 400 at 35, 1 in 100 at age 40, and 1 in 30 at age 45 (March of Dimes, 2012c). Children with Down syndrome have characteristic features that are usually identified at birth (**Fig. 10.18**). These common characteristics include:

- Small, low-set ears
- Hyperflexibility
- Muscle hypotonia
- Wide-spaced eyes
- Ulnar loop on the second digit
- Deep crease across palm (termed a *simian crease*)
- Flat facial profile
- Small white, crescent-shaped spots on irises
- Open mouth with protruding tongue
- Broad, short fingers (Kochhar, 2011)

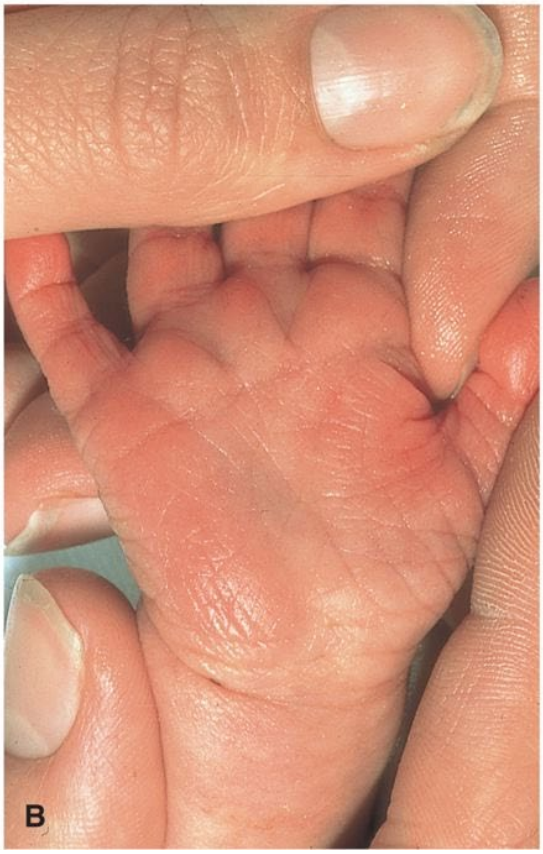


FIGURE 10.18

(**A**) Typical facial features of an infant with Down syndrome. (**B**) A simian line, a horizontal crease in the palm of children with Down syndrome.

The outlook for children with Down syndrome is much brighter now than it was years ago. Most children with Down syndrome have an intellectual disability 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, 2012c). Despite modern medical technology, the individual with Down syndrome has a shortened life span, with an average life expectancy of 55 years (March of Dimes, 2012c).

TRISOMY 18 AND TRISOMY 13

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. These conditions are associated with a high degree of infant mortality, with most dying before their first birthday (March of Dimes, 2012b). Trisomy 18, or Edward syndrome, occurs in 1 of every 5,000 newborns (March of Dimes, 2012b), with advanced maternal age as a causative factor. Prenatally, several findings are apparent on ultrasound: intrauterine growth restriction (IUGR), hydramnios or oligohydramnios, cardiac malformations, a single umbilical artery, and decreased fetal movement. Additionally, trisomy 18 has been associated with a 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 intellectual disability, growth deficiency of the cranium (microcephaly), low-set ears, facial malformations, small-for-gestational-age size, seizures, drooping eyelids, webbing of fingers, kidney and congenital heart defects, rocker-bottom feet, and severe hypotonia (National Organization for Rare Disorders [NORD], 2011e). 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 16,000 newborns (March of Dimes, 2012b). Forty-seven chromosomes (three of chromosome 13) are present. Maternal age is also thought to be a causative factor in this genetic disorder. The common abnormalities associated with trisomy 13 are microcephaly, cardiac defects, small eyes, kidney malformations, central nervous system anomalies, cleft lip and palate, cryptorchidism, polydactyly (**Fig. 10.19**), severe intellectual disability, severe hypotonia, and seizures. Life expectancy is only a few months for most infants with trisomy 13 (NORD, 2011d). Care for these infants is supportive.

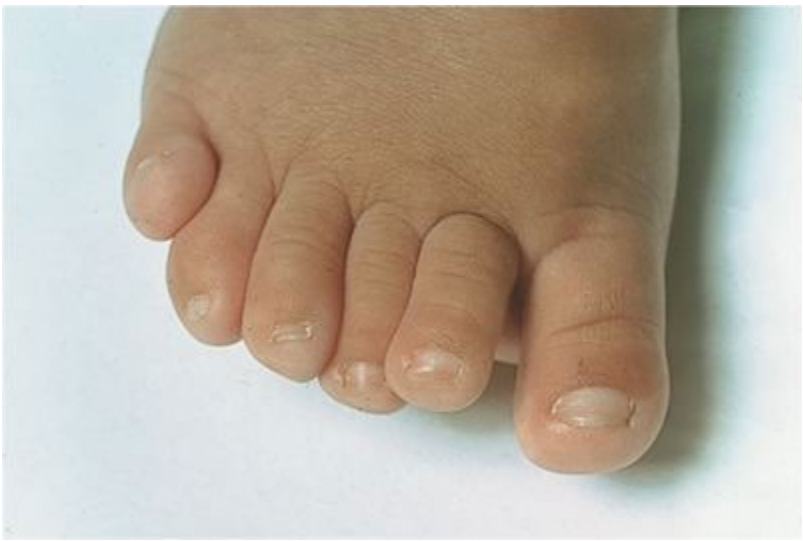


FIGURE 10.19

An infant with trisomy 13 has supernumerary digits (polydactyly).

Structural Abnormalities

Chromosomal abnormalities of structure usually occur when a portion of one or more chromosomes is broken or lost, and during the repair process the broken ends are rejoined incorrectly. Structural abnormalities usually lead to having too much or too little genetic material. Altered chromosome structure can take on several forms. Deletions occur when a portion of the chromosome is missing, resulting in a loss of that chromosomal material. Duplications are seen when a portion of the chromosome is duplicated and an extra chromosomal segment is present. Clinical findings vary depending on how much chromosomal material is involved. Inversions occur when a portion of the chromosome breaks off at two points and is turned upside down and reattached; therefore, the genetic material is inverted. With inversion, there is no loss or gain of chromosomal material and carriers are phenotypically normal, but they do have an increased risk for miscarriage and chromosomally abnormal offspring. Ring chromosomes are seen when a portion of a chromosome has broken off in two places and formed a circle.

The most clinically significant structural abnormality is a translocation. This occurs when part of one chromosome is transferred to another chromosome and an abnormal rearrangement is present.

Structural abnormalities can be balanced or unbalanced. Balanced abnormalities involve the rearrangement of genetic material with neither an overall gain nor loss. Individuals who inherit a balanced structural abnormality are usually phenotypically normal but are at a higher risk for miscarriages and having chromosomally abnormal offspring. Examples of structural rearrangements that can be balanced include inversions, translocations, and ring chromosomes. Unbalanced structural

abnormalities are similar to numerical abnormalities because genetic material is either gained or lost. Unbalanced structural abnormalities can encompass several genes and result in severe clinical consequences.

CRI DU CHAT SYNDROME

Cri du chat syndrome ("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 that resembles the mewing of a cat, which is due to a laryngeal defect. The incidence of the disorder is thought to be approximately 1 in 50,000 live births (NORD, 2011a). In addition to the cat-like, high-pitched cry in infancy, it is also associated with severe intellectual disability, 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 child may survive to adulthood: 75% of deaths occur during the first several months of life and almost 90% occur in the first year. Death occurs in 6% to 8% of the overall population affected with the syndrome. Pneumonia, aspiration pneumonia, congenital heart defects, and respiratory distress are the common causes of death (Chen, 2011a). Parents should be referred for genetic counseling.

FRAGILE X SYNDROME

Fragile X syndrome, also termed Martin-Bell syndrome, is a structural abnormality involving the X chromosome, which demonstrates breaks and gaps. The syndrome is usually diagnosed by molecular DNA studies. Conservative estimates report that fragile X syndrome affects approximately 1 in 4,000 males and 1 in 8,000 females (Jewell, 2011). 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. Fragile X syndrome is characterized by intellectual disability, hyperactivity, large head, long face, 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). It is the most common form of male intellectual disability (NORD, 2011b).

Aside from the morbidity associated with intellectual disability and cognitive/behavioral/neuropsychological problems, the life span of an individual with fragile X syndrome is unaffected. There is no cure for this disorder. Speech, occupational, and physical therapy services usually are needed, as well as special education and counseling.

Sex Chromosome Abnormalities

Chromosomal abnormalities can also involve sex chromosomes. These cases are usually less severe in their clinical effects than autosomal chromosomal abnormalities. Sex chromosome abnormalities are gender specific and involve a missing or extra sex chromosome. They affect sexual development and may cause infertility, growth abnormalities, and possibly behavioral and learning problems. Many

affected individuals lead essentially normal lives. Examples are Turner syndrome (in females) and Klinefelter's syndrome (in males).

TURNER SYNDROME

Turner syndrome is a common abnormality of the sex chromosome in which a portion or all of the X chromosome is missing. It affects about 1 in 2,000 live-born female infants (March of Dimes, 2012b). Clinical manifestations include a low posterior hairline and webbing of the neck, short stature, broad skeletal abnormalities, kidney abnormalities, osteoporosis, heart defects, a shield-like chest with widely spaced nipples, lymphedema, cataracts, scoliosis, puffy feet, underdeveloped secondary sex characteristics, and infertility (NORD, 2011f). Only about a third of cases are diagnosed as newborns; the remaining two thirds are diagnosed in early adolescence when they experience primary amenorrhea. No cure exists for this syndrome. Growth hormone typically is given; hormone replacement therapy also may be used to induce puberty and stimulate continued growth. Most females with Turner syndrome are of normal intelligence and usually live essentially normal lives (Postellon, 2011).

KLINEFELTER SYNDROME

Klinefelter syndrome is a sex chromosomal abnormality that occurs only in males. About 1 in 500 to 1,000 males are born with Klinefelter syndrome (Chen, 2011b). With this syndrome an extra X chromosome (XXY) is present. The extra genetic material causes abnormal development of the testicles, resulting in decreased production of sperm and male sex hormones. Clinical manifestations may include:

- Mild intellectual disability
- Small testicles
- Infertility
- Long arms and legs
- Enlarged breast tissue (gynecomastia)
- Scant facial and body hair
- Decreased sex drive (libido) (NORD, 2011c)

No treatment can correct this genetic abnormality, but testosterone replacement therapy can improve symptoms resulting from the deficiency. Surgery may be done to reduce gynecomastia. Most males with Klinefelter syndrome (XXY) are diagnosed in late puberty. Infertility is common and life expectancy is normal (Chen, 2011b).

Genetic Evaluation and Counseling

Genetic counseling is the process by which clients or relatives at risk for an inherited disorder are advised of the consequences and nature of the disorder, the probability of developing or transmitting it, and the options open to them in management and family planning in order to prevent, avoid, or ameliorate it (Lea, 2010). An individual should be referred for genetic counseling for any of a variety of reasons. **Box 10.2** lists those who may benefit from genetic counseling. In many cases, geneticists and genetic counselors provide information to families regarding genetic diseases. However, an experienced

family physician, pediatrician, or nurse who has received special training in genetics may also provide the information.

BOX 10.2: THOSE WHO MAY BENEFIT FROM GENETIC COUNSELING

- Maternal age 35 years or older when the baby is born
- Paternal age 50 years or older
- Previous child, parents, or close relatives with an inherited disease, congenital anomalies, metabolic disorders, developmental disorders, or chromosomal abnormalities
- Consanguinity or incest
- Pregnancy screening abnormality, including alpha-fetoprotein, triple screen, amniocentesis, or ultrasound
- Stillborn with congenital anomalies
- Two or more pregnancy losses
- Teratogen exposure or risk
- Concerns about genetic defects that occur frequently in their ethnic or racial group (for instance, those of African descent are most at risk for having a child with sickle cell anemia)
- Abnormal newborn screening
- Couples with a family history of X-linked disorders
- Carriers of autosomal recessive or dominant diseases
- Child born with one or more major malformations in a major organ system
- Child with abnormalities of growth
- Child with developmental delay, intellectual disability, blindness, or deafness

Adapted from Beery, T. A., & Wo*r*kman, M. L. *(*2011). *Genetics and genomics in nursing and health care.* Philadelphia, PA: F. A. Davis; Burke, W., Tarini, B., Press, N., & Evans, J. (2011). Genetic screening. *Epidemiologic Reviews, 33,*148–164; and Dayal, M. B., & Athanasiadis, I. (2011). Preimplantation genetic diagnosis. *eMedicine*. Retrieved from **http://emedicine.medscape.com/article/273415-overview#aw2aab6b3**.

A genetic consultation involves evaluation of an individual or a family. Its purposes are to confirm, diagnose, or rule out genetic conditions; to identify medical management issues; to calculate and communicate genetic risks to a family; to discuss ethical and legal issues; and to provide and arrange psychosocial support. Genetic counselors serve as educators and resource persons for other health care providers and the general public.

The ideal time for genetic counseling is before conception. Preconception counseling gives couples the chance to identify and reduce potential pregnancy risks, plan for known risks, and establish early prenatal care. Unfortunately, many women delay seeking prenatal care until their second or third trimester, after the crucial time of organogenesis. Therefore, it is important that preconception counseling be offered to all women as they seek health care throughout their childbearing years, especially if they are contemplating pregnancy. This requires health care providers to take a proactive role.

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 not stop drinking and smoking when I found out I was pregnant? 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. Is it was too late to do anything about this? I am 37 years old and alone. … I started to pray silently when the counselor opened the door. …

Thoughts: This woman is reviewing the past few weeks, 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?

Genetic screening and counseling can raise serious ethical and moral issues for a couple. The results of prenatal genetic testing can lead to the decision to terminate a pregnancy, even if the results are not conclusive but indicate a strong possibility that the child will have an abnormality. The severity of the abnormality may not be known, and some may find the decision to terminate unethical. Another difficult situation that provides an example of the ethical and moral issues surrounding genetic screening and counseling involves disorders that affect only one gender of offspring. A mother may find she is a carrier of a gene for a disorder for which there is no prenatal screening test available. In these cases the couple may decide to terminate any pregnancy where the fetus is the affected sex, even though there is a 50% chance that the child will not inherit the disorder. In these situations, the choice is the couple's and information and support must be provided in a nondirective manner.

Genetic counseling is particularly important if a congenital anomaly or genetic disease has been diagnosed prenatally or if a child is born with a life-threatening congenital anomaly or genetic disease. In these cases families need information urgently so they can make immediate decisions. If a diagnosis with genetic implications is made later in life, if a couple with a family history of a genetic disorder or a previous child with a genetic disorder is planning a family, or if there is suspected teratogen exposure, urgency of information is not such an issue. In these situations, the family needs time to ponder all their options. This may involve several meetings over a longer period of time.

A **teratogen** is any substance, organism, physical agent, or deficiency state present during gestation that is capable of inducing abnormal postnatal structure or function by interfering with normal embryonic and fetal development (March of Dimes, 2012a). Susceptibility to teratogenic agents is dependent on the timing of the exposure and the developmental stage

of the embryo or fetus. Teratogens include alcohol, certain drugs/medications, infections, and certain chemicals.

Genetic counseling involves gathering information regarding birth history, past medical history, and current health status as well as a family history of congenital anomalies, intellectual disability, genetic diseases, reproductive history, general health, and causes of death. A detailed family history is imperative and in most cases will include the development of a pedigree, which is like a family tree (**Fig. 10.20**). Information is ideally gathered on three generations, but if the family history is complicated, information from more distant relatives may be needed. Families receiving genetic counseling may benefit from being told in advance that this information will be necessary; they may need to discuss these sensitive, private issues with family members to obtain the needed facts. When necessary, medical records may be requested for family members, especially those who have a genetic disorder, to help ensure accuracy of the information. Sometimes a pedigree may reveal confidential information not known by all family members, such as an adoption, a child conceived through in vitro fertilization, or a husband not being the father of a baby. Therefore, maintaining confidentiality is extremely important. After careful analysis of the data obtained, referral to a genetic counselor when indicated is appropriate.

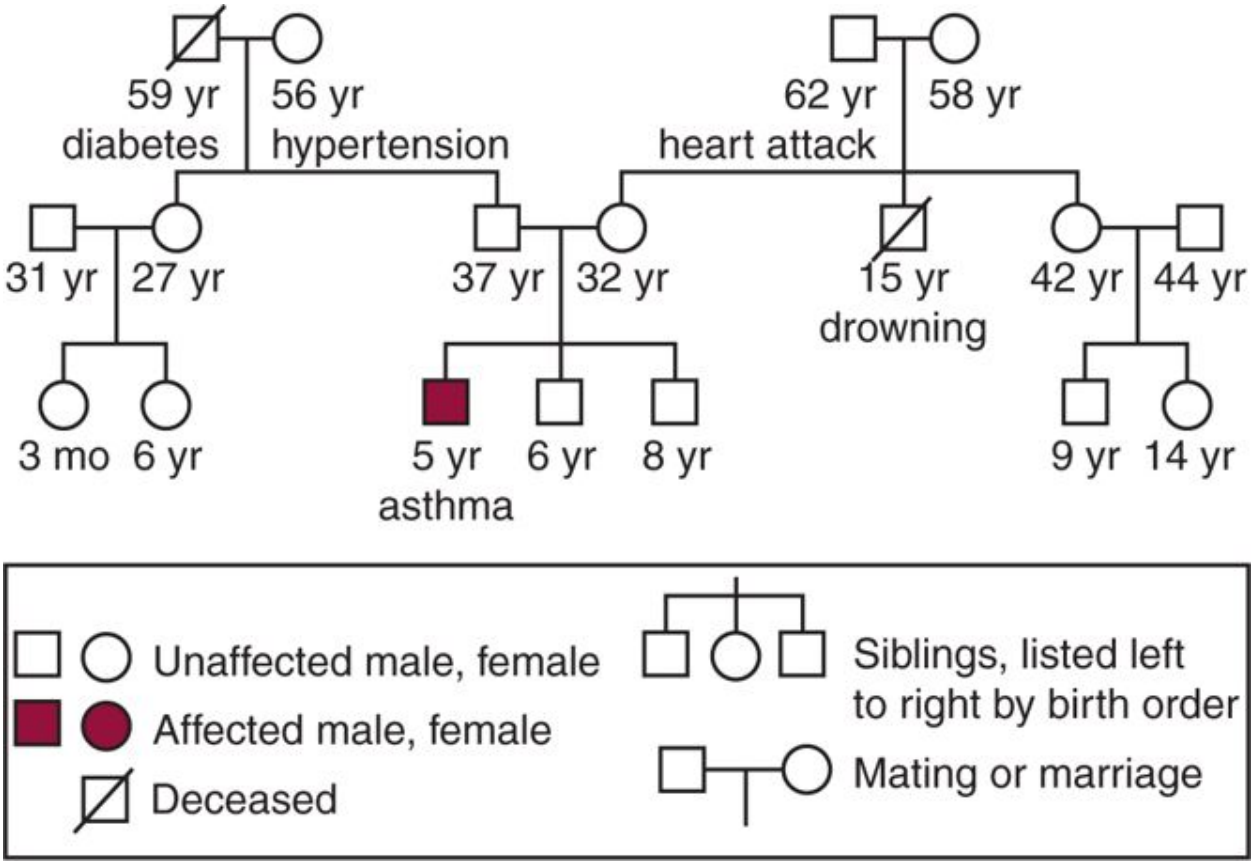


FIGURE 10.20

A pedigree is a diagram made using symbols that demonstrates the links between family members and focuses on medical and health information for each relative.

Medical genetic knowledge has increased dramatically during the past few decades. Not only is it possible to detect specific diseases with genetic mutations, but it is also possible to test for a genetic predisposition to various diseases or conditions and certain physical characteristics. This leads to complex ethical, moral, and social issues. Maintaining client privacy and confidentiality and administering care in a nondiscriminatory manner are essential while maintaining sensitivity to cultural differences. It is essential to respect client autonomy and present information in a nondirective manner.

NURSING ROLES AND RESPONSIBILITIES

The nurse is likely to interact with the client 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. Nurses are often the first health care providers to encounter women with preconception and prenatal issues. Nurses play an important role in beginning the preconception counseling process and referring women and their partners for further genetic testing when indicated.

An accurate and thorough family history is an essential part of preconception counseling. Nurses in any practice setting can obtain a client's history during the initial encounter. The purpose is to gather client and family information that may provide clues as to whether the client has a genetic trait, inherited condition, or inherited predisposition (Beery & Workman, 2011). At a basic level, all nurses should be able to take a family medical history to help identify those at risk for genetic conditions, and then initiate a referral when appropriate. **Box 10.3** presents examples of focused assessment questions that can be used. Based on the information gathered during the history, the nurse must decide whether a referral to a genetic specialist is necessary or whether further evaluation is needed. Families identified with genetic issues need unique clinical care including management of acute illnesses, screening for long-term complications, discussion of the etiology of the condition, connections to social supports, and clarification of the recurrence risks and prenatal testing and treatment options (Hartley, Greenberg, & Mhanni, 2011). Prenatal testing to assess for genetic risks and defects might be used to identify genetic disorders. These tests are described in **Common Laboratory and Diagnostic Tests 10.1**.

BOX 10.3: 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 childbearing, especially if older than 35 years?

Do any family members have an intellectual disability or developmental delays?

Do any family members have a known or suspected metabolic disorder such as PKU?

What is the maternal age of both parents presently with this pregnancy?

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?

Adapted from Edelman, C. L., & Mandle, C. L. (2010). *Health promotion throughout the lifespan* (7th ed.). St. Louis, MO: Mosby Elsevier; Gregg, A. R., & Simpson, J. L. (2010). *Genetic screening and counseling: An issue of obstetrics and gynecology clinics*. St. Louis, MO: Saunders Elsevier; and Jenkins, J. (2011). Family history as a genetic assessment tool: Where are the resources? *American Nurse Today*, *6*(10), 1–3.

Remember Robert and Kate Shafer? Based on the information gathered from their genetic history, they were referred to a genetic specialist. What prenatal tests might be ordered to assess their risk for genetic disorders? What would be the nurse's role related to genetic counseling?

Nurses working with families involved with genetic counseling typically have certain responsibilities. These include:

- Using interviewing and active listening skills to identify genetic concerns
- Knowing basic genetic terminology and inheritance patterns
- Explaining basic concepts of probability and disorder susceptibility
- Safeguarding the privacy and confidentiality of clients' genetic information
- Providing complete informed consent to facilitate decisions about genetic testing
- Discussing costs of genetic services and the benefits and risks of using health insurance to pay for genetic services, including potential risks of discrimination
- Recognizing and defining ethical, legal, and social issues
- Providing accurate information about the risks and benefits of genetic testing
- Using culturally appropriate methods to convey genetic information
- Monitoring clients' emotional reactions after receiving genetic analysis
- Providing information on appropriate local support groups
- Knowing their own limitations and making appropriate referrals (R. Lewis, 2011).

Talking with family members who have recently been diagnosed with a genetic disorder or who have had a child born with congenital anomalies is very difficult. Many times the nurse may be the one who has first contact with these parents and will be the one to provide follow-up care.

Genetic disorders are significant, life-changing, and possibly life-threatening situations. Genetic information is highly technical and the field is undergoing significant technological advances. Nurses need an understanding of who will benefit from genetic counseling and must be able to discuss the role of the genetic counselor with families. The goal is to ensure that families at risk are aware that genetic counseling is available before they attempt to have another baby.

Based on the results of their genetic tests, Robert and Kate are placed at moderate risk for having an infant with an autosomal recessive genetic disorder. The couple asks the nurse what all of this means. What information should the nurse provide about concepts of probability and disorder susceptibility for this couple? How can the nurse help this couple to make knowledgeable decisions concerning their reproductive future?

Nurses play an essential role in providing emotional support to the family through this challenging time. Genetics permeates all aspects of health care. Today, everyone is embracing quality and evidence-based care. Nurses who have an understanding of genetics and genomics will possess the foundation to provide quality,

evidence-based care especially with follow-up counseling after the couple or family has been to the genetic specialist.

COMMON LABORATORY AND DIAGNOSTIC TESTS 10.1: PRENATAL TESTS TO ASSESS RISK FOR GENETIC DISORDERS

Test	Description	Indication	Timing
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

Test	Description	Indication	Timing
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, tests are done to identify cystic fibrosis, sickle cell trait or disease	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, sickle cell trait or disease, Duchenne muscular dystrophy, cystic fibrosis, 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
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

Test	Description	Indication	Timing
Level III ultra-sound/fetal scan	Use of high-frequency sound waves to visualize the fetus	Enables early evaluation of structural changes	Typically performed after 18 weeks' gestation
Triple and quad screening tests	Triple screening includes alpha-fetoprotein, estriol, and beta-hCG; Quad screening includes alpha-fetoprotein, estriol, beat-hCG, and inhibin A	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 15 and 18 weeks' gestation
Preimplantation genetic diagnosis (PGD)	Genetic testing of embryos produced through in vitro fertilization (IVF)	Identifies embryos carrying specific genetic alterations that can cause disease. Only those without genetic alterations are later transferred into the woman's uterus to start a pregnancy. Prevents inheritable genetic disease before implantation.	Usually on day 3 after egg retrieval and 2 days after fertilization, a single blastomere is removed from the developing embryo to be evaluated.

Adapted from Burke, W., Tarini, B., Press, N., & Evans, J. (2011). Genetic screening. *Epidemiologic Reviews*, *33,*148–164; Dayal, M. B., & Athanasiadis, I. (2011). Preimplantation genetic diagnosis. *eMedicine*. Retrieved from **http://emedicine.medscape.com/article/273415-overview#aw2aab6b3**; March of Dimes. (2012e). *Routine prenatal tests*. Retrieved from **http://www.marchofdimes.com/pregnancy/prenatalcare_routinetests.html**; and MedlinePlus. (2012). *Prenatal testing*. Retrieved from **http://www.nlm.nih.gov/medlineplus/prenataltesting.html**.

Take Note!

ESSENTIALS of Maternity, Newborn, & Women's Health Nursing - **THIRD EDITION**

Susan Scott Ricci, **ARNP, MSN, MEd**

Nurses need to be actively engaged with clients and their families and help them consider the facts, values, and context in which they are making decisions. Nurses need to be open and honest with families as they discuss these sensitive and emotional choices.

Nurses should provide ongoing support and education for clients and their families. This includes coping with the disease burden, helping clients and families adapt to a condition in the family, and ensuring adequate understanding of the genetic risks and the available prenatal diagnostic and reproductive choices.

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. Referral to appropriate agencies, support groups, and resources, such as a social worker, a chaplain, or an ethicist, is another key role when caring for families with suspected or diagnosed genetic disorders.

KEY CONCEPTS

- Fertilization, which takes place in the outer third of the ampulla of the fallopian tube, leads to the formation of a zygote. The zygote undergoes cleavage, eventually implanting in the endometrium about 7 to 10 days after conception.
- Three embryonic layers of cells are formed: ectoderm, which forms the central nervous system, special senses, skin, and glands; mesoderm, which forms the skeletal, urinary, circulatory, and reproductive systems; and endoderm, which forms the respiratory system, liver, pancreas, and digestive system.
- Amniotic fluid surrounds the embryo and increases in volume as the pregnancy progresses, reaching approximately a liter 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, which would choke 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. Each person has a unique genetic constitution, or genotype.
- Research from the Human Genome Project has provided a better understanding of the genetic contribution to disease.

- Genetic disorders can result from abnormalities in patterns of inheritance or chromosomal abnormalities involving chromosomal number or structure.
- Autosomal dominant inheritance occurs when a single gene in the heterozygous state is capable of producing the phenotype. Autosomal recessive inheritance occurs when two copies of the mutant or abnormal gene in the homozygous state are necessary to produce the phenotype. X-linked inheritance disorders are those associated with altered genes present on the X chromosome. They can be dominant or recessive. Multifactorial inheritance is thought to be caused by multiple gene and environmental factors.
- In some cases of genetic disorders, a chromosomal abnormality occurs. Chromosomal abnormalities do not follow straightforward patterns of inheritance. These abnormalities occur on autosomal as well as sex chromosomes and can result from changes in the number of chromosomes or changes in the structure of the chromosomes.
- Genetic counseling involves evaluation of an individual or a family. Its purpose is to confirm, diagnose, or rule out genetic conditions, identify medical management issues, calculate and communicate genetic risks to a family, discuss ethical and legal issues, and assist in providing and arranging psychosocial support.
- Legal, ethical, and social issues that can arise related to genetic testing include the privacy and confidentiality of genetic information, who should have access to personal genetic information, psychological impact and stigmatization due to individual genetic differences, use of genetic information in reproductive decision making and reproductive rights, and whether testing is to be performed if no cure is available.
- Preconception screening and counseling can raise serious ethical and moral issues for a couple. The results of prenatal genetic testing can lead to the decision to terminate a pregnancy.
- Nurses play an important role in beginning the preconception counseling process and referring women and their partners for further genetic information when indicated. Many times the nurse is the one who has first contact with these women and will be the one to provide follow-up care.
- Nurses need to have a solid understanding of who will benefit from genetic counseling and must be able to discuss the role of the genetic counselor with families, ensuring that families at risk are aware that genetic counseling is available before they attempt to have another baby.
- Nurses play an essential role in providing emotional support and referrals to appropriate agencies, support groups, and resources when caring for families with suspected or diagnosed genetic disorders. Nurses can assist clients with their decision making by referring them to a social worker, a chaplain, or an ethicist.