chapter 27

# Human Development and Heredity

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## Key Terms

**amnion** (p. 514)
**blastocyst** (p. 513)
**chorion** (p. 514)
**chromosomes** (p. 525)
**embryo** (p. 514)
**fertilization** (p. 511)
**fetus** (p. 518)
**genes** (p. 525)
**human chorionic gonadotropin (hCG)** (p. 513)
**karyotype** (p. 525)
**mammary glands** (p. 522)
**placenta** (p. 514)
**sex-linked trait** (p. 527)
**teratogens** (p. 518)
**umbilical cord** (p. 517)
**zygote** (p. 512)

## Objectives

1. Describe the process of fertilization: when, where, and how it occurs.
2. Do the following regarding prenatal development:
   - Describe the process of development: cleavage, growth, morphogenesis, and differentiation.
   - Explain the three periods of prenatal development: early embryonic, embryonic, and fetal.
   - State two functions of the placenta.
3. Explain hormonal changes during pregnancy.
4. Describe the hormonal changes and stages of labor.
5. Describe the structure of the breast and lactation.
6. Describe immediate postnatal changes and lifelong developmental stages.
7. Discuss heredity and how genetic structures are related, including:
   - Describe the relationships among deoxyribonucleic acid (DNA), chromosomes, and genes.
   - Define *karyotype*.
8. Explain how the gender of the child is determined.
9. State the difference between congenital and hereditary diseases.

Nine months after conception, the reproductive process produces a baby. The "bundle of joy" has arrived on the scene. Note that this term of endearment does not suggest "up, awake, and playing at 2 AM," and diapers, diapers, and more diapers. Nonetheless, Baby is cute, and the urge to reproduce is very strong. Let us follow Baby's start from fertilization through development and birth. Finally, we'll see what is meant by statements like "He's got his father's nose and his mother's smile." This is the genetic story.

## FERTILIZATION

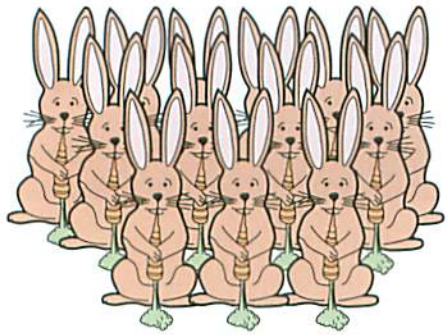
**Fertilization**, also called *conception*, refers to the union of the nuclei of the egg and the sperm. When, where, and how does this union take place?

### WHEN FERTILIZATION OCCURS

Timing is everything. In the female, ovulation occurs at midcycle, around day 14 (see Chapter 26). The egg lives for about 24 hours after ovulation. Sperm usually live between 12 and 48 hours, with some surviving up to 72 hours. Generally speaking, for fertilization to occur, sexual intercourse must take place around the time of ovulation, generally no earlier than 72 hours (3 days) before ovulation and no later than 24 hours (1 day) after ovulation.

ALERT: Evidence suggests that some women are reflex ovulators. These women ovulate in response to having intercourse. Think about it; the chance of pregnancy goes *waaay* up. Remember that rabbits are

reflex ovulators, and we all know about the rabbit population.



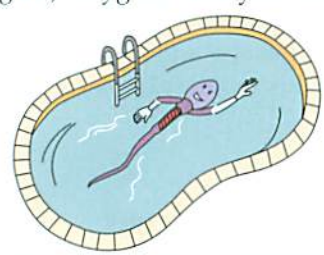
## WHERE FERTILIZATION OCCURS

After ovulation, the egg enters the fallopian tube. Fertilization normally occurs in the first third of the fallopian tube.

## HOW FERTILIZATION OCCURS

During intercourse, about 200 to 600 million sperm are deposited in the vagina, near the cervix of the uterus. Although many of the sperm are killed by the acidic environment of the vagina, about 100,000 survive and swim through the uterus and into the fallopian tube toward the egg. Within 1 to 2 hours after intercourse, thousands of sperm are gathered around the egg in the fallopian tube. Life comes at you fast!

The acrosome on the head of the sperm ruptures and releases enzymes. The enzymes digest the linings of cells that surround the egg. Then, one (and only one) sperm penetrates the membrane of the egg. Upon penetration of the egg, the nuclei of the egg and sperm unite, thereby completing fertilization. The fertilized egg is called a **zygote** (ZYE-goht). "Zygote" was your first name!



The single-cell zygote has 46 chromosomes: 23 from the egg and 23 from the sperm. The zygote begins to divide, forming a cluster of cells that slowly makes its way through the fallopian tube toward the uterus. When this cluster of cells reaches the uterus, it implants itself into the plush endometrial lining, where it grows and develops into a human being with billions of cells.

### Re-Think

1. Where does fertilization occur?
2. Why is it possible for a fertilized ovum to take up residence within the abdominal cavity rather than the uterus?

# HUMAN DEVELOPMENT

Development is a process that begins with fertilization and ends with death. Human development is divided into two phases: prenatal development and postnatal development. Prenatal development begins with fertilization and ends at birth. The time of prenatal development is called *pregnancy*, or *gestation*. The normal gestation period lasts 38 weeks, or about 9 months. Pregnancy is divided into trimesters (3-month periods): the first trimester is the first 3 months of pregnancy; the second trimester is months 4, 5, and 6; and the third, or last, trimester is months 7, 8, and 9. Postnatal development begins with birth and ends with death; it is what we are all doing now—living *life*.

# PRENATAL DEVELOPMENT

What does prenatal development include? Prenatal development includes cleavage, growth, morphogenesis, and differentiation. Cleavage is cell division by mitosis. Mitosis produces two identical cells from a single cell. Thus, one cell splits into two cells, the two cells split into four cells, four cells split into eight cells, and so on. Each new cell is identical to the parent cell. Mitotic cell division increases the numbers of cells but not their actual size. The size of the cell increases through growth. As development progresses, therefore, both the number and size of the cells increase.

Morphogenesis (mohr-foh-JEN-eh-sis) is the shaping of the cell cluster. Certain cells migrate to specific areas in the cell cluster. This process changes the shape of the cell mass. For example, cells migrate to the side of the cell mass and take the appearance of tiny buds. These buds eventually become legs. Through morphogenesis, the round cluster of cells develops into an intricately and wondrously formed infant. Baby is shaping up!

Differentiation is the process whereby a cell becomes specialized. A cell differentiates to become a nerve cell, muscle cell, blood cell, or some other cell.

What are the periods of prenatal development? There are three periods: early embryonic, embryonic, and fetal periods.

## EARLY EMBRYONIC PERIOD

### FROM ZYGOTE TO BLASTOCYST ... OR FROM ZYGOTE TO EGGPLANT

The early embryonic period lasts for 2 weeks after fertilization. During this period, the zygote undergoes mitosis and travels from the fallopian tube into the uterus. After fertilization (Figure 27-1, *A*) the zygote undergoes cleavage. Cleavage is accomplished by mitosis, or cell division that increases the numbers of cells. The cells formed by mitotic cell division are blastomeres (see Figure 27-1, *B*). Note the two-cell, four-cell, and eight-cell cluster.

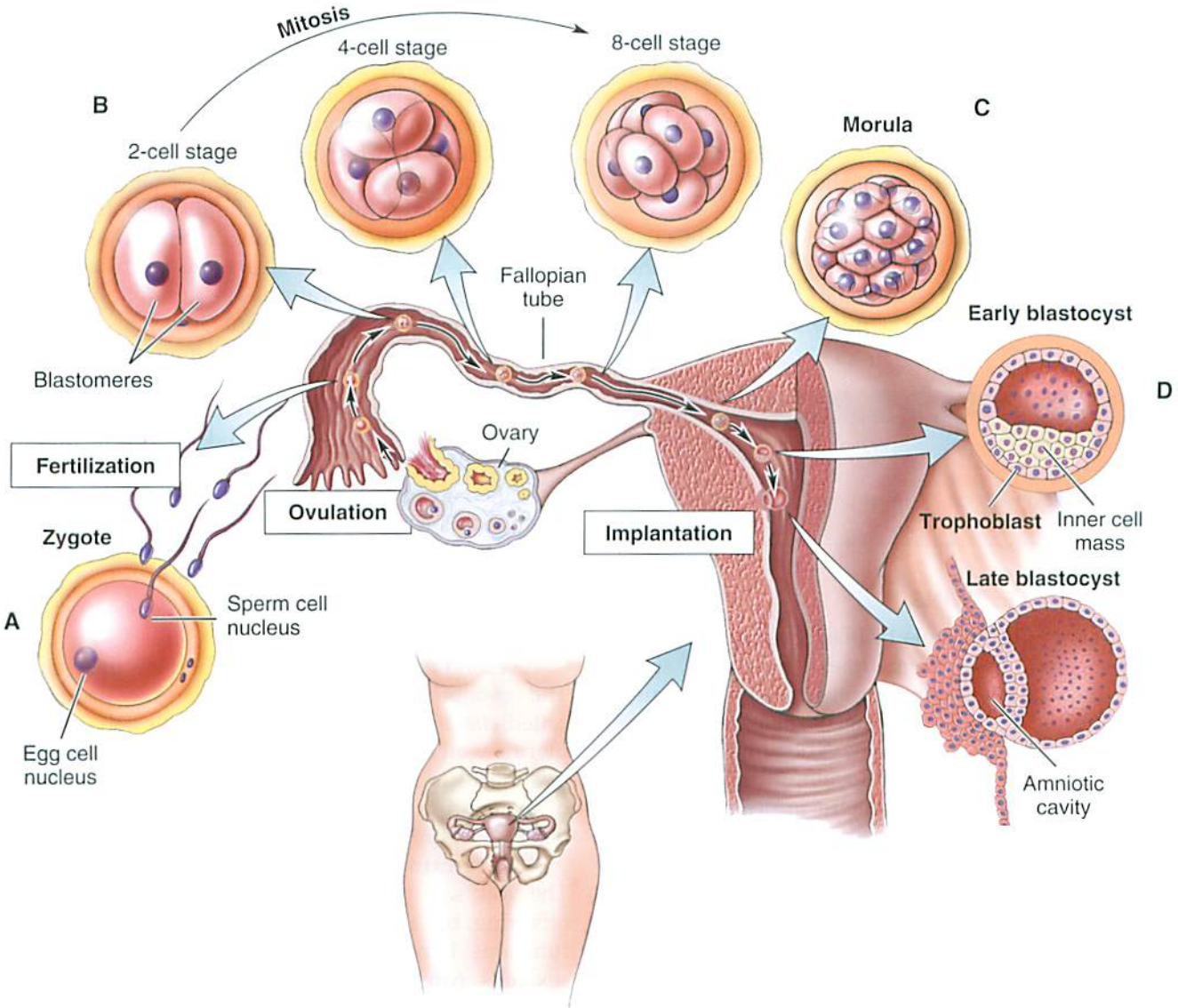



FIGURE 27-1 **A-D,** Stages of early embryonic development, from zygote to blastocyst.

When the number of cells increases to 16, the collection of cells is called a *morula* (see Figure 27-1, *C*). The morula looks like a raspberry, so tiny that it is visible only through a microscope. Transformation from the zygote to a morula takes about 3 days. The morula enters the uterine cavity, where it floats around for 3 to 4 days and continues to undergo mitosis. By the end of the fifth day, the morula has developed into a **blastocyst**.

Note the structure of the early blastocyst (see Figure 27-1, *D*). The early blastocyst contains a hollow cavity surrounded by a single layer of flattened cells and a cluster of cells at one side. The single layer of flattened cells surrounding the cavity is called the *trophoblast*. These cells will help form the placenta and also secrete an important hormone. The cluster of cells within the blastocyst is called the *inner cell mass*. These cells will eventually form Baby.

The late blastocyst develops by day 7. This stage shows the beginnings of the amniotic cavity. The late blastocyst burrows into the endometrial lining of the uterus, where it is gradually covered over by cells of the endometrial lining. The burrowing process is called *implantation*. We now have an "eggplant"!

During implantation, the blastocyst also functions as a gland. The trophoblast, the flattened layer of cells that surrounds the cavity, secretes a hormone called **human chorionic gonadotropin (hCG)**, which travels through the blood to the ovary, where it prevents the deterioration of the corpus luteum. In response to hCG, the corpus luteum continues to secrete estrogen and progesterone. The estrogen and progesterone stimulate the growth of the uterine wall and prevent menstruation.

The secretion of hCG continues at a high level for about 2 months and then steadily declines as the

placenta develops. The placenta eventually takes over the role of the corpus luteum by secreting large amounts of estrogen and progesterone. The blastocyst thus helps preserve its own survival through its secretion of hCG. Think about it: Baby helps Baby survive. With the implantation of the blastocyst and the organization of the inner cell mass, the early embryonic period comes to a close.

hCG forms the basis of pregnancy tests. hCG is secreted in early pregnancy and can be detected in the mother's urine or blood. A pregnancy test may indicate positive results within about 8 to 10 days after fertilization.

**Re-Think**

1. List the stages of development from fertilization to implantation.
2. Explain the significance of the trophoblastic secretion of hCG.

## SEEING DOUBLE: TWINS

Each cell within the morula or blastocyst can become a complete individual. Sometimes, these cells split and two embryos begin developing at the same time, thereby producing two offspring (twins) rather than one. These are identical, or monozygotic (mon-oh-zye-GOH-tik), twins because they develop from the same zygote and have identical genetic information. For monozygotic twins to develop, one sperm fertilizes one egg, and the zygote then splits.

Sometimes a woman ovulates two eggs, which are then fertilized by two different sperm. Because two babies are produced, they are called *twins*. These twins, however, are not identical. They do not develop from the same egg and do not have the same genetic information. They are called *fraternal*, or *dizygotic* (dye-zye-GOH-tik), *twins* (meaning that they come from two different zygotes). Triplets, quadruplets, and other multiple births can develop in the same two ways.

**Re-Think**

1. What is the difference between monozygotic and dizygotic twins?
2. Identical twins are always the same gender. Why is that not true for fraternal twins?

## EMBRYONIC PERIOD

Embryonic development lasts for 6 weeks, from week 3 through week 8. During this period, the baby-to-be is called an **embryo**. The embryonic period involves the formation of extraembryonic membranes, the placenta, and all of the organ systems in the body.

### EXTRAEMBRYONIC MEMBRANES

The extraembryonic membranes form outside the embryo; hence, the term *extra*embryonic. The membranes help protect and nourish the embryo; they are also involved in the embryonic excretion of waste. At birth, the membranes are expelled along with the placenta as the afterbirth. The four extraembryonic membranes are the amnion, chorion, yolk sac, and allantois.

The **amnion** (AM-nee-on) enlarges and forms a sac around the embryo (Figure 27-2). The sac is called the *amniotic sac* and is filled with a fluid called the *amniotic fluid*. The amniotic fluid forms a protective cushion around the embryo and helps protect it from bumps and changes in temperature. The amniotic fluid also nourishes embryos as they drink and digest it. About 1 L of amniotic fluid occupies the amniotic sac at full term. Interestingly, much of the amniotic fluid is from the urine produced by the fetal kidneys!

The embryo secretes waste and sheds cells into the amniotic fluid. This process is the basis for a diagnostic test called an *amniocentesis*, in which a sample of amniotic fluid is aspirated from the amniotic cavity and examined for evidence of fetal abnormalities (see Figure 27-2, *B*). The amniotic sac is often called the *bag of water*. It breaks before delivery and generally signals the onset of labor.

A second extraembryonic membrane is the chorion. The **chorion** develops many finger-like projections called *chorionic villi*. The chorionic villi penetrate the uterine wall and interact with the tissues of the mother's uterus to form the placenta. Sampling of the cells of the chorionic villi is another way to detect genetic defects (Figure 27-3 and Figure 27-2, *C*).

A third extraembryonic membrane is the yolk sac. The yolk sac in birds and reptiles helps nourish the offspring, but the yolk sac in humans serves different functions: it produces red blood cells and immature sex cells. After the sixth week, the yolk sac ceases to function. The embryonic liver and spleen then produce red blood cells, and by the seventh month the bone marrow has assumed this function. By then, the yolk sac has become part of the umbilical cord.

The allantois (ah-LAN-toh-is) is the fourth extraembryonic membrane. The allantois contributes to the formation of several structures, including the urinary bladder. The blood vessels of the allantois also help form the umbilical blood vessels, which transport blood to and from the placenta. After the second month, the allantois deteriorates and becomes part of the umbilical cord.

**Re-Think**

List four extraembryonic membranes and provide a function of each.

### PLACENTA

The **placenta** is a disc-shaped structure about 7 inches (15 to 20 cm) in diameter and 1 inch (2.5 cm) thick. Normally, the placenta develops in the upper portion of the uterus. The placenta is a highly vascular

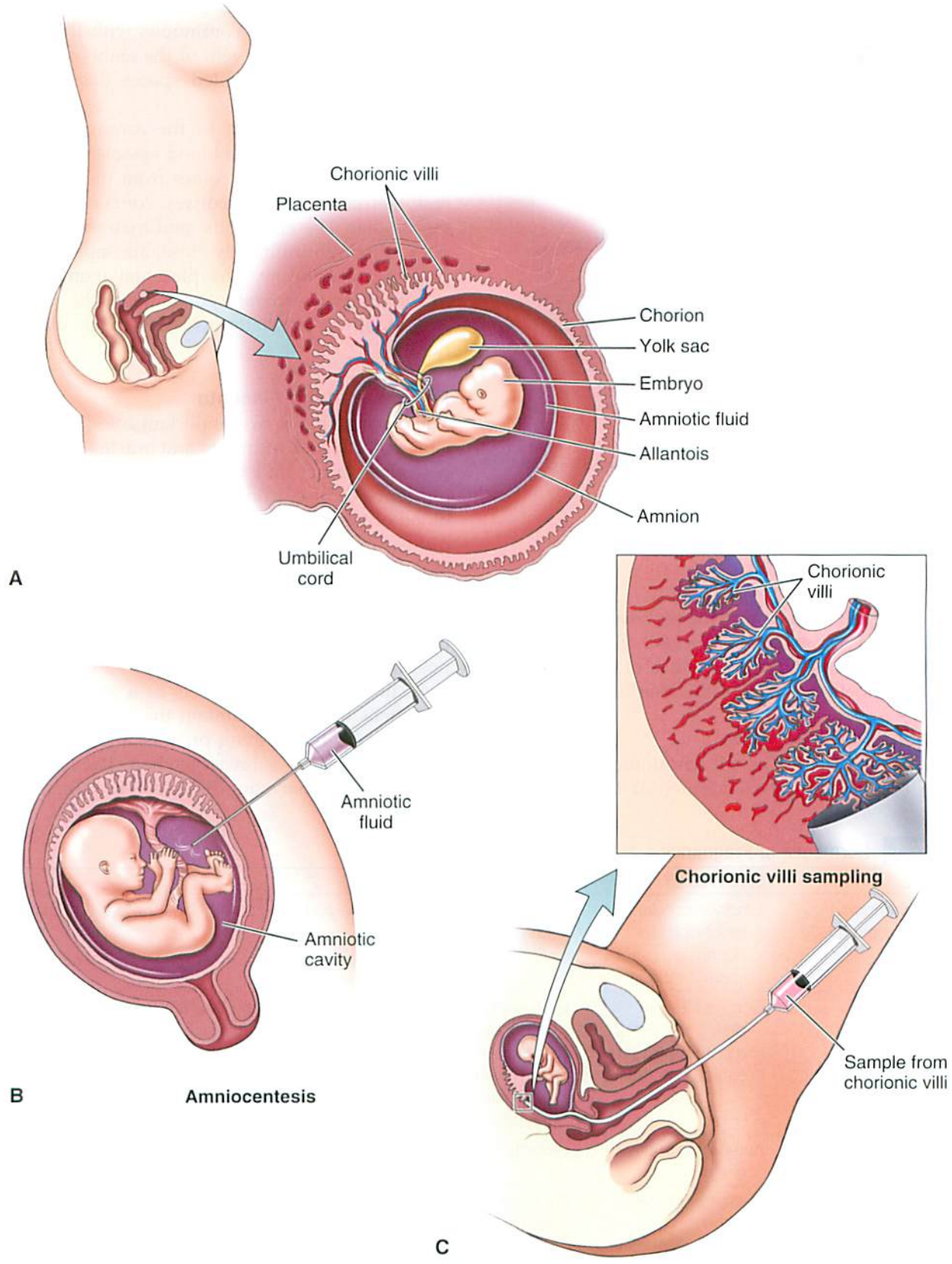



FIGURE 27-2 Extraembryonic membranes and the formation of the placenta. **A,** The embryo is surrounded by the amnion and the amniotic fluid. **B,** Amniocentesis. **C,** Chorionic villi sampling.

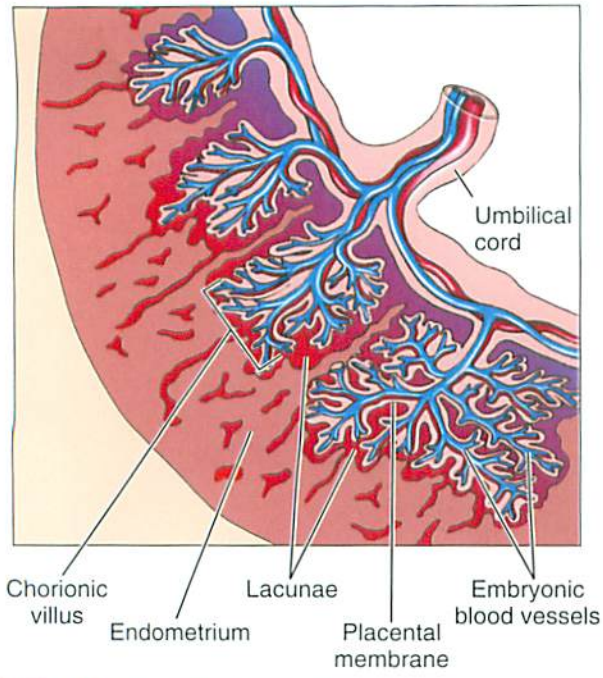



FIGURE 27-3 Cross-section of the chorionic villi.

structure formed from embryonic and maternal tissue. By the end of the embryonic period (8 weeks), the placenta is functional. After the birth of the baby, the placenta is expelled as part of the afterbirth.

### Formation of the Placenta

The placenta develops as the chorionic villi of the embryo burrow into the endometrial lining of the uterus (see Figure 27-3). The chorionic villi contain blood vessels that are continuous with the umbilical arteries and umbilical vein of the embryo. The chorionic villi sit in blood-filled spaces (lacunae) in the mother's endometrium.

Pay special attention to the arrangement of the embryonic and maternal blood vessels. The chorionic villi contain blood that comes from the embryo. The endometrial spaces (lacunae) contain blood from the mother. The embryonic and maternal blood supplies, although intimately close, are separated by the placental membrane. The placental membrane thus maintains two separate circulations: the embryonic and maternal circulations. The two circulations do not mix!

### Functions of the Placenta

The placenta plays two important roles. First, it is the site across which nutrients and waste are exchanged between mother and baby. Oxygen, food, and other nutrients diffuse from the mother's blood into the blood of the embryo. Carbon dioxide and other waste diffuse from the embryo's blood into the mother's blood and are then excreted. Baby-to-be "breathes," "eats," and excretes at the placenta. If the placenta is injured in any way, the oxygen supply to the embryo may be cut off, causing irreversible brain damage and possibly death.

Secondly, the placenta acts as a gland. It secretes hormones that help maintain the pregnancy, prepare the body for the birthing process, and promote postnatal events such as breast-feeding. The hormones of pregnancy are listed and described in Table 27-1.

**Table 27-1 Hormones of Pregnancy**

| HORMONE | SECRETED BY | EFFECTS |
|---|---|---|
| Human chorionic gonadotropin (hCG) | Embryonic cells (trophoblasts) during implantation | Maintains the function of the corpus luteum; forms the basis of the pregnancy test |
| Estrogen and progesterone | Corpus luteum during the first 2 months; placenta after 2 months | Both estrogen and progesterone stimulate the development of the uterine lining and mammary glands. Progesterone inhibits uterine contractions during pregnancy. Estrogen causes relaxation of the pelvic joints. At the beginning of labor, estrogen opposes the quieting effects of progesterone on uterine contractions and sensitizes the myometrium to oxytocin. |
| Prolactin | Anterior pituitary gland | Prolactin stimulates the breast to secrete milk. |
| Oxytocin | Posterior pituitary gland | Oxytocin causes the release of milk from the breast (part of the milk let-down reflex initiated by suckling). It causes uterine contractions (participates in labor and postpartum uterine contractions to decrease bleeding). |
| Prostaglandins | Placenta | Prostaglandins stimulate uterine contractions. |
| Aldosterone | Adrenal cortex | Aldosterone expands blood volume. |
| Human placental lactogen (HPL) | Placenta | HPL affects the maternal energy metabolism to make more nutrients available to the fetus. |

**Re-Think**

What are two functions of the placenta?

## HOOK UP: THE UMBILICAL CORD

How does the embryo connect with the mother? The **umbilical cord** is the structure that connects embryo and mother at the placenta (see Figure 27-2). The umbilical cord contains two umbilical arteries and one umbilical vein (see Figure 18-8). Because the umbilical cord carries oxygen-rich blood to the developing infant, it is literally the baby's lifeline. Compression or injury to the umbilical cord can cause severe distress and possibly the death of the baby.

When the baby is delivered, the umbilical cord is cut, severing the placenta. The stump of the cord shrivels up, drops off, and leaves the navel (belly button). The baby's organs, such as the lungs, kidneys, and digestive system, must then take over the functions previously performed by the placenta.

**Re-Think**

Why can umbilical cord compression or premature separation of the placenta cause fetal death?

## ORGANOGENESIS

The embryonic period is a time of organogenesis (or-gah-no-JEN-eh-sis), or the formation of body organs and organ systems. The inner cell mass of the blastocyst forms a flattened structure called the *embryonic disc*. The embryonic disc, in turn, gives rise to three primary germ layers: the ectoderm, mesoderm, and endoderm.

All the tissues and organs of the body develop from these germ layers. For example, the ectoderm gives rise to the nervous system, portions of the special senses, and the skin. The skin is one of the earliest organs to develop and forms during the third week. The mesoderm gives rise to muscle, bone, blood, and many of the structures of the cardiovascular system. The endoderm gives rise to the epithelial lining of the digestive tract, respiratory tract, and parts of the urinary tract. By the end of the embryonic period (week 8), the main internal organs are established. The embryo weighs about 1 g, is about 1 inch (2.5 cm) in length, and has a human appearance (Figure 27-4).

## BE CAREFUL: TERATOGENS

Because the organs of the body are being formed at this time, the embryonic period is most critical for

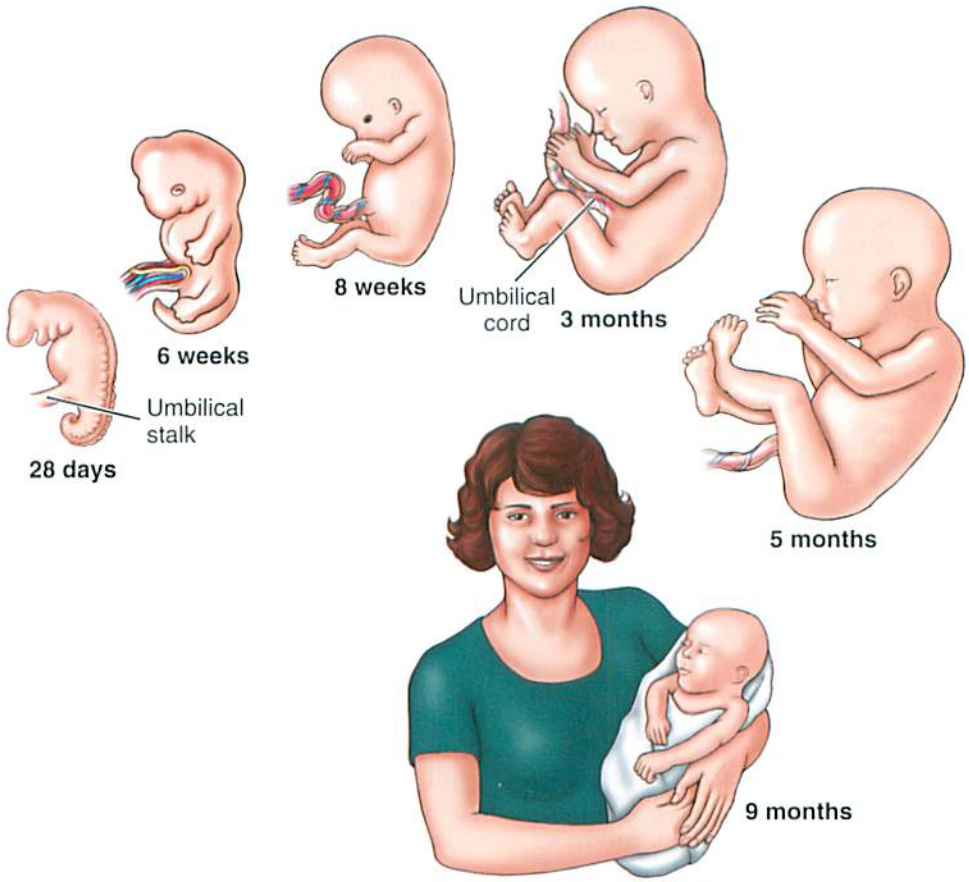



FIGURE 27-4 From embryo to fetus to baby.

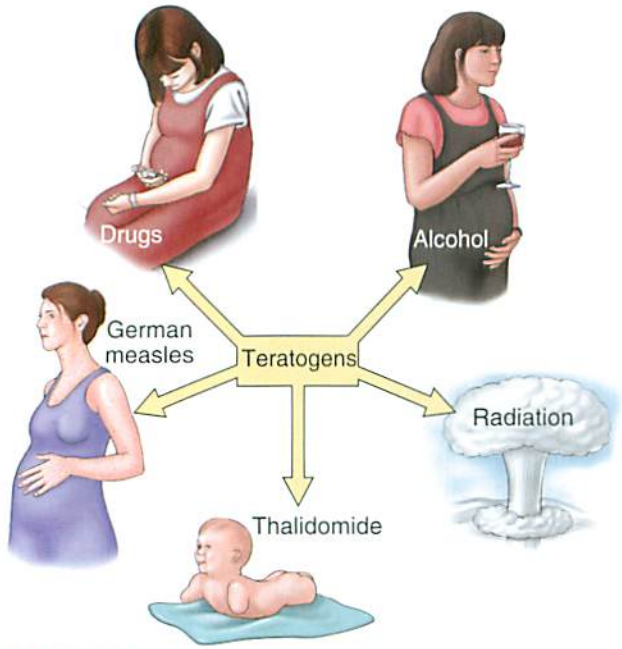



FIGURE 27-5 Teratogens: "monster-producing" agents.

development. Toxic substances such as alcohol, drugs, and certain pathogens can cross the placental membrane and interfere with embryonic development, causing severe birth defects. These toxic substances are called **teratogens** (TER-ah-toh-jens), a word that means "monster-producing" and attests to the severity of teratogenic birth defects. Hazardous conditions such as exposure to radiation can also act as teratogens (Figure 27-5).

### Do You Know...

**How You Get That Extra Inch, or "Slackus the Urachus"?**

Following delivery of a baby, the umbilical cord is tied off or clamped. Traditionally, the cord of a girl baby was tied close to the abdominal wall. The cord of a male baby, however, was tied farther away from the abdominal wall. Why the difference in cord length?

In the baby, a cordlike structure called the *urachus* extends from the top of the urinary bladder to the navel. It was thought that the urachus would be pulled up tight if the cord were tied too close to the abdominal wall. The tight urachus in turn would pull up on the penis, thereby stunting its growth. Conversely, by leaving the umbilical cord long, the urachus would relax, and the penis would grow longer. This obsession with penile length gave rise to the motto "slackus the urachus." Today, the umbilical cord in both male and female babies is clamped about 1 to 2 cm from the abdominal wall. No penile shortening has been recorded.

Alcohol is a potent teratogen. It can cause a cluster of birth defects known as *fetal alcohol syndrome*. In addition to causing facial deformities, alcohol interferes with neurological and mental development. The tranquilizer thalidomide is another drug that produces teratogenic effects. The development of finlike appendages instead of arms and legs has been correlated with the ingestion of thalidomide during pregnancy. Because of the sensitivity of the embryo to teratogenic agents, the mother must be extremely careful to protect her unborn child from toxic substances and hazardous conditions. Many drugs are teratogenic and most pharmacological clinical trials do not involve pregnant women!

### Do You Know...

**What a Sonogram Is?**

A sonogram is an image produced by sound waves as they encounter different tissues and organs. During the procedure, sound waves are directed through the mother's abdomen. As the sound waves "hit" the fetus, an image appears on the scope, showing an outline of the fetus. The size, position, and gender of the fetus can be determined by sonography, as can certain fetal abnormalities.

## FETAL PERIOD

The fetal period extends from week 9 to birth. At this time, the developing offspring is called a **fetus**. The fetal period is primarily a time of growth and maturation; only a few new parts appear. Body proportions, however, continue to change. For example, at 8 weeks, the head is almost as large as the body. At birth, the head is proportionately much smaller than the body. Note the changes in body size and proportion in Figure 27-4.

Table 27-2 summarizes prenatal development. Note that a primitive nervous system begins to form in the third week. The heart and blood vessels originate during the second week. The heart is pumping blood to all the organs of the embryo by the second month, and a heartbeat can be detected during the third month, when the gender of the fetus can also be determined. Once the testes differentiate, they produce the male sex hormone testosterone. Testosterone stimulates the growth of the male external genitals. In the absence of testosterone, female genitals form.

The mother first feels the fetus move during the fifth month; this experience is called *quickening*. As the fetus grows, its skin becomes covered by a fine downy hair called *lanugo* (lah-NOO-go) (during the fifth month). The skin is covered by a white, cheeselike substance called the *vernix caseosa*. The vernix is thought to protect the delicate fetal skin from the amniotic fluid. By the fifth month, the fetus is in crowded quarters and is flexed in the fetal position. During the last 2 months, the baby is gaining weight rapidly as fat is deposited in the subcutaneous tissue. As the time for birth approaches, the fetus rotates so that the head is pointed toward the cervix. At the end of 38 weeks, the fetus is full term. During this period, the fetal weight has increased from less than 0.5 oz (14 g) to 7.5 lb (3.4 kg; average weight of a full-term infant). The fetus has grown in length from about 1 to 21 inches (2.5 to 53 cm). Baby is ready to face the world!

**Table 27-2 Human Development**

| TIME | DEVELOPMENTAL EVENT |
|---|---|
| **Embryonic** | |
| Second week | Implantation occurs. The inner cell mass is giving rise to the primary germ layers; beginning of placental development. |
| Third week | Beginning of the nervous system |
| Fourth week | Appearance of limb buds. Heart is beating. Embryo has tail. Other organ systems begin. |
| Fifth week | Enlarged head; nose, eyes, and ears are noticeable. |
| Sixth week | Fingers and toes are present; appearance of cartilage skeleton |
| Second month | All organ systems are developing. Cartilaginous skeleton is being replaced by bone (ossification). Embryo is about 1.5 inches (3.8 cm) long. |
| **Fetal** | |
| Third month | Facial features present in crude form. Can determine gender (the external reproductive organs are distinguishable as male or female). |
| Fourth month | Sensory organs are present; eyes and ears attain shape and position. Eyes blink and the baby begins sucking movements. Skeleton is visible. |
| Fifth month | Vernix caseosa and a fine downy hair (lanugo) cover the skin. Heartbeat can be heard during a prenatal visit. Quickening occurs. |
| Sixth month | Continues to grow. Myelination of the spinal cord begins. |
| Seventh month | Eyes are open. Testes descend into scrotum. Weighs about 3 lb (135 g). Bone marrow becomes the only site of blood cell formation. |
| Eighth month | Body is lean and well proportioned. Subcutaneous fat begins to be deposited. |
| Ninth month | Full term and ready to be delivered. Average weight is 7.5 lb (338 g), average length is 21 inches (53.3 cm). |

Sometimes, the embryo or fetus is born too early, before the 9-month gestation period is completed. A number of terms are used to describe early birth. An *abortion* is the loss of an embryo or fetus any time during the gestational period; most commonly, abortion refers to the termination of the pregnancy before the twentieth week of development. A *spontaneous abortion* occurs naturally, with no artificial interference. Usually, a spontaneous abortion is caused by some fetal abnormality. A *miscarriage* is the layperson's term for a spontaneous abortion. An *induced abortion* is an abortion deliberately caused by some artificial or mechanical means. An unwanted pregnancy is a common cause of an induced abortion. A *therapeutic abortion* is performed by a physician as a form of treatment for the mother. For example, a pregnancy that threatens the life of the mother may be terminated to save her life or improve her medical condition.

A baby born before 38 weeks but capable of living outside the womb is a premature or preterm infant. A 20-week-old fetus is considered viable—that is, able to live outside the womb. A premature baby is small but, more importantly, it is immature and may require medical support. In particular, the hypothalamus is too immature to regulate body temperature well, and the surfactant produced by the fetal lungs is inadequate to maintain breathing. Generally, the more premature the birth, the greater the need for medical support.

### 2+2 Sum It Up!

Fertilization takes place in the fallopian tube, when the nuclei of a sperm and egg unite, producing a zygote. The zygote gradually moves through the fallopian tube into the uterus, where it develops into an infant over a 9-month period. Prenatal development includes the processes of cleavage (mitosis), growth, morphogenesis, and differentiation. Prenatal development is divided into three periods: the early embryonic period (2 weeks), the embryonic period (6 weeks), and the fetal period (7 weeks to birth at 38 weeks). The zygote undergoes mitosis and develops into a blastocyst; trophoblastic cells help the blastocyst to implant. Other major accomplishments of the embryonic period include the formation of extraembryonic membranes and the development of the placenta and umbilical cord. The embryonic period is also the period of organogenesis. The fetal period is primarily a period of rapid growth and maturation. Baby shapes up, fattens up, and moves about.

### ? Re-Think

Why is the embryo more susceptible to teratogens than a seventh-month fetus?

## CHANGES IN THE MOTHER'S BODY DURING PREGNANCY

Throughout pregnancy, the mother supplies all the food and oxygen for the fetus and eliminates all the

waste. This added burden requires many changes in the mother's physiology:

- The rate of metabolism increases. For example, the mother secretes greater amounts of the thyroid hormones triiodothyronine ($T_3$) and thyroxine ($T_4$). Human placental lactogen circulates within the maternal circulation; among other growth-promoting actions, this hormone makes additional amounts of glucose available to the growing fetus.
- The mother's blood volume expands by as much as 40% to 50%. The increase in blood volume is caused by an increase in the secretion of aldosterone by the adrenal cortex. To pump the additional blood and meet the demands of an increased metabolism, the activity of the cardiovascular system increases. For example, heart rate, stroke volume, and cardiac output increase.
- Respiratory activity increases to provide additional oxygen and eliminate excess carbon dioxide.
- The kidneys work harder and produce more urine because they must eliminate waste for both the mother and fetus.
- Under the influence of estrogen and progesterone, the size and weight of the uterus increase dramatically as the fetus grows to full term. To accommodate the growth of the uterus, the pelvic cavity expands as the sacroiliac joints and symphysis pubis become more flexible. With growth, the uterus pushes the abdominal organs upward. In the later months of pregnancy especially, the upward displacement of abdominal organs exerts pressure on the diaphragm and hampers the mother's breathing.
- The mother's nutritional needs increase as the maternal organs (uterus and breasts) grow and she provides for the growing fetus. In particular, the need for calcium increases, because an increase in parathyroid hormone (PTH) extracts calcium from the mother's bone, making it available to the growing fetus.

Pregnancy brings some discomforts for some women. Nausea and vomiting, generally referred to as *morning sickness*, commonly occur in the first 3 months. Morning sickness may be caused by hormonal changes, especially to the elevated levels of hCG. During the later months of pregnancy, the woman gains approximately 2 to 3 lb/month. The added weight causes a shift in the mother's center of gravity, thereby affecting her balance and forcing her to adjust her walking style. Eventually, many women appear to be waddling.

The added weight may also cause discomfort in the lower back and many other aches as the uterine ligaments and other supporting structures stretch. The expanding uterus stretches the abdominal skin, causing stretch marks, or striae (STRYE-ay). It also displaces the stomach upward, causing heartburn. Frequent urination results from increased urine formation and compression of the urinary bladder by the uterus. Finally, the expanded uterus hampers the return of blood through the veins of the lower body region. This inhibited blood flow, in turn, may cause varicose veins and hemorrhoids. No wonder the mother-to-be looks forward to giving birth!

Although these discomforts of pregnancy are normal, several pregnancy-related conditions are not normal but are dangerous to both the mother and child. For example, the mother may develop a toxemia of pregnancy. This condition is characterized by an elevated blood pressure and progresses in severe cases to generalized seizures. The early stage is called *pre-eclampsia*, and the later seizure stage is called *eclampsia*.

**2+2 Sum It Up!**

Pregnancy causes many changes in the mother. Hormonal changes are numerous and complex. Secretion of hCG, estrogen, and progesterone help maintain the pregnancy and prepare the organs of reproduction for 9 months of pregnancy. Other hormones, such as aldosterone, thyroid hormones, and parathyroid hormone, prepare the mother's body to nourish and sustain the growing unborn child. Almost every maternal organ responds to the presence of the fetus; the heart pumps more blood, the kidneys excrete more waste, and the increased metabolic rate indicates that every cell is working harder.

## BIRTH OF BABY

Finally, Baby is ready to face the world. The birth process is called *parturition* (pahr-too-RIH-shun). Labor is the process whereby forceful contractions expel the fetus from the uterus. Once labor starts, forceful and rhythmic contractions begin at the top of the uterus and travel down its length, forcing the fetus through the birth canal.

### LABOR

#### HORMONAL BASIS OF LABOR

The precise mechanism that starts labor is unknown. A number of hormonal stimuli do, however, play a role. For example, progesterone, which normally quiets uterine contractions during pregnancy, is secreted in decreasing amounts after the seventh month. This decrease coincides with an increase in the secretion of estrogen. Estrogen has two effects on the uterus: it opposes the quieting effect of progesterone on uterine contractions, and it sensitizes the myometrium (uterine muscle) to the stimulatory effects of oxytocin.

The secretion of prostaglandins by the placenta also plays a role in initiating labor. Prostaglandins stimulate uterine contractions. Finally, the stretching of the uterine and vaginal tissue in the late stage of pregnancy stimulates nerves that send signals to the hypothalamus. The hypothalamus, in turn, stimulates the release of oxytocin from the posterior pituitary gland. Oxytocin exerts a powerful stimulating effect on the

myometrium and is thought to play an important role in labor.

### Do You Know...

**Why Aspirin and Ibuprofen Can Inhibit the Onset of Labor?**

Aspirin and ibuprofen are antiprostaglandin drugs. Because prostaglandins stimulate uterine contractions, suppression of prostaglandin secretion by these drugs may inhibit uterine contractions, thereby inhibiting labor.

Labor can, however, have a false start. Sometimes, a very pregnant and embarrassed mother is admitted to the hospital in false labor. What has happened? She has indeed felt uterine contractions. However, these contractions—called *Braxton Hicks contractions*—are weak, irregular, and ineffectual, and they normally occur during late pregnancy. These contractions are caused by the increased responsiveness of the uterus to various hormones, particularly changing concentrations of estrogen and progesterone. The mother returns home to await the onset of true labor.

## STAGES OF LABOR

The three stages of true labor are the dilation stage, expulsion stage, and placental stage (Figure 27-6). The dilation stage begins with the onset of labor and ends with full dilation of the cervix (10 cm). This stage is characterized by rhythmic and forceful contractions, rupture of the amniotic sac (the bag of water), and cervical dilation. It is the longest stage of labor and generally lasts between 6 and 12 hours. The expulsion stage extends from complete cervical dilation to the expulsion of the fetus through the vagina (birth canal). This stage generally lasts less than 1 hour. During this stage, the mother has the urge to push with her abdominal muscles.

In a normal delivery, the head is delivered first. A head-first delivery allows the baby to be suctioned free of mucus and to breathe even before the body has fully exited from the birth canal. Because the vaginal orifice may not expand enough to deliver the baby, however, an episiotomy may be performed. An episiotomy (eh-piz-ee-OT-oh-mee) is a surgical incision into the perineum, the tissue between the vaginal opening and the anus. The incision enlarges the vaginal opening and facilitates the delivery of the baby.

The third stage is the placental stage and occurs 10 to 15 minutes after the birth. It involves the separation of the placenta from the uterine wall and expulsion of the placenta and attached membranes by forceful uterine contractions. The placenta and the attached membranes are collectively called the *afterbirth*. In addition to expelling the fetus and the placenta, the uterine contractions also cause vasoconstriction of the uterine blood vessels, thereby minimizing blood loss. Uterine contractions also help the uterus return to its nonpregnant size and shape. About 500 mL of blood is lost during delivery.

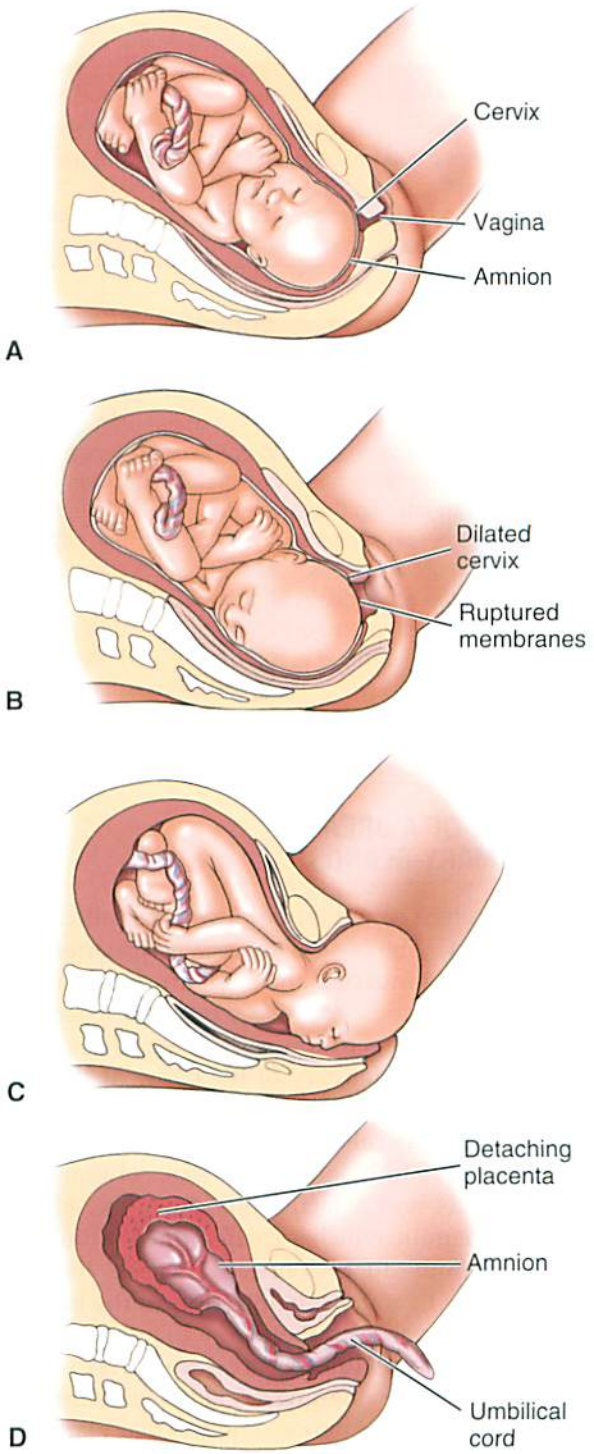



FIGURE 27-6 Stages of labor. **A,** Before labor begins. **B,** Dilation stage. **C,** Expulsion stage. **D,** Placental stage.

Sometimes, the fetus does not come out head first. Instead, another part of the body, such as the buttocks, is delivered first. This presentation is called a *breech birth*. A breech presentation makes delivery more difficult for both the mother and the infant.

There are a number of conditions that prevent a safe vaginal delivery, thereby requiring surgery. A cesarean section (C-section) refers to the delivery of the infant

through a surgical incision in the abdominal and uterine walls.

### 2+2 Sum It Up!

The birth process is called *parturition*. Labor is the forceful contractions that expel the baby and afterbirth from the uterus, through the birth canal. Labor begins in response to various hormones, particularly oxytocin. The three stages of labor are the dilation stage, the expulsion stage, and the placental stage.

### Do You Know...

**What Placenta Previa and Abruptio Placentae Are?**

Sometimes, the placenta forms too low in the uterus, near the cervix. When the cervix dilates during the later stages of pregnancy, the placenta detaches from the uterine wall, causing bleeding in the mother and depriving the fetus of an adequate supply of oxygen and nutrients. This condition is called *placenta previa*. *Abruptio placentae* refers to the premature separation of an implanted placenta at about 20 or more weeks of pregnancy. Without immediate treatment, abruptio placentae results in severe hemorrhage in the mother and death for the fetus.

## FEMALE BREAST AND LACTATION

### STRUCTURE OF A BREAST: THE MAMMARY GLANDS

The anterior chest contains two elevations called *breasts* (Figure 27-7, *A*). The breasts are located anterior to the pectoralis major muscles and contain adipose tissue and mammary glands. **Mammary glands** are accessory organs of the female reproductive system. They secrete milk following the delivery of the baby. At the tip of each breast is a nipple surrounded by a circular area of pigmented skin called the *areola*. Each mammary gland contains 15 to 20 lobes. Each lobe contains many alveolar glands and a lactiferous duct. The alveolar glands secrete milk, which is carried toward the nipple by the lactiferous duct. Connective tissue, including the suspensory ligaments, helps support the breast.

Until a child reaches puberty, the mammary glands of male and female children are similar. At puberty, however, the female mammary glands are stimulated by estrogen and progesterone. The alveolar glands and ducts enlarge, and adipose tissue is deposited around these structures. The male breast does not develop because there is no hormonal stimulus to do so. If a male is given female hormones, however, he too develops breasts.

### GOT MILK?

#### HORMONES OF LACTATION

During pregnancy, the increased secretion of estrogen and progesterone has a profound effect on the breasts. The breasts may double in size in preparation for lactation (milk production) following birth. Usually, there is no milk production during pregnancy because lactation requires prolactin, a hormone secreted by the anterior pituitary gland (see Figure 27-7, *B*). High plasma levels of estrogen and progesterone inhibit prolactin secretion during pregnancy. After delivery, however, plasma levels of estrogen and progesterone decrease, allowing the anterior pituitary gland to secrete prolactin. Milk production takes 2 to 3 days to begin. In the meantime, the mammary glands produce colostrum (koh-LOH-strohm), a yellowish watery fluid rich in protein and antibodies.

Prolactin is necessary for milk production, but a second hormone, oxytocin, is necessary for the release of milk from the breast. How does the release of milk happen? When the baby suckles, or nurses, at the breast, nerve impulses in the areola are stimulated. Nerve impulses then travel from the breast to the hypothalamus; the hypothalamus, in turn, stimulates the posterior pituitary gland to release oxytocin. The oxytocin travels through the blood to the breast, causing contraction of the smooth muscle of the lobules. This process squeezes milk into the ducts, where it can be sucked out of the nipple by the nursing infant (see Figure 27-7, *B*).

The effect of suckling and oxytocin release is called the *milk let-down reflex*. Note that the stimulus for the milk let-down reflex is suckling at the breast. Thus, nursing mothers are encouraged to suckle their infants often. Breast-feeding encourages a good flow of milk. Also note the distinction between the effects of prolactin and oxytocin. Prolactin stimulates milk production, or lactation. Oxytocin stimulates the milk let-down reflex and stimulates the flow of milk.

In addition to its effect on the flow of milk, oxytocin causes the uterus to contract. Uterine contraction helps minimize blood loss and more quickly returns the uterus to its nonpregnant state. (The return of the uterus to its nonpregnant size is called *involution*.) A class of drugs called *oxytocic agents* are sometimes administered to the mother after childbirth. Like oxytocin, these drugs (ergot preparations) cause uterine contractions and minimize postpartum bleeding.

### ? Re-Think

1. List three pregnancy-induced physiological adjustments.
2. Differentiate between the effects of prolactin and oxytocin with regard to lactation.
3. What is colostrum?

## POSTNATAL CHANGES AND DEVELOPMENTAL STAGES

### IMMEDIATE ADJUSTMENTS

Immediately after birth, the baby must make many important adjustments to survive. Most importantly,

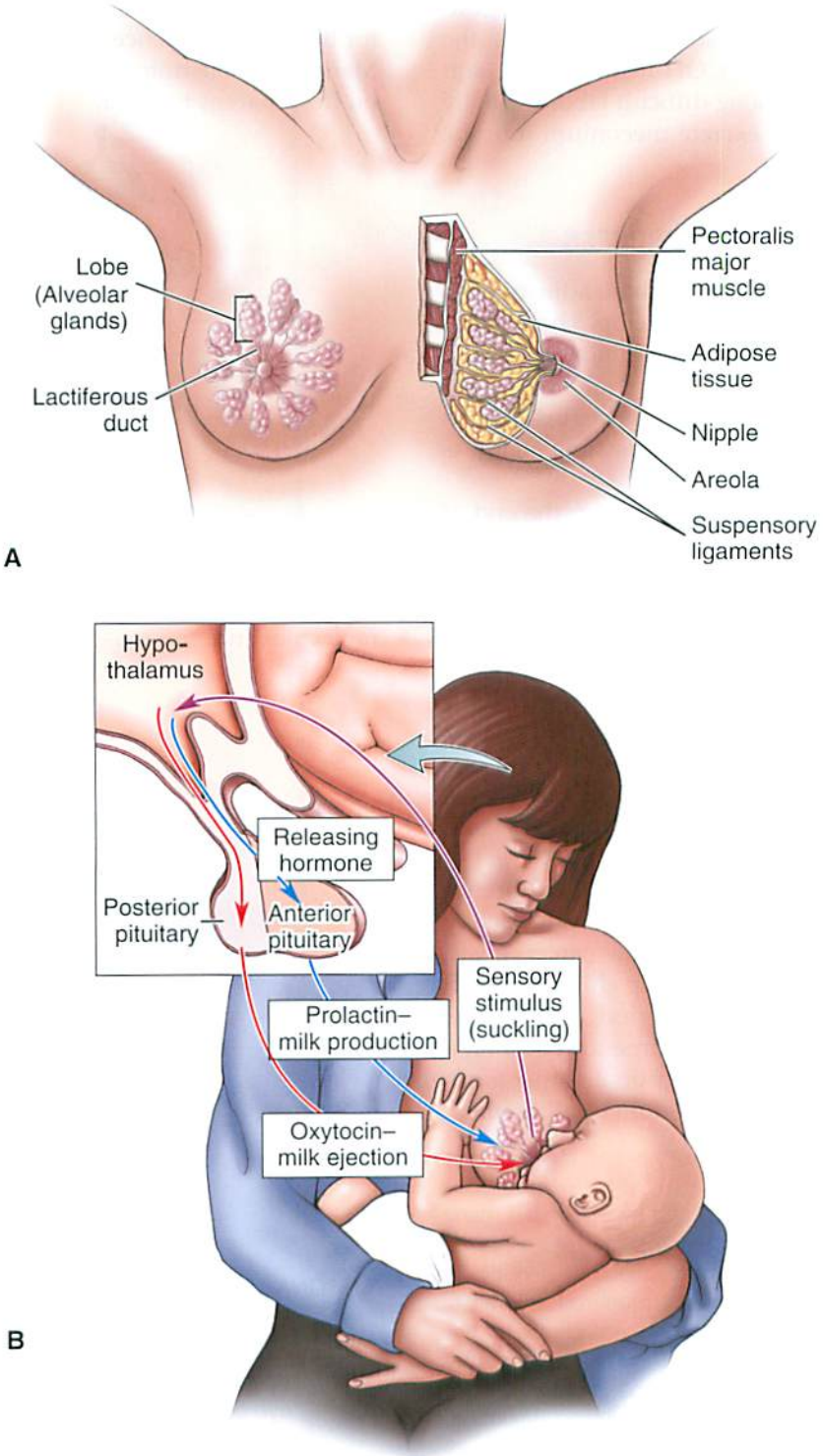



FIGURE 27-7 **A,** Breast and mammary glands. **B,** Hormones involved in breast-feeding; the milk let-down reflex.

the baby must begin breathing. The first deep breaths, drawn as the baby cries, expand the lungs and provide the infant with life-giving oxygen. The cardiovascular system also makes major adjustments, the most important being the establishment of blood flow to the lungs. Pulmonary blood flow occurs when the fetal heart structures, such as the foramen ovale and ductus arteriosus, close. (Review fetal circulation in Chapter 18.)

Other organ systems continue functioning as they did before birth. For example, the kidneys, having once secreted their urine into the amniotic fluid, become diaper-seeking organs, and the digestive

system continues its lifelong career of eating, digesting, and excreting. The first stool produced by the newborn—meconium (meh-COH-nee-um)—is soft and dark green. (During a long difficult labor the baby may become stressed and excrete meconium into the amniotic fluid—not good.)

## DEVELOPMENT AS A LIFELONG PROCESS

After the newborn makes the immediate adjustments, the infant continues to grow and develop. Throughout life, the person will pass through the following developmental stages:

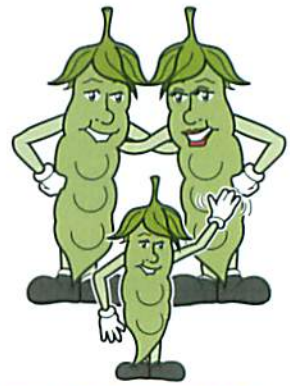
- *Neonatal period.* The neonatal period begins at birth and lasts for 4 weeks. During this time, Baby is called a *neonate,* or *newborn.*
- *Infancy.* The period of infancy lasts from the end of the first month to the end of the first year. Baby's first birthday marks the end of this stage.
- *Childhood.* The period of childhood lasts from the beginning of the second year to puberty.
- *Adolescence.* The period of adolescence lasts from puberty to adulthood. One word characterizes this stage: hormones. The period of adolescence is a period of tremendous growth and upheaval. Adolescents are physically capable of reproduction. The teen moves toward adulthood, leaving behind childish ways and coming to grips with becoming an adult.
- *Adulthood.* Adulthood is the period from adolescence to old age. During this period, the person is usually concerned with family matters and career goals. (Most of us are still trying to give up childish ways!)
- *Senescence.* Senescence is the period of old age, ending in death. It is not only a time to reflect on a life well lived, but also a time to pursue other goals and to pass on the collected wisdom of a lifetime.

### 2+2 Sum It Up!

After the birth of the baby, both baby and mother make many physiological adjustments. The mother's body returns to its nonpregnant state. For example, cardiac output and blood volume decrease. She is physiologically prepared to breastfeed. Through the actions of prolactin and oxytocin, her mammary glands are producing milk and making it readily available to the suckling infant. The baby has made an initial adjustment to life on the outside and is breathing, urinating, and eating. The newborn continues postnatal development as a neonate. From there, it is on to infancy, childhood, adolescence, adulthood, and senescence.

## HEREDITY

"He has his father's nose and his mother's smile." How often have we made that type of statement when we recognize the traits of a parent in a child? The transmission of characteristics from parent to child is called *heredity,* and the science that studies heredity is called *genetics.* The work of an Austrian monk, Gregor Mendel, in the early nineteenth century paved the way for the modern science of genetics. Using garden peas, Mendel demonstrated a pattern of specific traits passed on from parent to child.



## DNA, GENES, AND CHROMOSOMES

How are genetic structures related? Genetic information is located in the deoxyribonucleic acid (DNA) molecule and, more specifically, in the DNA base

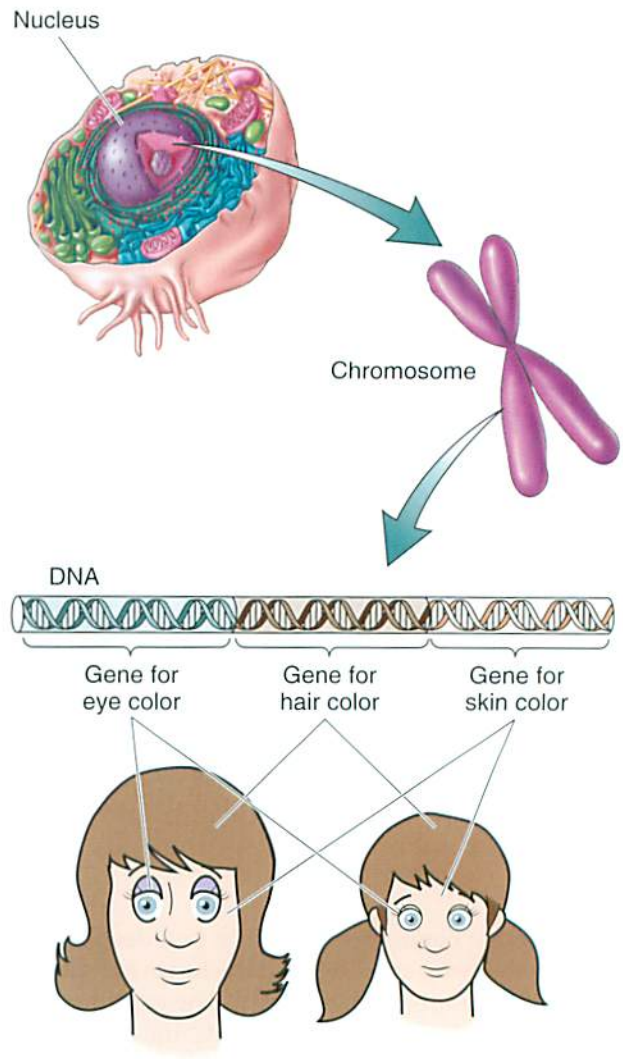



FIGURE 27-8 Chromosomes and genes.

**Table 27-3** Examples of Genetic Traits

| TRAIT | DOMINANT | RECESSIVE |
|---|---|---|
| Hairline | Widow's peak | Continuous hairline |
| Hair color | Dark | Light |
| Hair texture | Curly | Straight |
| Hair on back of hand | Present | Absent |
| Freckles | Present | Absent to few |
| Dimples | Present | Absent |
| Eye color | Dark | Light |
| Color vision | Normal | Color-blind |
| Ear lobes | Unattached | Attached |
| Cleft chin | Present | Absent |
| Rh factor | Present | Absent |

sequencing (Figure 27-8; see Chapter 4). DNA is tightly wound into threadlike structures called **chromosomes** found in the nucleus of most cells in the body. **Genes** are segments of the DNA strand and carry information for a specific trait such as skin color, freckles, and blood type. See Table 27-3 for other genetically determined characteristics. Some traits are determined by a single pair of genes, whereas other traits, such as height, require input from several genes. Each chromosome may carry thousands of genes, and each gene occupies a specific position on a chromosome.

Chromosomes exist in pairs. With the exception of the sex cells (egg and sperm), there are 23 pairs, or 46 chromosomes, in almost all human cells (red blood cells are the other exception). During fertilization, the egg and sperm each contribute 23 chromosomes to the zygote for a total of 23 pairs, or 46 chromosomes. One member of each pair comes from the egg; the other comes from the sperm. Thus, for each trait, genetic instructions come from both the mother and the father. Forty-four chromosomes (22 pairs) are called *autosomes* (AW-toh-sohms). The autosomal gene pairs are numbered from 1 to 22. Two (one pair) of the 46 chromosomes are sex chromosomes; each is either an X or Y chromosome.

**? Re-Think**

What happens to the chromosomal number during meiotic cell division? Why is this important?

## GENETIC ART: THE KARYOTYPE

It is possible to photograph the chromosomes in the cell. The photograph of the chromosomes is then cut apart, and the chromosomes are arranged in pairs by size and shape. The resulting display of the paired chromosomes is called a **karyotype** (KAIR-ee-oh-type) (Figure 27-9). This genetic artwork displays 22 pairs of autosomes and one pair of sex chromosomes. The karyotype is a diagnostic tool. It can reveal structural abnormalities and errors in the numbers of chromosomes.

## DOMINANT, RECESSIVE, AND CODOMINANT GENES

Remember that each cell inherits two genes for each trait—tall or short, straight nose or curved nose, stubby fingers or long fingers, dark eyes or light eyes. Hence, the choice. Will it be long or short, or curved or straight? Genes can be dominant, recessive, or codominant. A dominant gene expresses itself; it gets noticed. The dominant gene overshadows the recessive gene and keeps it unnoticed, or unexpressed. Thus, a recessive gene is not expressed if it is paired with a dominant gene. For example, the genes for dark eyes are dominant, whereas the genes for light eyes are recessive. If the dominant genes (dark) and the recessive (light) genes are paired, the genes for dark eyes will be expressed (Figure 27-10). The genes for light eyes will not be expressed. Codominant genes express a trait equally. AB blood type is an example of codominance.

If recessive genes carry light eye coloring, how can an offspring develop blue eyes? Although blue eye coloring is recessive, a baby develops blue eyes because both the mother and father are carrying the genes for blue eyes, a recessive trait (see Figure 27-10). If either the mother or father had passed on a dominant gene for dark eyes, the child would have brown eyes.

The question can be put another way. If I have brown eyes, can any of my children have blue eyes? Yes! If I am carrying both a dominant (brown) and recessive (blue) gene for eye color, my child has a chance of having blue eyes. My recessive gene might pair with a recessive gene from my mate. If so, the pairing of two recessive genes produces a blue-eyed offspring. Brown-eyed me is a carrier, one who shows no evidence of a trait (like blue eyes) but carries a recessive gene for that trait.

## TOO MANY OR TOO FEW CHROMOSOMES

A person normally inherits 22 pairs of autosomal chromosomes. Sometimes, however, a person inherits too many or too few autosomal chromosomes. The most common autosomal abnormality is called *trisomy 21*, or *Down syndrome*. A child with trisomy 21 has three copies of chromosome 21 instead of two copies. Other types of trisomy occur very infrequently in live births. Trisomy 18, called *Edwards syndrome*, is caused by three copies of chromosome 18, whereas trisomy 13, called *Patau syndrome*, is caused by three copies of chromosome 13. Autosomal abnormalities are usually caused by nondisjunction.

Nondisjunction is the failure of the chromosomes to separate during meiosis, thereby causing the

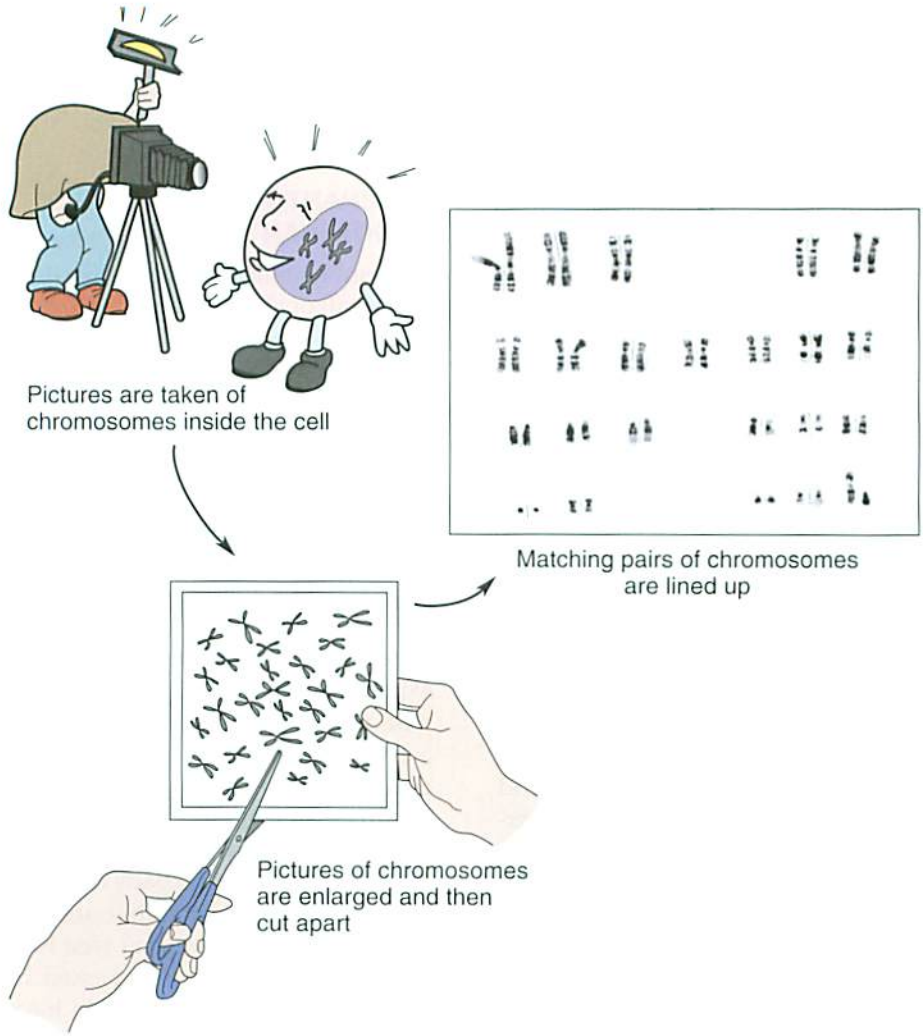



FIGURE 27-9 Genetic art: the karyotype.

formation of eggs or sperm with too many or too few chromosomes. If these eggs and sperm lead to pregnancy, the embryo may have one too many or too few chromosomes. Most pregnancies with an unbalanced number of chromosomes miscarry in the first trimester. Down syndrome is the most common chromosomal abnormality because the condition is least likely to cause the pregnancy to miscarry. Even so, an estimated 70% of pregnancies with Down syndrome spontaneously miscarry, usually in the first trimester.

## GENETIC EXPRESSION

Genetic expression determines what the offspring looks like. A person's genetic expression can be influenced by a number of factors, including the person's gender, the influence of other genes, and environmental conditions. For example, certain types of baldness and color blindness may be inherited by males and females, but these traits are more apt to appear in the male. A child may also have the genetic capability of growing very tall. If the child is deprived of adequate nutrition and exercise, however, that child may not grow as tall as the genetic makeup has predicted.

## GENETIC MUTATIONS

Normally, DNA replicates all information with few mistakes. Because of this precision, information is passed along reliably and efficiently to the next generation. Sometimes, however, a change occurs in a gene, or a chromosome breaks in some unexpected way. The result may be a unique feature or a birth defect. This change in the genetic code is called a *mutation*. Some mutations occur spontaneously; others are caused by mutagenic agents, such as chemicals, drugs, and radiation. Mutations can be beneficial or harmful and may even cause the death of the offspring. For example, a mutation in the cells of the immune system may render a child resistant to a particular disease, thereby enhancing health. Another mutation, however, may weaken the immune system, making it more susceptible to pathogens.

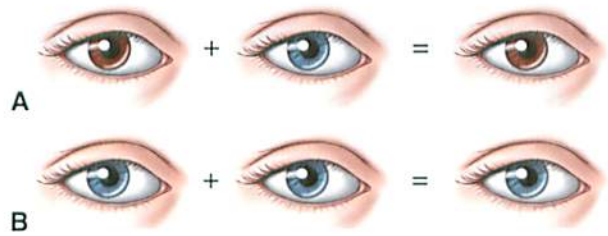



FIGURE 27-10 Eye color, with dominant and recessive genes. **A,** The dominant brown-eye gene is expressed over the recessive blue-eye gene. **B,** Two recessive blue-eye genes produce a blue-eyed offspring.

**Re-Think**

Define *nondisjunction*, *trisomy*, and *genetic mutation*.

## IT'S A BOY, IT'S A GIRL: HOW THE SEX OF THE CHILD IS DETERMINED

### Xs AND Ys

Each human cell has 22 pairs of autosomal chromosomes and one pair of sex chromosomes (X and Y chromosomes). The female has two X chromosomes in her cells, a pair designated XX. A male has both an X and Y chromosome in his cells, a pair designated XY (Figure 27-11).

### SEX DETERMINATION: A MALE THING

The sex cells, the egg and the sperm, divide by a special type of cell division called *meiosis*. The important step in meiosis is the reduction (by half) of the chromosomes. In other words, meiosis reduces the numbers of chromosomes from 46 to 23. The meiotic cell reduction also reduces the numbers of sex chromosomes by half. Consequently, each egg contains one X chromosome, and the sperm contains either an X chromosome or a Y chromosome. If a sperm containing an X chromosome fertilizes an egg, the child has an XX sex chromosome pair and is therefore female. If a sperm containing a Y chromosome fertilizes an egg, the child has an XY chromosome pair and is therefore male. Thus, the sperm (male) determines the sex of the child.

Sometimes, a person inherits an abnormal number of sex chromosomes. For example, a female with Turner syndrome inherits only one sex chromosome, an X chromosome. Turner syndrome is designated as XO. The X signifies the female chromosome; the O signifies the absence of the second sex chromosome. A child with Turner syndrome does not develop secondary sex characteristics, is shorter than average, and has a webbed neck. People with Turner syndrome have normal intelligence.

**Re-Think**

Explain why the father determines the gender of the child.

### SEX-LINKED TRAITS

The X and Y chromosomes differ structurally. The female X chromosome is larger than the Y chromosome and carries many genes for traits in addition to determining sex. The male Y chromosome is much smaller than the X chromosome and does not carry as much genetic information as the X chromosome. Any trait that is carried on a sex chromosome is called a **sex-linked trait**. Because the X chromosome has more genetic information than the Y chromosome, most sex-linked traits are carried on the female X chromosome. Sex-linked traits carried on the X chromosomes are also called *X-linked traits*.

Although most sex-linked traits are carried on the X chromosome, they are expressed, or appear, in the male. Sex-linked diseases include hemophilia, Duchenne muscular dystrophy, and fragile X syndrome. Less serious sex-linked traits include baldness and red–green color blindness.

## CONGENITAL AND HEREDITARY DISEASE

You need to distinguish between hereditary diseases and congenital diseases and defects. Hereditary diseases are genetically transmitted; congenital conditions are present at the time of birth. Congenital disorders include those that are inherited and those that are not. For example, hemophilia is genetically transmitted and is therefore inherited. Because hemophilia is present at birth, it is also congenital.

A disease can, however, be congenital but not inherited. For example, a mother may give birth to an infant who was exposed to the rubella virus (German measles) during her first trimester of pregnancy. This child may be born with cardiovascular, ocular, and neural tube defects. These defects were not transmitted genetically from the parents to the child and are therefore not hereditary. The defects are congenital, however, because they were present at birth. Only 15% of congenital defects have a known genetic cause. For 70% of congenital birth defects, the cause is unknown.

Gene therapy offers hope for the treatment and eventual cure of genetic disorders. *Gene therapy* refers to the insertion of normal genes into cells that have abnormal genes. For example, a person with congenitally high cholesterol levels might be successfully treated with genes that code for normal cholesterol production. Although still in its early experimental stages, gene therapy provides hope for those with such genetic conditions as sickle cell disease, cystic fibrosis, and muscular dystrophy.

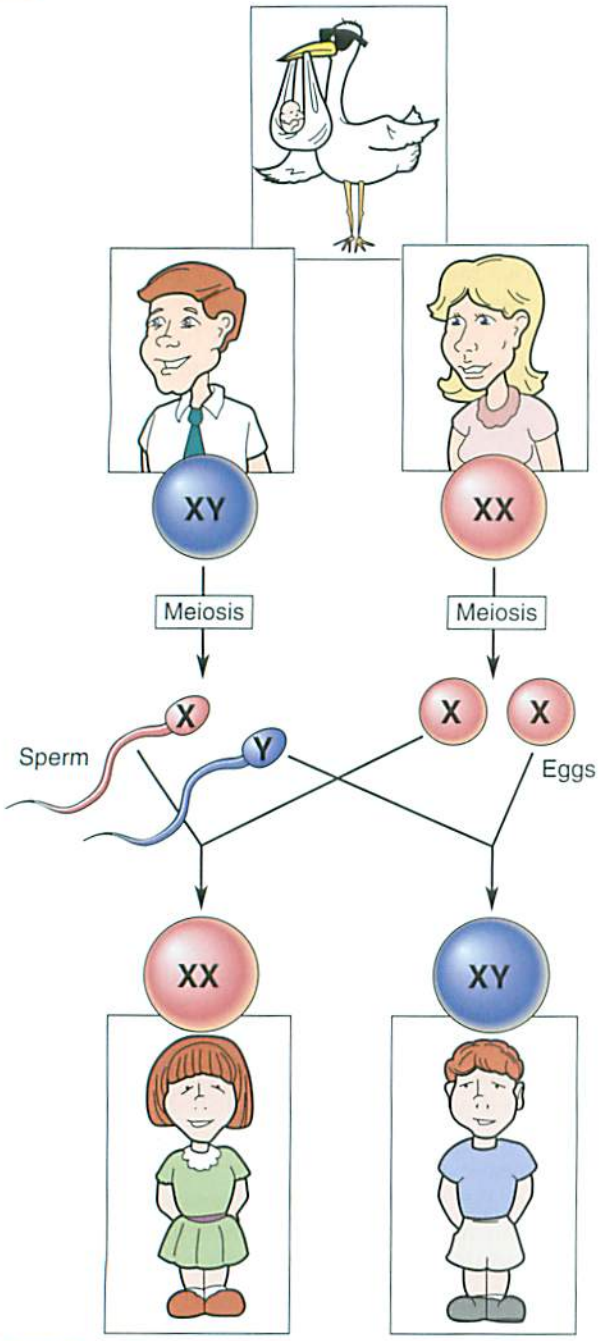



FIGURE 27-11 Determination of sex; Xs and Ys.

## Re-Think

1. What is a sex-linked trait?
2. Explain how a disorder can be congenital but not hereditary.

## 2+2 Sum It Up!

A child resembles the parents because he or she inherits genetic information from each parent. Genetic information is stored in the DNA molecules, which are arranged in strands called *chromosomes*. Almost every human cell contains 22 pairs of autosomal chromosomes and one pair of sex chromosomes. Genes are segments of DNA that contain codes for specific traits such as eye color or blood type. The child receives two genes for each inherited trait—one from the mother and one from the father.

Genes are dominant, recessive, or codominant. The sex chromosomes are designated X and Y. At fertilization, an XX combination produces a female child, whereas an XY combination produces a male. The father, with his Y chromosome, determines the sex of the child. Genetic information is passed along efficiently and reliably. Occasionally, incorrect or unhealthy information is passed along, thereby producing genetic diseases.

*Note: The Medical Terminology and Disorders table related to this chapter appears in Chapter 26.*

# Get Ready for Exams!

## Summary Outline

The purpose of the reproductive system is to produce offspring whose genetic information is faithfully transmitted from generation to generation.

**I. Fertilization to Birth**

A. Fertilization
   1. *Fertilization* (conception) refers to the union of an egg and a sperm, called a *zygote*.
   2. Fertilization takes place around the time of ovulation.
   3. Fertilization normally occurs within the fallopian tube.

B. Human development: begins with fertilization and ends with death

C. Prenatal development
   1. Prenatal development processes: cleavage, growth, morphogenesis, and differentiation
   2. Prenatal period: early embryonic, embryonic, and fetal periods
      a. The early embryonic period lasts for 2 weeks after fertilization. The zygote develops and

implants into the uterine endometrial lining; its trophoblastic cells secrete hCG (maintains the corpus luteum).
   b. There are two types of twins: monozygotic (identical) twins and dizygotic or fraternal (nonidentical) twins.
   c. The embryonic period lasts for 6 weeks and involves the formation of the extraembryonic membranes, the placenta, and all the organ systems.
   d. The four extraembryonic membranes are the amnion, chorion, yolk sac, and allantois.
   e. The placenta develops as the chorionic villi of the embryo burrow into the endometrial lining of the uterus.
   f. The placenta has two functions: the site of "exchange" and glandular function.
   g. The embryo is hooked up to the placenta by the umbilical cord.
   h. The embryonic period is a period of organogenesis.
   i. The fetal period extends from week 9 to birth; it is a time of growth and maturation.
   j. Hormonal changes during pregnancy are summarized in Table 27-1.

D. Birth (parturition)
   1. Labor is the forceful contractions that expel the fetus from the uterus.
   2. Labor is caused by hormones.

E. Breasts and lactation
   1. The breasts contain mammary (milk-secreting) glands and adipose tissue.
   2. Hormonal control of breast function: prolactin and oxytocin (milk let-down reflex)

**II. Postnatal Development**

A. Immediate postnatal changes
   1. The baby takes a first breath and the cardiovascular system makes some major adjustments.
   2. Baby is no longer eating, drinking, and breathing at the placenta.

B. Developmental periods: neonatal period, infancy, childhood, adolescence, adulthood, and senescence

**III. Heredity**

A. DNA, genes, and chromosomes
   1. Genetic information is stored in the DNA of genes, which are arranged into chromosomes.
   2. Chromosomes exist in pairs. With the exception of sex cells, most human cells have 23 pairs of chromosomes, or 46 chromosomes.
   3. Genes are dominant, recessive, or codominant.
   4. The sex chromosomes are designated X and Y. A female has two X chromosomes and a male has one X chromosome and one Y chromosome. Because only the male has the Y chromosome, the father determines the sex of the child.
   5. Sex-linked traits are carried on the sex chromosomes. Most are carried on the X chromosome and are therefore also called *X-linked traits*.

B. Congenital and hereditary diseases
   1. A congenital disorder is a condition present at birth.
   2. Congenital disorders include inherited and noninherited birth defects and diseases. A hereditary disease is transmitted genetically from parent to child.

## Review Your Knowledge

### Matching: Fertilization and Development

Directions: Match the following words with their descriptions below.

**a.** primary germ layers
**b.** fetus
**c.** zygote
**d.** fertilization
**e.** embryo
**f.** parturition
**g.** trophoblasts
**h.** placenta
**i.** implantation
**j.** umbilical cord

**1.** ___ The fertilized ovum
**2.** ___ Cells that secrete hCG
**3.** ___ A zygote-making event
**4.** ___ The site where Baby breathes, eats, and excretes
**5.** ___ Baby's lifeline; contains the umbilical blood vessels
**6.** ___ Baby's name at age 3 to 8 weeks
**7.** ___ Process whereby the blastocyst baby-to-be burrows into the endometrium
**8.** ___ The birth process
**9.** ___ Endoderm, mesoderm, and ectoderm
**10.** ___ Baby's name from 9 weeks to birth

### Matching: Hormones

Directions: Match the following words with their descriptions below. Some words may be used more than once.

**a.** hCG
**b.** prolactin
**c.** oxytocin
**d.** aldosterone

**1.** ___ Hormone that stimulates the mammary glands to make milk
**2.** ___ Posterior pituitary hormone involved in the milk let-down reflex
**3.** ___ Hormone that sustains the corpus luteum
**4.** ___ Hormone that stimulates the contraction of the myometrium
**5.** ___ Secretion of this hormone continues at a high level for about 2 months and then steadily declines as the placenta takes over.
**6.** ___ Hormone that stimulates $Na^+$ reabsorption and expands blood volume during pregnancy

### Matching: Heredity

Directions: Match the following words with their descriptions on the next page.

**a.** sex-linked trait
**b.** mutation
**c.** autosomes
**d.** sex chromosomes
**e.** genes

1. ___ Segments of a DNA strand that carry the code for a specific trait, such as eye color
2. ___ Twenty-two pairs (numbered 1 to 22) of chromosomes
3. ___ X and Y chromosomes
4. ___ Any trait that is carried on an X or Y chromosome
5. ___ A change in the genetic code that may express itself as a change in a particular trait

### Multiple Choice

1. Implantation
   a. normally occurs within the fallopian tubes.
   b. is achieved by the morula.
   c. is a uterine event achieved by the blastocyst.
   d. occurs within the ovaries.
2. Human chorionic gonadotropin (hCG)
   a. promotes the maturation of the egg.
   b. is responsible for female characteristics.
   c. maintains the corpus luteum.
   d. promotes the transformation of the corpus luteum into the corpus albicans.
3. Trophoblastic cells
   a. secrete oxytocin.
   b. are responsible for the milk let-down reflex.
   c. assist with implantation.
   d. are incorporated within the graafian follicle.
4. The morula
   a. only is formed if there is an ectopic pregnancy.
   b. is the unfertilized ovum that gets discharged with the menstrual blood.
   c. refers to the pre-embryonic cluster of cells.
   d. spends its life embedded within the endometrium.
5. Which of the following is least true of the placenta?
   a. Is the site at which the baby-to-be breathes
   b. Is very vascular
   c. Replaces the glandular secretion of the corpus luteum
   d. Nourishes the zygote as it matures into the morula
6. The chorion is an extraembryonic membrane that
   a. develops finger-like projections that penetrate the uterine wall forming the placenta.
   b. forms the inner cell mass.
   c. secretes the hormones that initiate labor.
   d. secretes vernix caseosa that covers the fetus.
7. The myometrium
   a. sloughs during menstruation.
   b. is responsive to oxytocin and oxytocic drugs.
   c. is the uterine lining that is most responsive to estrogen and progesterone.
   d. forms finger-like projections that penetrate the uterine lining forming the placenta.
8. Which of the following happens first?
   a. Zygote formation
   b. Fertilization
   c. Ovulation
   d. Early blastocyst
9. Prolactin and oxytocin
   a. are secreted by the adenohypophysis.
   b. stimulate the milk let-down reflex.
   c. stimulate the mammary glands to make milk.
   d. target the mammary glands.
10. Colostrum is
    a. secreted by the mammary glands.
    b. located within the amniotic sac.
    c. the vernix caseosa that covers the fetus.
    d. a hormone that initiates labor and terminates pregnancy.

## Go Figure

1. **According to Figure 27-1**
   a. The morula splits to form a zygote.
   b. A zygote forms within the fallopian tube.
   c. The late blastocyst is a fallopian tube dweller.
   d. Implantation is an ovarian event.
2. **According to Figure 27-1**
   a. Repeated mitosis of the zygote forms a morula.
   b. Trophoblastic cells, located in the ovary, assist in the implantation of the embryo.
   c. Fertilization usually occurs within the corpus luteum.
   d. Trophoblastic cells secrete LH, which is responsible for ovulation.
3. **According to Figures 27-2 and 27-3**
   a. The chorionic villi are located within the umbilical cord.
   b. Figure 27-2, *C*, illustrates the procedure for amniocentesis.
   c. None of the extraembryonic membranes are illustrated in Figure 27-2, *A*.
   d. The chorionic villi interact with the maternal uterine lining to form the placenta.
4. **According to Figure 27-7**
   a. Alveolar glands secrete milk into the lactiferous ducts.
   b. Oxytocin is secreted reflexively in response to suckling.
   c. Prolactin stimulates the breast to make milk.
   d. All of the above are true.
5. **According to Figure 27-7**
   a. The stimulus for the milk let-down reflex is the level of prolactin in the mother's blood.
   b. Oxytocin is a posterior pituitary hormone that causes milk to flow in response to suckling.
   c. Prolactin is a neurohypophyseal hormone that mediates the milk let-down reflex.
   d. All of the above are true.
6. **According to Figure 27-11**
   a. The father determines the gender of the offspring.
   b. The father provides sperm that only carry the Y chromosome.
   c. The mother provides ova that only carry the Y chromosome.
   d. An infant with an X and Y chromosome is female.