CHAPTER 2

Biological and Environmental Foundations and Prenatal Development

Learning Objectives

2.1 Describe the process of cell reproduction and patterns of genetic inheritance.

2.2 Define and provide examples of genetic disorders and chromosomal abnormalities.

2.3 Explain how the dynamic interactions of heredity and environment influence development.

2.4 Discuss the stages of prenatal development, stages of childbirth, and challenges for infants at risk.

2.5 Identify the principles of teratology, types of teratogens, and ways that teratogens can be used to predict prenatal outcomes.

Digital Resources

audio Growth Hormone

Lives in context Fostering Gross Motor Skills in Early Childhood

web Brain-Based Education (p. 172)

FPO

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"Roger and Ricky couldn't be more different," marveled their mother. "People are surprised to find out they are brothers." Roger is tall and athletic, with blond hair and striking blue eyes. He spends most afternoons playing ball with his friends and often invites them home to play in the yard. Ricky, two years older than Roger, is much smaller, thin and wiry. He wears thick glasses over his brown eyes that are nearly as dark as his hair. Unlike his brother, Ricky prefers solitary games and spends most afternoons at home playing video games, building model cars, and reading comic books. How can Roger and Ricky have the same parents and live in the same home yet differ markedly in appearance, personality, and preferences? In this chapter, we discuss the process of genetic inheritance and principles that can help us to understand how members of a family can share a great many similarities—and many differences. We also examine the process by which a single cell containing genes from two biological parents develops over a short period of time into an infant.

GENETIC FOUNDATIONS OF DEVELOPMENT

LO 2.1 Describe the process of cell reproduction and patterns of genetic inheritance.

Although Roger is quite different from his older brother, Ricky, he shares so many of his father's characteristics that most people comment on the strong physical resemblance. In other ways, however, Roger is more like his highly sociable mother. Ricky also shares similarities with each of his parents: In physical appearance, he resembles his mother and her brothers, but his quiet personality is similar to that of his father. Most of us learn early in life, and take it for granted, that children tend to resemble their parents. But to understand just how parents transmit their inborn characteristics and tendencies to their children, we must consider the human body at a cellular level.

GENETICS

The human body is composed of trillions of units called cells. Within each cell is a nucleus that contains 23 matching pairs of rod-shaped structures called **chromosomes** (Plomin, DeFries, Knopik, & Neiderhiser, 2013). Each chromosome holds the basic units of heredity, known as genes, composed of stretches of **deoxyribonucleic acid (DNA)**, a complex molecule shaped like a twisted ladder or staircase. The 20,000 to 25,000 genes that reside within our chromosomes are the blueprint for creating all of the traits that organisms carry (Barlow-Stewart, 2012; Finegold, 2013). People around the world share 99.7% of their genes (Watson, 2008). Although all humans share the same basic genome, or set of genetic instructions, every person has a slightly different code, making him or her genetically distinct from other humans.

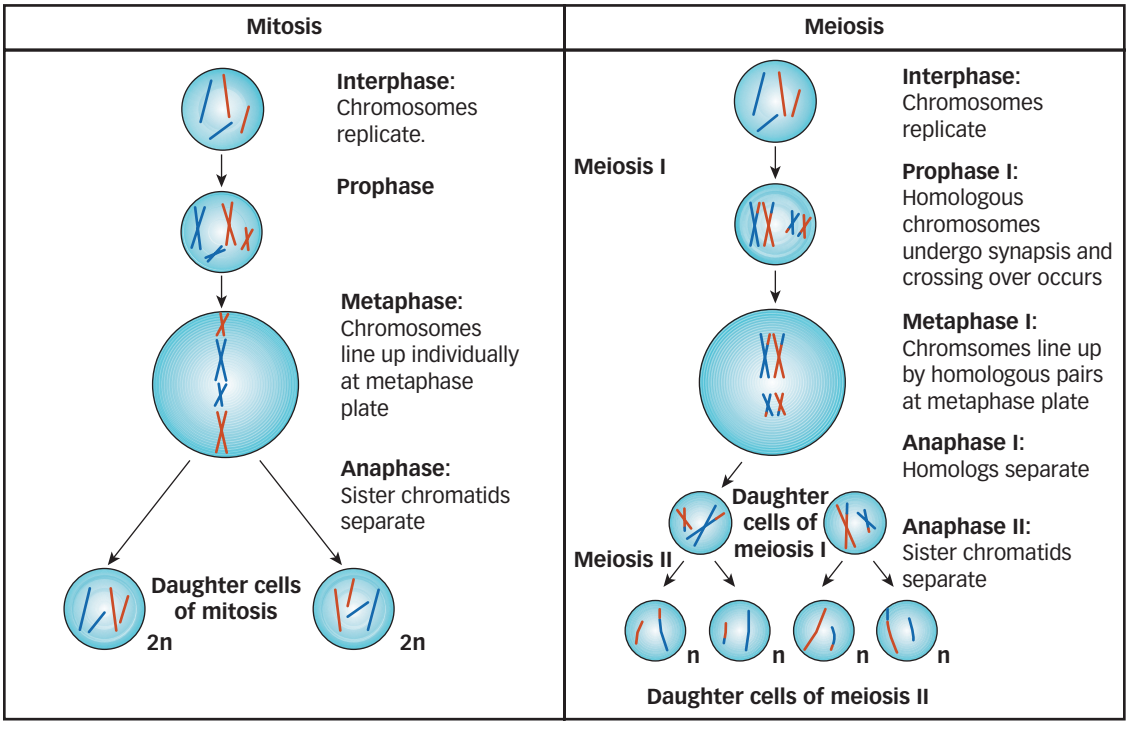
Cell Reproduction

Most cells in the human body reproduce through a process known as **mitosis,** in which DNA replicates itself, permitting the duplication of chromosomes and, ultimately, the formation of new cells with identical genetic material (Sadler, 2015). Sex cells reproduce in a different way, called **meiosis,** which results in gametes (sperm in males and ova in females; see Figure 2.1). Gametes each contain 23 chromosomes (one-half of the 46 chromosomes, or 23 pairs, present in body cells). This permits the joining of sperm and ovum at fertilization to produce a fertilized egg, or **zygote**, with 46 chromosomes forming 23 pairs, half from the biological mother and half from the biological father. Each gamete has a unique genetic profile. It is estimated that individuals can produce millions of versions of their own chromosomes (National Library of Medicine, 2013).

As shown in Figure 2.2, 22 of the 23 pairs of chromosomes are matched; they contain similar genes in almost identical positions and sequence, reflecting the distinct genetic blueprint of the biological mother and father. The 23rd pair are sex chromosomes that specify the biological sex of the individual. In females, sex chromosomes consist of two large X-shaped chromosomes (XX). Males' sex chromosomes consist of one large X-shaped chromosome and one much smaller Y-shaped chromosome (XY; Moore & Persaud, 2016; Plomin et al., 2013).

Because females have two X sex chromosomes, all ova contain one X sex chromosome. Males' sex chromosome pair includes both X and Y chromosomes. Therefore, one half of the sperm males produce contains an X chromosome and one half contains a Y. Whether the fetus develops into a boy or girl is determined by which sperm fertilizes the ovum. If the ovum is fertilized by a Y sperm, a male fetus will develop, and if the ovum is fertilized by an X sperm, a female fetus will form, as shown in Figure 2.3.

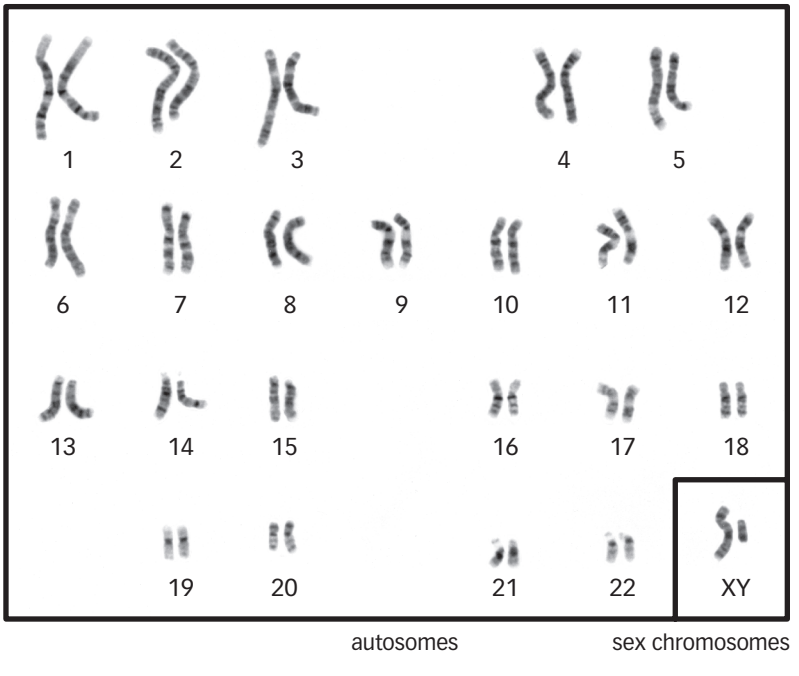
FIGURE 2.1: Meiosis and Mitosis



Genes Shared by Twins

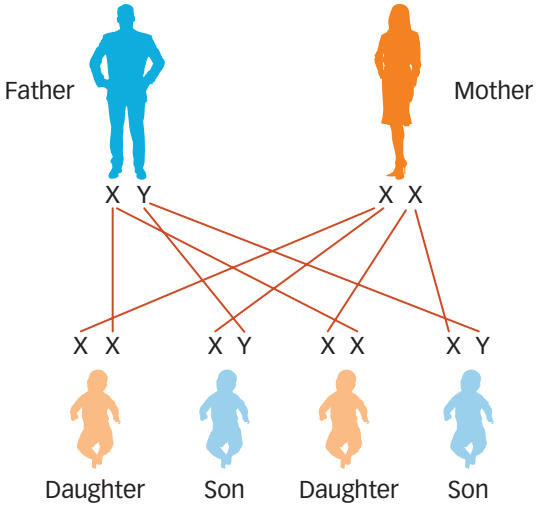
Twins are siblings who share the same womb. Twins occur in about 1 out of every 30 births in the United States (Martin, Hamilton, & Osterman, 2012). About two-thirds of naturally conceived twins are **dizygotic (DZ) twins**, or fraternal twins, conceived when a woman releases more than one ovum and each is fertilized by a different sperm. DZ twins share about one-half of their genes and, like other siblings, most fraternal twins differ in appearance, with different hair color, eye color, and height. In about half of fraternal twin pairs, one twin is a boy and the other a girl. DZ twins tend to run in families, suggesting a genetic component that controls the tendency for a woman to release more than one ovum each month. However, rates of DZ twins also increase with in vitro fertilization, maternal age, and with each subsequent birth (Fletcher, Zach, Pramanik, & Ford, 2012; Martin et al., 2012).

FIGURE 2.2: Chromosomes



Monozygotic (MZ) twins, or identical twins, originate from the same zygote, sharing the same genotype with identical instructions for all physical and psychological characteristics. MZ twins occur when the zygote splits into two separate but identical zygotes that develop into two infants. It is estimated that MZ twins occur in 4 of every 1,000 U.S. births (Fletcher et al., 2012). The causes of MZ twinning are not well understood. Temperature fluctuations are associated with

FIGURE 2.3: Sex Determination



AP/Charlie Neibergall

PHOTO 2.1: Genes Shared by Twins
Monozygotic, or identical, twins share 100% of their DNA.

MZ births in animals, but it is unknown whether similar effects occur in humans (Aston, Peterson, & Carrell, 2008). In vitro fertilization and advanced maternal age (35 and older) may increase the occurrence of MZ twins (Aston et al., 2008; Knopman et al., 2014).

PATTERNS OF GENETIC INHERITANCE

Although the differences among various members of a given family may appear haphazard, they are the result of a genetic blueprint unfolding. Researchers are just beginning to uncover the instructions contained in the human genome, but we have learned that traits and characteristics are inherited in predictable ways.

Dominant–Recessive Inheritance

Lynn has red hair while her brother, Jim, does not—and neither do their parents. How did Lynn end up with red hair? These outcomes can be explained by patterns of genetic inheritance, how the sets of genes from each parent interact. As we have discussed, each person has 23 pairs of chromosomes, one pair inherited from the mother and one from the father. The genes within each chromosome can be expressed in different forms, or *alleles*, that influence a variety of physical characteristics. When alleles of the pair of chromosomes are alike with regard to a specific characteristic, such as hair color, the person is said to be **homozygous** for the characteristic and will display the inherited trait. If they are different, the person is **heterozygous**, and the trait expressed will depend on the relations among the genes (Moore & Persaud, 2016; National Center for Biotechnology Information, 2004). Some genes are passed through **dominant–recessive inheritance**, in which some genes are dominant and are always expressed regardless of the gene they are paired with. Other genes are recessive and will be expressed only if paired with another recessive gene (see Table 2.1).

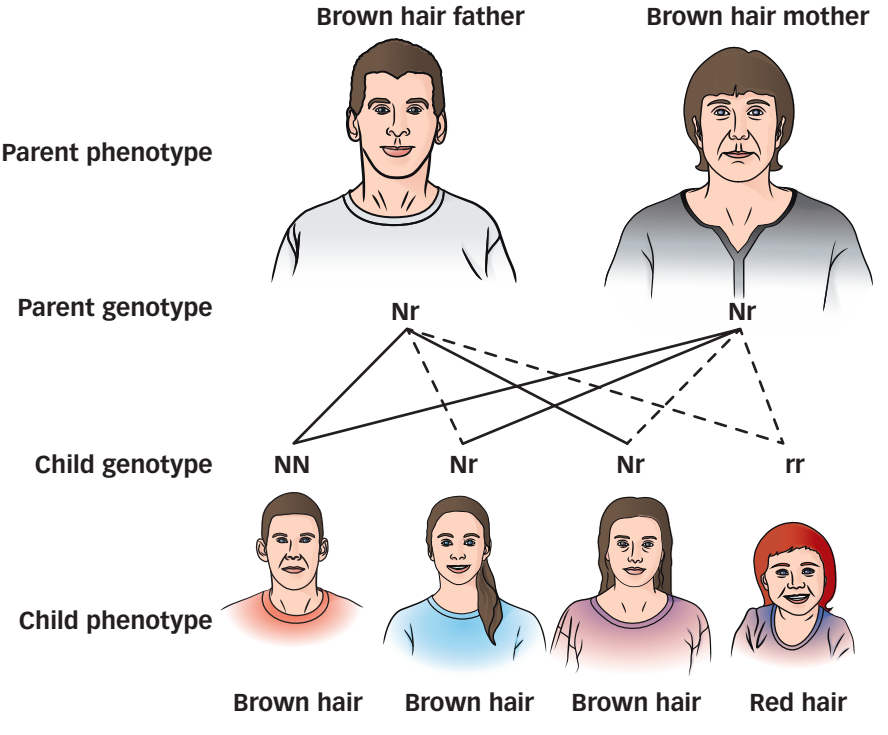
TABLE 2.1 • Dominant and Recessive Characteristics

DOMINANT TRAIT	RECESSIVE TRAIT
Dark hair	Blond hair
Curly hair	Straight hair
Hair	Baldness
Non-red hair	Red hair
Facial dimples	No dimples
Brown eyes	Blue, green, hazel eyes
Second toe longer than big toe	Big toe longer than second toe
Type A blood	Type O blood
Type B blood	Type O blood
Rh-positive blood	Rh-negative blood
Normal color vision	Color blindness

Source: McKusick (1998); McKusick-Nathans Institute of Genetic Medicine (2014).

Lynn and Jim's parents are heterozygous for red hair; both have dark hair, but they each carry a recessive gene for red hair. When an individual is heterozygous for a particular trait, the dominant gene is expressed, and the person becomes a carrier of the recessive gene, as shown in Figure 2.4.

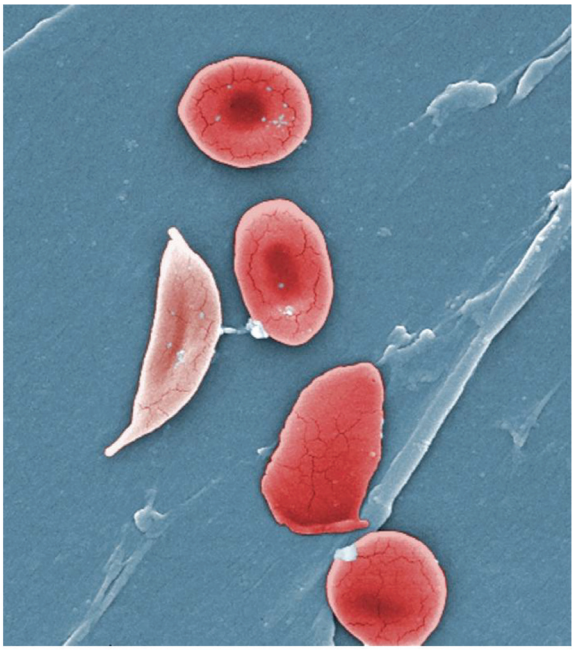
FIGURE 2.4: Dominant-Recessive Inheritance



Incomplete Dominance

In most cases, dominant–recessive inheritance is an oversimplified explanation for patterns of genetic inheritance. **Incomplete dominance** is a genetic inheritance pattern in which both genes influence the characteristic (Plomin et al., 2013). For example, consider blood type. Neither the alleles for blood type A and B dominate each other. A heterozygous person with the alleles for blood type A and B will express both A and B alleles and have blood type AB.

A different type of inheritance pattern is seen when a person inherits heterozygous alleles in which one allele is stronger than the other yet does not completely dominate. In this situation, the stronger allele does not mask all of the effects of the weaker allele. Therefore some, but not all, characteristics of the recessive allele appear. For example, the trait for developing normal blood cells does not completely mask the allele for developing sickle-shaped blood cells. About 8% of African Americans (and relatively few Caucasians or Asian Americans) carry the recessive **sickle cell trait** (Ashley-Koch, Yang, & Olney, 2000; Ojodu, Hulihan, Pope, & Grant, 2014). Sickle cell alleles cause red blood cells to become crescent, or sickle, shaped. Cells that are sickle-shaped cannot distribute oxygen effectively throughout the circulatory system (Ware, de Montalembert, Tshilolo, & Abboud, 2017). However, sickle cell carriers do not develop full-blown sickle cell anemia. Carriers of the trait for sickle cell anemia may function normally but may show some symptoms such as reduced oxygen distribution throughout the body and exhaustion after exercise. Only individuals who are homozygous for the recessive sickle cell trait develop sickle cell anemia.



Wikimedi

PHOTO 2.2: Incomplete Dominance

Recessive sickle cell alleles cause red blood cells to become crescent shaped and unable to distribute oxygen effectively throughout the circulatory system. Alleles for normal blood cells do not mask all of the characteristics of recessive sickle cell alleles, illustrating incomplete dominance.

Polygenic Inheritance

Hereditary influences act in complex ways, and researchers cannot trace most characteristics to only one or two genes. Most traits are a function of the interaction of many genes, known as **polygenic inheritance**. Examples of polygenic traits include height, intelligence, temperament, and susceptibility to certain forms of cancer (Bouchard, 2014; Plomin et al., 2013). As the number of genes that contribute to a trait increases, so does the range of possible traits. Genetic propensities interact with environmental influences to produce a wide range of individual differences in human traits.

Genomic Imprinting

The principles of dominant–recessive and incomplete dominance inheritance can account for more than 1,000 human traits (McKusick, 2007). However, a few traits are determined by a process known as **genomic imprinting**. Genomic imprinting refers to the instance in which the expression of a gene is determined by whether it is inherited from the mother or the father (Kelly & Spencer, 2017; National Library of Medicine, 2013). For example, consider two conditions that illustrate genomic imprinting: Prader-Willi syndrome and Angelman syndrome. Both syndromes are caused by an abnormality in the 15th chromosome (Kalsner & Chamberlain, 2015). If the abnormality occurs on chromosome 15 acquired by the father, the individual—whether a daughter or son—will develop Prader-Willi syndrome, a set of specific physical and behavioral characteristics including obesity, insatiable hunger, short stature, motor slowness, and mild to moderate intellectual impairment. If the abnormal chromosome 15 arises from the mother, the individual—again, whether it is a daughter or a son—will develop Angelman syndrome, characterized by hyperactivity, thin body frame, seizures, disturbances in gait, and severe learning disabilities including severe problems with speech. Prader-Willi and Angelman syndromes each occur in about 1 in 15,000 persons (Everman & Cassidy, 2000). Patterns of genetic inheritance can be complex, yet they follow predictable principles. For a summary of patterns of genetic inheritance, refer to Table 2.2.

TABLE 2.2 • Summary: Patterns of Genetic Inheritance

INHERITANCE PATTERN	DESCRIPTION
Dominant–recessive inheritance	Genes that are dominant are always expressed, regardless of the gene they are paired with, and recessive genes are expressed only if paired with another recessive gene.
Incomplete dominance	Both genes influence the characteristic, and aspects of both genes appear.
Polygenic inheritance	Polygenic traits are the result of interactions among many genes.
Genomic imprinting	The expression of a gene is determined by whether it is inherited from the mother or the father.

Thinking in Context 2.1

1. Why do twins occur? From an evolutionary developmental perspective, does twinning serve an adaptive purpose for our species? Why or why not?
2. Consider your own physical characteristics, such as hair and eye color. Are they indicative of recessive traits or dominant ones?
3. Do you think that you might be a carrier of recessive traits? Why or why not?

CHROMOSOMAL AND GENETIC PROBLEMS

LO 2.2 Define and provide examples of genetic disorders and chromosomal abnormalities.

Many disorders are caused by inherited genes. Some disorders and abnormalities are the result of dominant–recessive inheritance to which one or both parents contribute. Others are the result of variations in chromosomes.

GENETIC DISORDERS

Disorders and abnormalities that are inherited through the parents' genes include such well-known conditions as cystic fibrosis and sickle cell anemia, as well as others that are rare and, in some cases, never even noticed throughout the individual's life.

Dominant–Recessive Disorders

Recall that in dominant–recessive inheritance, dominant genes are always expressed, regardless of the gene they are paired with, and recessive genes are expressed only if paired with another recessive gene. Table 2.3 illustrates diseases that are inherited through dominant–recessive inheritance. Few severe disorders are inherited through dominant–recessive inheritance because individuals who inherit the allele often do not survive long enough to reproduce and pass it to the next generation. One exception is Huntington's disease, a fatal disease in which the central nervous system deteriorates (National Library of Medicine, 2013; Sadler, 2015). Individuals with the Huntington's allele develop normally in childhood, adolescence, and young adulthood. Symptoms of Huntington's disease do not appear until age 35 or later. By then, many individuals have already had children, and one half of them, on average, will inherit the dominant Huntington's gene.

Phenylketonuria (PKU) is a common recessive disorder that prevents the body from producing an enzyme that breaks down the amino acid phenylalanine from proteins (Blau, van Spronsen, & Levy, 2010; Romani et al., 2017). Without treatment, the phenylalanine builds up quickly to toxic levels that damage the central nervous system, contributing to intellectual developmental disability, once known as mental retardation. PKU illustrates how genes interact with the environment to produce developmental outcomes because

TABLE 2.3 • Diseases Inherited Through Dominant–Recessive Inheritance

DISEASE	OCCURRENCE	MODE OF INHERITANCE	DESCRIPTION	TREATMENT
Huntington's disease	1 in 20,000	Dominant	Degenerative brain disorder that affects muscular coordination and cognition	No cure; death usually occurs 10 to 20 years after onset
Cystic fibrosis	1 in 2,000–2,500	Recessive	An abnormally thick, sticky mucus clogs the lungs and digestive system, leading to respiratory infections and digestive difficulty	Bronchial drainage, diet, gene replacement therapy
Phenylketonuria (PKU)	1 in 8,000–10,000	Recessive	Inability to digest phenylalanine that, if untreated, results in neurological damage and death	Diet
Sickle cell anemia	1 in 500 African Americans	Recessive	Sickling of red blood cells leads to inefficient distribution of oxygen throughout the body that leads to organ damage and respiratory infections	No cure; blood transfusions, treat infections, bone marrow transplant; death by middle age
Tay-Sachs disease	1 in 3,600 to 4,000 descendants of Central and Eastern European Jews	Recessive	Degenerative brain disease	None; most die by 4 years of age

Source: McKusick-Nathans Institute of Genetic Medicine (2014).

Chris Walker/MCT/Newscom

PHOTO 2.3: Dominant–Recessive Disorders

This young man is diagnosed with Fragile X syndrome, a recessive disorder carried on the X chromosome and the most common form of inherited intellectual impairment.

intellectual disability results from the interaction of the genetic predisposition and exposure to phenylalanine from the environment (Blau, 2016a). The United States and Canada require all newborns to be screened for PKU (Blau, Shen, & Carducci, 2014). If the disease is discovered, the infant is placed on a diet low in phenylalanine. Children who maintain a strict diet usually attain average or near-average levels of intelligence (Blau, 2016b; Widaman, 2009). Some cognitive and psychological problems may appear in childhood and persist into adulthood, particularly difficulty in attention and planning skills, emotional regulation, depression, and anxiety (Blau et al. 2010; Enns et al., 2010; Huijbregts, Gassió, & Campistol, 2013).

X-Linked Disorders

Some recessive genetic disorders are carried on the X chromosome, like the gene for hemophilia, a condition in which the blood does not clot normally (Barlow-Stewart, 2012). Males are more likely to be affected by X-linked genetic disorders because they have only one X chromosome, and therefore any genetic marks on their X chromosome are displayed. Females (XX) have two X chromosomes; a recessive gene located on one X chromosome will be masked by a dominant gene on the other X chromosome. Females are, therefore, less likely to display X-linked genetic disorders because both of their X-chromosomes must carry the recessive genetic disorder for it to be displayed. In contrast, **fragile X syndrome** is an example of a dominant–recessive disorder carried on the X chromosome (Hagerman, 2011). Because the gene is dominant, it need appear on only one X chromosome to be displayed. That means that fragile X syndrome occurs in both males and females. Table 2.4 illustrates diseases acquired through X-linked inheritance.

TABLE 2.4 • Diseases Acquired Through X-Linked Inheritance

SYNDROME/ DISEASE	OCCURRENCE	DESCRIPTION	TREATMENT
Color blindness	1 in 12 males	Difficulty distinguishing red from green; less common is difficulty distinguishing blue from green	No cure
Duchenne muscular dystrophy	1 in 3,500 males	Weakness and wasting of limb and trunk muscles; progresses slowly but will affect all voluntary muscles	Physical therapy, exercise, body braces; survival rare beyond late 20s
Fragile X syndrome	1 in 2000 males	Symptoms include cognitive impairment; attention problems; anxiety; unstable mood; long face; large ears; flat feet; and hyperextensible joints, especially fingers	No cure
Hemophilia	1 in 3,000–7,000 males	Blood disorder in which the blood does not clot	Blood transfusions

Source: McKusick-Nathans Institute of Genetic Medicine (2016)

CHROMOSOMAL ABNORMALITIES

Chromosomal abnormalities are the result of errors during cell reproduction, meiosis, or mitosis or damage caused afterward. Occurring in 1 of about every 700 births, the most widely known chromosome disorder is trisomy 21, more commonly called **Down syndrome** (Parker et al., 2010). Down syndrome occurs when a third chromosome appears alongside the 21st pair of chromosomes. Although individuals with Down syndrome vary in the severity of their symptoms, Down syndrome is associated with marked physical, health, and cognitive attributes, including a short, stocky build and striking facial features, such as a round face, almond-shaped eyes, and a flattened nose (Davis & Escobar, 2013; Kruszka et al., 2017). Children with Down syndrome tend to show delays in physical and motor development relative to other children and health problems such as congenital heart defects, vision impairments, poor hearing, and immune system deficiencies (Ram & Chinen, 2011; Zampieri et al., 2014). Down syndrome is the most common genetic cause of intellectual developmental disability (Davis & Escobar, 2013), but children's abilities vary. Children who participate in early intervention and receive sensitive caregiving and encouragement to explore their environment show positive outcomes, especially in the motor, social, and emotional areas of functioning (Hazlett, Hammer, Hooper, & Kamphaus, 2011).

Digital Light Source/Universal/Getty

PHOTO 2.4: Chromosomal Abnormalities

Down syndrome is the most common cause of intellectual disability. Children with Down syndrome show more positive developmental outcomes when adults are sensitive to their needs. Interventions that encourage children to interact with their environment can promote motor, social, and emotional development.

Advances in medicine have addressed many of the physical health problems associated with Down syndrome so that today, many individuals with Down syndrome live well into middle age, with an average life expectancy of 60 (Glasson, Dye, & Bittles, 2014; Torr, Strydom, Patti, & Jokinen, 2010). As more adults age with Down syndrome, we have discovered a link between Down syndrome and Alzheimer's disease, a brain degenerative disease that typically strikes in older adulthood (Hithersay, Hamburg, Knight, & Strydom, 2017; Wiseman et al., 2015). This is an example of how disorders and illnesses can be influenced by multiple genes and complex contextual interactions; in this case, Down syndrome and Alzheimer's disease share genetic markers.

Some of the most common chromosomal abnormalities concern the 23rd pair of chromosomes: the sex chromosomes. Given their different genetic makeup, sex chromosome abnormalities yield different effects in males and females. They are summarized in Table 2.5.

MUTATION

Not all inborn characteristics are inherited. Some result from **mutations**, sudden changes and abnormalities in the structure of genes that occur spontaneously or may be induced by exposure to environmental toxins such as radiation and agricultural chemicals in food (Burns & Bottino, 1989; Lewis, 2006). A mutation may involve only one gene or many. It is estimated that as many as one-half of all conceptions include mutated chromosomes (Plomin et al., 2013). Most mutations are fatal—the developing organism dies very soon after conception, often before the woman knows she is pregnant (Lewis, 2006; Rimoin, Connor, & Pyeritz, 1997).

Sometimes mutations are beneficial. This is especially true if the mutation is induced by stressors in the environment and provides an adaptive advantage to the individual. For example, the sickle cell gene is a mutation that originated in areas where malaria is widespread, such as Africa. Children who inherited a single sickle cell allele were more resistant to malarial infection and more likely to survive and pass it along to their offspring

TABLE 2.5 • Sex Chromosome Abnormalities

FEMALE GENOTYPE	SYNDROME	DESCRIPTION	PREVALENCE
XO	Turner	As adults, they are short in stature, often have small jaws with extra folds of skin around their necks (webbing), lack prominent female secondary sex characteristics, such as breasts, and show abnormal development of the ovaries. Elevated risk for thyroid disease, vision and hearing problems, heart defects, diabetes, and autoimmune disorders.	1 in 2,500 females
XXX	Triple-X	Grow about an inch or so taller than average, with unusually long legs and slender torsos, and show normal development of sexual characteristics and fertility. Because many cases of triple-X syndrome often go unnoticed, little is known about the syndrome.	Unknown
MALE GENOTYPE	**SYNDROME**	**DESCRIPTION**	**PREVALENCE**
XXY	Klinefelter	Symptoms range in severity from unnoticeable to severe symptoms such as a high-pitched voice, feminine body shape, breast enlargement, and infertility. Many boys and men with Klinefelter syndrome have short stature, a tendency to be overweight, and language and short-term memory impairments that can cause difficulties in learning.	1 in 500 to 1 in 1,000
XYY	XYY, Jacob's Syndrome	Accompanied by high levels of testosterone.	Prevalence of XYY syndrome is uncertain as most men with XYY syndrome are unaware that they have a chromosomal abnormality

Sources: Bardsley et al. (2013); Bird & Hurren (2016); Herlihy & McLachlan (2015); National Library of Medicine (2013); Otter, Schrander-Stumpel, & Curfs (2009); Pinsker (2012); Powell & Schulte (2011).

(Allison, 2004; Gong, Parikh, Rosenthal, & Greenhouse, 2013). The sickle cell gene is not helpful in places of the world where malaria is not a risk. The frequency of the gene is decreasing in areas of the world where malaria is uncommon. For example, only 8% of African Americans are carriers, compared with as many as 30% of black Africans in some African countries (Maakaron & Taher, 2017). Therefore, the developmental implications of genotypes—and mutations—are context specific, posing benefits in some contexts and risks in others.

PREDICTING AND DETECTING GENETIC DISORDERS

The likelihood of genetic disorders often can be predicted before conception. Moreover, advances in technology permit abnormalities to be detected earlier than ever before.

Genetic Counseling

When considering having children, many couples seek genetic counseling to determine the risk of their children inheriting genetic defects and chromosomal abnormalities (Uhlmann, Schuette, & Yashar, 2009). The genetic counselor constructs a family history of heritable disorders for both prospective parents. If either member of the couple appears to carry a genetic disorder, genetic screening blood tests may be carried out on both parents to detect chromosomal abnormalities and the presence of dominant and recessive genes for various disorders.

Candidates for genetic counseling include those whose relatives have a genetic condition, couples who have had difficulties bearing children, women over the age of 35, and couples from the same ethnic group. Once prospective parents learn about the risk of conceiving a child with a disorder, they can determine how to proceed—whether to conceive a child naturally or through the use of in vitro fertilization—after screening gametes for the disorders of concern. Given advances in our knowledge of genetic disorders and ability to screen for them, some argue that genetic counseling should be available to all prospective parents (Minkoff & Berkowitz, 2014).

Prenatal Diagnosis

Prenatal testing is recommended when genetic counseling has determined a risk for genetic abnormalities, when the woman is older than age 35, when both parents are members of an ethnicity at risk for particular genetic disorders, or when fetal development appears abnormal (Barlow-Stewart & Saleh, 2012). Technology has advanced rapidly, equipping professionals with an array of tools to assess the health of the fetus. Table 2.6 summarizes methods of prenatal diagnosis.

PHOTO 2.5: Genetic Counseling

TABLE 2.6 • Methods of Prenatal Diagnosis

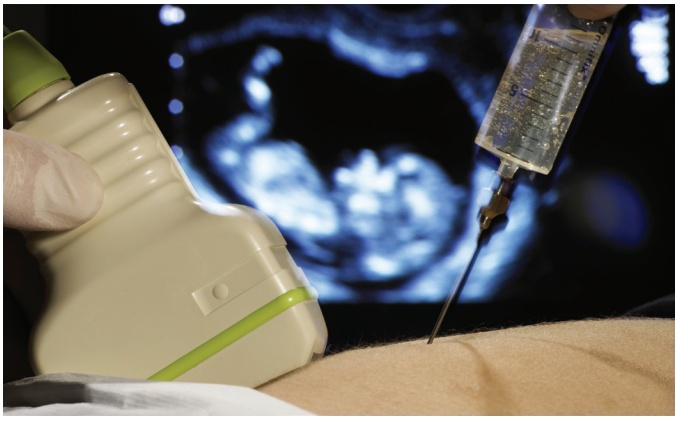
METHOD	EXPLANATION	ADVANTAGES	DISADVANTAGES
Ultrasound	High-frequency sound waves directed at the mother's abdomen provide clear images of the womb projected on to a video monitor.	Ultrasound enables physicians to observe the fetus, measure fetal growth, reveal the sex of the fetus, and to determine physical abnormalities in the fetus.	Many abnormalities and deformities cannot be easily observed.
Amniocentesis	A small sample of the amniotic fluid that surrounds the fetus is extracted from the mother's uterus through a long, hollow needle inserted into the mother's abdomen. The amniotic fluid contains fetal cells. The fetal cells are grown in a laboratory dish in order to create enough cells for genetic analysis.	It permits a thorough analysis of the fetus's genotype. There is 100% diagnostic success rate.	Safe, but poses a greater risk to the fetus than ultrasound. If conducted before the 15th week of pregnancy, it may increase the risk of miscarriage.
Chorionic villus sampling (CVS)	Chorionic villus sampling requires studying a small amount of tissue from the chorion, part of the membrane surrounding the fetus, for the presence of chromosomal abnormalities. The tissue sample is obtained through a long needle inserted either abdominally or vaginally, depending on the location of the fetus.	It permits a thorough analysis of the fetus's genotype. CVS is relatively painless, and there is a 100% diagnostic success rate. Can be conducted earlier than amniocentesis, between 10 and 12 weeks.	It may pose a higher rate of spontaneous abortion and limb defects when conducted prior to 10 weeks' gestation.
Noninvasive prenatal testing (NIPT)	Cell-free fetal DNA is examined by drawing blood from the mother.	There is no risk to the fetus. It can diagnose several chromosomal abnormalities.	It cannot yet detect the full range of abnormalities. It may be less accurate than other methods. Researchers have identified the entire genome sequence using NIPT, suggesting that someday NIPT may be as effective as other, more invasive techniques.

Sources: Akolekar, Beta, Picciarelli, Ogilvie, & D'Antonio (2015); Chan, Kwok, Choy, Leung, & Wang (2013); Fan et al. (2012); Gregg et al. (2013); Odibo (2015); Shahbazian, Barati, Arian, & Saadati (2012); Shim et al. (2014); Tabor & Alfirevic (2010); Theodora et al. (2016).

Prenatal Treatment of Genetic Disorders

What happens when a genetic or chromosomal abnormality is found? Advances in genetics and in medicine have led to therapies that can be administered prenatally to reduce the effects of many genetic abnormalities. For example, hormones and other drugs, as well as blood transfusions, can be given to the fetus by inserting a needle into the uterus (Fox & Saade, 2012; Lindenburg, van Kamp, & Oepkes, 2014). Most strikingly, fetal surgery

can repair defects of the heart, lung, urinary tract, and other areas (Danzer & Johnson, 2014; Sala et al., 2014). Researchers believe that one day we may be able to treat many heritable disorders thorough genetic engineering, by synthesizing normal genes to replace defective ones. It may someday be possible to sample cells from an embryo, detect harmful genes and replace them with healthy ones, then return the healthy cells to the embryo, where they will reproduce and correct the genetic defect (Coutelle & Waddington, 2012). This approach has been used to correct certain heritable disorders in animals and holds promise for treating humans.



Saturn Stills / Science Source

PHOTO 2.6: Prenatal Diagnosis

During amniocentesis, ultrasound is used to guide the insertion of a long, hollow needle into the mother's abdomen in order to extract a sample of the amniotic fluid that surrounds the fetus. The amniotic fluid contains fetal cells, which are grown in a laboratory dish and tested for genetic and chromosomal anomalies and defects.

Thinking in Context 2.2

1. Discuss how PKU illustrates the following two themes in human development: (1) the role of nature and nurture in development and (2) interactions among domains of development.
2. Identify risk factors for genetic and chromosomal disorders. What can prospective parents do to minimize the risks? What specific advice do you give?
3. Suppose you are a 36-year-old woman pregnant with your first child. What would be the advantages and disadvantages of the four types of prenatal diagnostic testing described in Table 2.6? What information would your health care provider need in order to recommend testing appropriate for your particular case?

HEREDITY AND ENVIRONMENT

LO 2.3 Explain how the dynamic interactions of heredity and environment influence development.

We have learned a great deal about genetic inheritance. Most human traits, however, are influenced by a combination of genes working in concert with environmental influences. Our genetic makeup, inherited from our biological parents, consists of a complex blend of hereditary characteristics known as genotype. Our **genotype** is a biological influence on all of our traits, from hair and eye color to personality, health, and behavior. However, our **phenotype**, the traits we ultimately show, such as our specific eye or hair color, is not determined by genotypes alone. Phenotypes are influenced by the interaction of genotypes and our experiences.

BEHAVIORAL GENETICS

Behavioral genetics is the field of study that examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors (Maxson, 2013; Plomin et al., 2013). Genotypes alone do not determine people's traits, characteristics, or personalities; instead, development is the process by which our genetic inheritance (genotype) is expressed in observable characteristics and behaviors (phenotype). Behavioral geneticists recognize that even traits that have a strong genetic component, such as height, are modified by environmental influences (Dubois et al., 2012; Plomin, DeFries, Knopik, & Neiderhiser, 2016). Moreover, most human traits, such as intelligence, are influenced by multiple genes, and there are often multiple variants of each gene (Bouchard, 2014; Chabris, Lee, Cesarini, Benjamin, & Laibson, 2015).

Methods of Behavioral Genetics

Behavioral geneticists devise ways of estimating the heritability of specific traits and behaviors. **Heritability** refers to the extent to which variation among people on a given characteristic is due to genetic differences. The remaining variation not due to genetic differences is instead a result of the environment and experiences. Heritability research therefore examines the contributions of the genotype but also provides information on the role of experience in determining phenotypes (Plomin & Daniels, 2011). Behavioral geneticists assess the hereditary contributions to behavior by conducting selective breeding and family studies (Maxson, 2013).

Using selective breeding studies, behavioral geneticists deliberately modify the genetic makeup of animals to examine the influence of heredity on attributes and behavior. For example, in a classic study, behavioral geneticists demonstrated that they can breed mice to be very physically active or sedentary. They selectively breed highly active mice only with each other and, similarly, breed mice with a very low level of activity with each other. Over subsequent generations, mice bred for high levels of activity become many times more active than those bred for low levels of activity (DeFries, Gervais, & Thomas, 1978). Selective breeding in rats, mice, and other animals such as chickens has revealed genetic contributions to many traits and characteristics, such as aggressiveness, emotionality, sex drive, and even maze learning (Plomin et al., 2016).

Behavioral geneticists conduct *family studies* to compare people who live together and share varying degrees of relatedness. Two kinds of family studies are common: twin studies and adoption studies (Koenen, Amstadter, & Nugent, 2012). Twin studies compare identical and fraternal twins to estimate how much of a trait or behavior is attributable to genes. If genes affect the attribute, identical twins should be more similar than fraternal twins because identical twins share 100% of their genes whereas fraternal twins share about only 50%. Adoption studies, on the other hand, compare the degree of similarity between adopted children and their biological parents whose genes they share (50%) and their adoptive parents with whom they share no genes. If the adopted children share similarities with their biological parents, even though they were not raised by them, it suggests that the similarities are genetic.

Adoption studies also shed light on the extent to which attributes and behaviors are influenced by the environment. For example, the degree to which two genetically unrelated adopted children reared together are similar speaks to the role of environment. Comparisons of identical twins reared in the same home with those reared in different environments can also illustrate environmental contributions to phenotypes. If identical twins reared together are more similar than those reared apart, an environmental influence can be inferred.

Genetic Influences on Personal Characteristics

Research examining the contribution of genotype and environment to intellectual abilities has found a moderate role for heredity. Twin studies have shown that identical twins consistently have more highly correlated scores than do fraternal twins. For example, a study of intelligence in over 10,000 twin pairs showed a correlation of .86 for identical and .60 for fraternal twins (Plomin & Spinath, 2004). Table 2.7 summarizes the results of comparisons of intelligence scores from individuals who share different genetic relationships with each other. Note that correlations for all levels of kin are higher when they are reared together, supporting the role of environment. Average correlations also rise with increases in shared genes.

Genes contribute to many other traits, such as sociability, anxiety, temperament, obesity, happiness, and susceptibility to various illnesses such as heart disease and cancer, poor mental health, and a propensity to be physically aggressive (H. Chen et al., 2013; Pemment, 2013; Veroude et al., 2016; Yoon-Mi, 2009). Yet even traits that are thought to be heavily influenced by genetics can be modified by physical and social interventions.

TABLE 2.7 • Average Correlation of Intelligence Scores From Family Studies for Related and Unrelated Kin Reared Together or Apart

	REARED TOGETHER	REARED APART
MZ twins (100% shared genes)	.86	.72
DZ twins (50% shared genes)	.60	.52
Siblings (50% shared genes)	.47	.24
Biological parent/child (50% shared genes)	.42	.22
Half-siblings (25% shared genes)	.31	—
Unrelated (adopted) siblings (0% shared genes)*	.34	—
Nonbiological parent/child (0% shared genes)*	.19	—

Notes: * Estimated correlation for individuals sharing neither genes nor environment = .0; MZ = monozygotic; DZ = dizygotic.

Source: Adapted from Bouchard & McGue (1981).

For example, growth, body weight, and body height are largely predicted by genetics, yet environmental circumstances and opportunities influence whether genetic potentials are realized (Dubois et al., 2012). Even identical twins who share 100% of their genes are not 100% alike. Those differences are due to the influence of environmental factors, which interact with genes in a variety of ways.

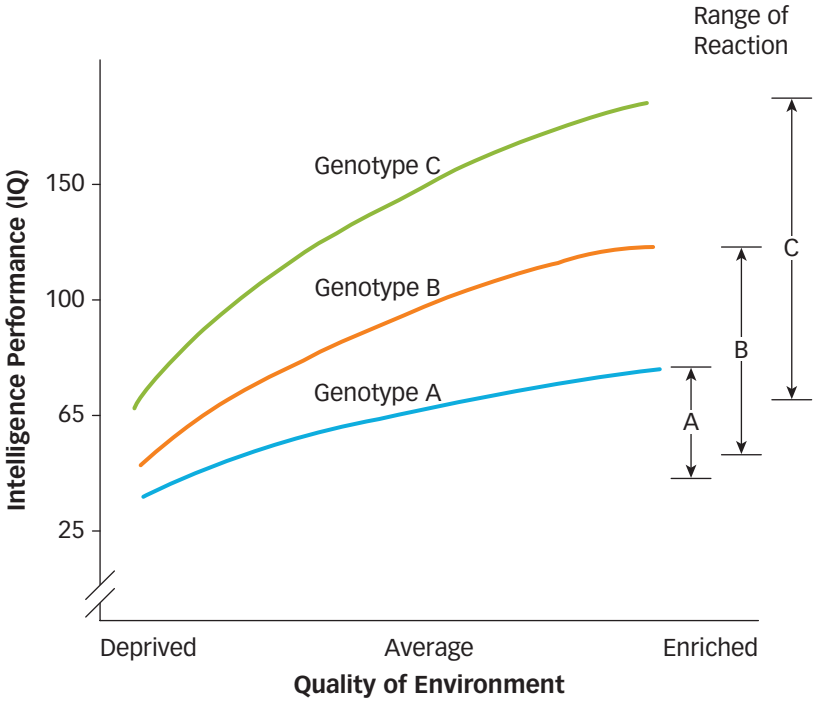
GENE-ENVIRONMENT INTERACTIONS

"You two are so different. Edward and Evan, are you sure you're twins?" kidded Aunt Joan. As fraternal twins, Edward and Evan share 50% of their genes and are reared in the same home. One might expect them to be quite similar, but their similar genes are not the whole story. Genes do not act alone in shaping our development. Instead, genes and the environment work together in complex way to determine our characteristics; behavior; physical, cognitive, and social development; and health (Chabris et al., 2015; Rutter, 2012). **Gene–environment interactions** refer to the dynamic interplay between our genes and our environment. Several principles illustrate these interactions.

Range of Reaction

Everyone has a different genetic makeup and therefore responds to the environment in a unique way. In addition, any one genotype can be expressed in a variety of phenotypes. There is a **range of reaction** (see Figure 2.5), a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints (Gottlieb, 2000). For example, consider height. Height is largely a function of genetics, yet an individual may show a range of sizes depending on environment and behavior. Suppose that a child is born to two very tall parents. She may have the genes to be tall, but unless she has adequate nutrition, she will not fulfill her genetic potential for height. In societies in which nutrition has improved dramatically over a generation, it is common for children to tower over their parents. The enhanced environmental opportunities, in this case nutrition, enabled the children to fulfill their genetic potential for height. Therefore, a genotype sets boundaries on the range of possible phenotypes, but the phenotypes ultimately displayed vary in response to different environments (Manuck & McCaffery, 2014). In this way, genetics sets

FIGURE 2.5: Range of Reaction



Source: Gottlieb (2007).

the range of development outcomes and the environment influences where, within the range, that person will fall.

Canalization

Some traits illustrate a wide reaction range. Others are examples of **canalization**, in which heredity narrows the range of development to only one or a few outcomes. Canalized traits are biologically programmed, and only powerful environmental forces can change their developmental path (Flatt, 2005; Waddington, 1971). For example, infants follow an age-related sequence of motor development, from crawling, to walking, to running. Around the world, most infants walk at about 12 months of age. Generally, only extreme experiences or changes in the environment can prevent this developmental sequence from occurring. For example, children reared in impoverished Romanian and Ethiopian orphanages and exposed to extreme environmental deprivation demonstrated delayed motor development, with some children not walking by 2 years of age (Miller, Tseng, Tirella, Chan, & Feig, 2008; Wilson, 2003). The Lives in Context feature examines gene–environment interactions and responses to child maltreatment.

Motor development is not entirely canalized, however, because some minor changes in the environment can subtly alter its pace and timing. For example, practice facilitates stepping movements in young infants, prevents the disappearance of stepping movements in the early months of life, and leads to an earlier onset of walking (Ulrich, Lloyd, Tiernan, Looper, & Angulo-Barroso, 2008; Zelazo, Zelazo, Cohen, & Zelazo, 1993). These observations demonstrate that even highly canalized traits, such as motor development, which largely unfolds via maturation, can be subtly influenced by contextual factors.

Gene–Environment Correlations

Heredity and environment are each powerful influences on development. Not only do they interact, but heredity and environmental factors are often correlated with each other (Plomin & Asbury, 2001; Scarr & McCartney, 1983). **Gene–environment correlation** refers to the idea that many of our traits are supported by both our genes and environment (Plomin, DeFries, & Loehlin, 1977). Genes give rise to behaviors, which are associated with the environment (Knafo & Jaffee, 2013). There are three types of gene–environment correlations—passive, reactive, and active—as shown in Figure 2.6.

Parents create homes that reflect their own genotypes. Because parents are genetically similar to their children, the homes that they create are not only in line with their own interests and preferences but they also correspond with the child's genotype—an example of a *passive gene–environment correlation* (Wilkinson, Trzaskowski, Haworth, & Eley, 2013). For example, parents might provide genes that predispose a child to develop music ability and also provide a home environment that supports the development of music ability, such as by playing music in the home and owning musical instruments. This type

LIVES IN CONTEXT

Gene-Environment Interactions and Responses to Child Maltreatment

PHOTO 2.7: Lives in Context: Gene-Environment Interactions and Responses to Child Maltreatment

Children who are maltreated or abused by their parents are at risk for developing many problems, including aggression and violent tendencies. Yet not all children who are maltreated become violent adolescents and adults. Why? A classic study examined this question.

Caspi and colleagues (2002) followed a sample of males from birth until adulthood and observed that not all maltreated boys developed problems with violence. Only boys who carried a certain type of gene were at risk for becoming violent after experiencing maltreatment. Specifically, there are two versions of a gene that controls monoamine oxidase A (MAOA), an enzyme that regulates specific chemicals in the brain; one produces high levels of the enzyme and the other produces low levels. Boys who experienced abuse and other traumatic experiences were about twice as likely to develop problems with aggression, violence, and to even be convicted of a violent crime—but only if they carried the low-MAOA gene. Maltreated boys who carried the high-MAOA gene were no more likely to become violent than non-maltreated boys. In addition, the presence of the low MAOA gene itself was not associated with violence. The low-MAOA gene predicted violence only for boys who experience abuse early in life. These findings have been replicated in another 30-year longitudinal study of boys (Fergusson, Boden, Horwood, Miller, & Kennedy, 2011) as well as a meta-analysis of 27 studies (Byrd & Manuck, 2014).

Similar findings of a MAOA gene x environment interaction in which low-MAOA, but not high-MAOA, predicts negative outcomes in response to childhood adversity has been extended to include other mental health outcomes such as antisocial personality disorder and depression (Beach et al., 2010; Cicchetti, Rogosch, & Sturge-Apple, 2007; Manuck & McCaffery, 2014; Nikulina, Widom, & Brzustowicz, 2012). Many of these studies have examined only males. Females show a more mixed pattern with some studies showing that girls display the MAOA gene x environment interaction but to a much lesser extent than boys whereas other studies suggest no relationship (Byrd & Manuck, 2014).

Although there is no single gene that will predict general developmental outcomes, these findings suggest that some genes may increase or decrease our risk for problems in the presence of particular contexts (Belsky & Hartman, 2014a; Conradt, 2017).

In addition, some genes might increase our sensitivity to, and the effectiveness of, environmental interventions (Bakermans-Kranenburg & van IJzendoorn, 2015). Just as we may adjust contextual factors to contribute to successful developmental outcomes and resilience, in the future we might learn how to "turn on" protective genes and "turn off" those that contribute to risk.

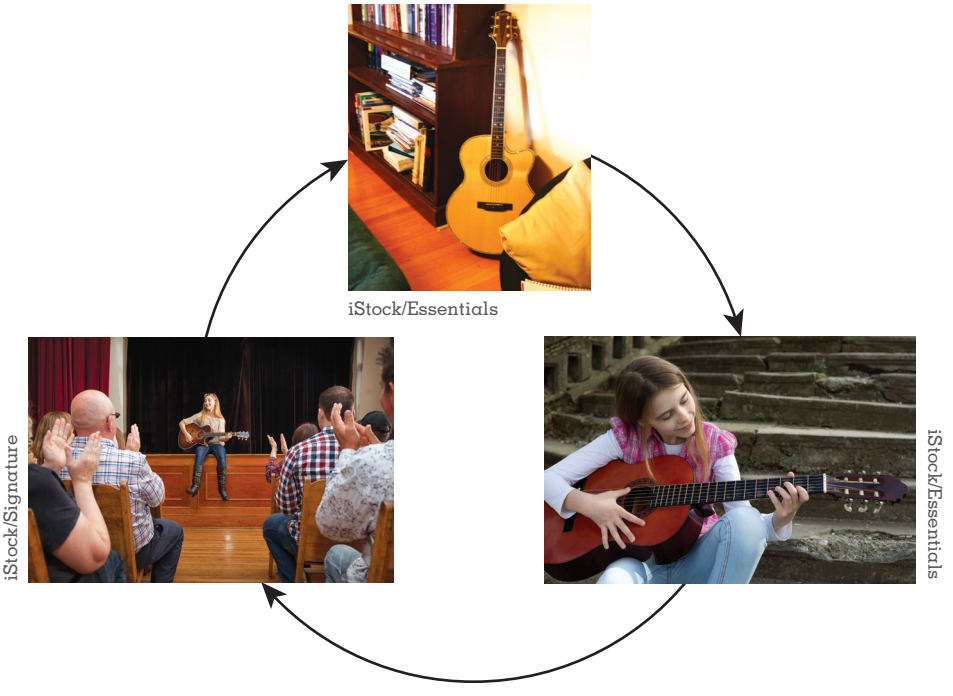
What Do You Think?

1. **In your view, how important are genetic contributors to development?**
2. **If some genes may be protective in particular contexts, should scientists learn how to turn them on? Why or why not? What about genes that may be harmful in particular contexts?**

of gene–environment correlation is seen early in life because children are reared in environments that are created by their parents, who share their genotype.

People naturally evoke responses from others and the environment, just as the environment and the actions of others evoke responses from the individual. In an *evocative gene–environment correlation*, a child's genetic traits (e.g., personality characteristics including openness to experience) influence the social and physical environment, which

FIGURE 2.6: Gene-Environment Correlation



The availability of instruments in the home corresponds to the child's musical abilities and she begins to play guitar (passive gene-environment correlation). As she plays guitar, she evokes positive responses in others, increasing her interest in music (evocative gene-environment correlation). Over time she seeks opportunities to play, such as performing in front of an audience (niche picking).

shape development in ways that support the genetic trait (Burt, 2009; Klahr, Thomas, Hopwood, Klump, & Burt, 2013). For example, active, happy infants tend to receive more adult attention than do passive or moody infants (Deater-Deckard & O'Connor, 2000), and even among infant twins reared in the same family, the more outgoing and happy twin receives more positive attention than does the more subdued twin (Deater-Deckard, 2001). Why? Babies who are cheerful and smile often influence their social world by evoking smiles from others, which in turn support the genetic tendency to be cheerful. In this way, genotypes influence the physical and social environment to respond in ways that support the genotype. Children who engage in disruptive play tend to later experience problems with peers (Boivin et al., 2013). To return to the music example, a child with a genetic trait for music talent will evoke pleasurable responses (e.g., parental approval) when she plays music; this environmental support, in turn, encourages further development of the child's musical trait. In addition, some individuals may be more affected by environmental stimuli due to their genetic makeup (Belsky & Hartman, 2014b).

Children also take a hands-on role in shaping their development. Recall from Chapter 1 that a major theme in understanding human development is the finding that individuals are active in their development; here we have an example of this pattern. As children grow older, they have increasing freedom in choosing their own activities and environments. An *active gene–environment correlation* occurs when the child actively creates experiences and environments that correspond to and influence his genetic predisposition. For example, the child with a genetic trait for interest and ability in music actively seeks experiences and environments that support that trait, such as friends with similar interests and after-school music classes. This tendency to actively seek out experiences and environments compatible and supportive of our genetic tendencies is called **niche-picking** (Scarr & McCartney, 1983).

The strength of passive, evocative, and active gene–environment correlations changes with development, as shown in Figure 2.7 (Scarr, 1992). Passive gene–environment correlations are common at birth as caregivers determine infants' experiences. Correlations between their genotype and environment tend to occur because their environments are made by genetically similar parents. Evocative gene–environment correlations also occur from birth, as infants' inborn traits and tendencies influence others, evoking responses that support their own genetic predispositions. In contrast, active gene–environment correlations take place as children grow older and more independent (Scarr & McCartney, 1983). As they become increasingly capable of controlling parts of their environment, they engage in niche-picking by choosing their own interests and activities, actively shaping their own development. Niche-picking contributes to the differences we see in siblings, including fraternal twins, as they grow older. But identical twins tend to become more similar over time, perhaps because they are increasingly able to select the environments that best fit their genetic propensities (Bouchard et al., 2004; Steves, Spector, & Jackson, 2012). As they age, identical twins—even those reared apart—become alike in attitudes, personality, cognitive ability, intelligence, and preferences; as well, they select similar spouses and best friends (Briley & Tucker-Drob, 2013; Plomin & Deary, 2015; Rushton & Bons, 2005).

EPIGENETIC FRAMEWORK

We have seen that every aspect of our development is the result of dynamic interactions of heredity and environment. Without a doubt, genes provide a biological foundation for our development. However, genes never act alone in determining human characteristics. Moreover, genes themselves may show stable changes not due to DNA (Holliday, 2006a; Lux, 2013). The dynamic interplay between heredity and environment is known as the **epigenetic framework** (Gottlieb, 2003, 2007; Lickliter & Honeycutt, 2013). From this perspective, development results from ongoing reciprocal interactions between genetics and environment.

Genes provide a blueprint for development, determining a range of reaction in which characteristics may develop, depending on environmental circumstances. Not all genes are expressed, however. Genetic expression is influenced by epigenetics (Crews, Gillette, Miller-Crews, & Gore, 2014; Holliday, 2006b; Lester, Conradt, & Marsit, 2016). The term *epigenetics* literally means "above the gene." The epigenome is a molecule that stretches along the length of DNA and provides instructions to genes, determining how they are expressed and whether they are turned on or off. Epigenetic mechanisms determine how genetic instructions are carried out to determine the phenotype. At birth, each cell in our body turns on only a fraction of its genes. Genes continue to be turned on and off over the

FIGURE 2.7: Development Stage and Gene–Environment Correlations

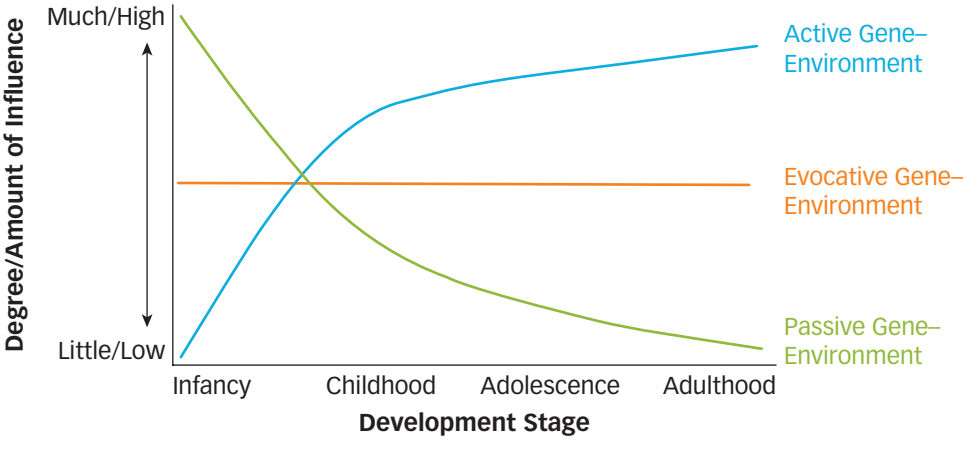
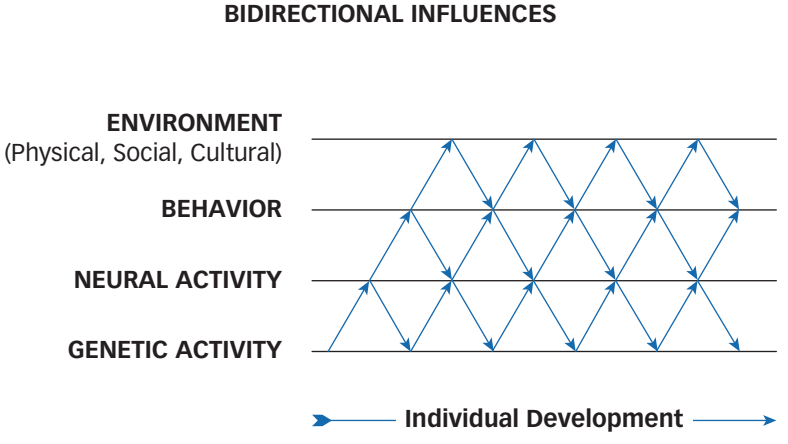


FIGURE 2.8: Epigenetic Framework



SOURCE: Gottlieb (2007).

course of development and also in response to the environment (Gottlieb, 2000). In this way, even traits that are highly canalized can be influenced by the environment. Environmental factors such as toxins, injuries, crowding, diet, and responsive parenting can influence the expression of genetic traits.

For example, consider brain development. Providing an infant with a healthy diet and opportunities to explore the world will support the development of brain cells, governed by genes that are switched on or off. Brain development influences motor development, further supporting the infant's exploration of the physical and social world, thereby promoting cognitive and social development. Active engagement with the world encourages connections among brain cells. Exposure to toxins might suppress the activity of some genes, potentially influencing brain development and its cascading effects on motor, cognitive, and social development. In this way, brain development, like all other aspects of development, is influenced by dynamic interactions between biological and environmental factors.

Evocative gene–environmental correlations and niche-picking illustrate the ways in which genetically expressed characteristics can influence the environment. Genes, and the epigenome, influence development and experience, yet gene expression is also influenced by development and experience, as illustrated in Figure 2.8 (Dodge & Rutter, 2011). These complex gene–environment interactions mean that humans are more than their genes. Interactions between heredity and environment change throughout development as does the role we play in constructing environments that support our genotypes, influence our epigenome, and determine who we become. For a striking example of epigenetics, see the Applied Developmental Science feature.

Thinking in Context 2.3

To answer the following questions, begin by thinking about how your own development reflects interactions among your genes and sociocultural context. Then, describe a skill, ability, or hobby in which you excel.

1. How might a passive gene–environment correlation account for this ability? For example, in what ways has the context in which you were raised shaped this ability?
2. In what ways might this ability be influenced by an evocative-genetic-environment correlation?
3. Provide an example of how this ability might reflect an active gene–environment correlation.
4. Which genetic-environment correlation do you think most accurately accounts for your skill, ability, or hobby?
5. How might you apply the epigenetic framework to account for your ability?

PRENATAL DEVELOPMENT

LO 2.4 Discuss the stages of prenatal development, stages of childbirth, and challenges for infants at risk.

Remarkably, a human infant progresses from fertilization to birth in just 166 days or 38 weeks. Conception, the union of **ovum** and sperm, marks the beginning of prenatal development,

APPLIED DEVELOPMENTAL SCIENCE

Altering the Epigenome

Wikimedia

PHOTO 2.8: Applied Developmental Science: Altering the Epigenome

These two mice are genetically identical. Both carry the agouti gene but in the yellow mouse the agouti gene is turned on all the time. In the brown mouse it is turned off.

One of the earliest examples of epigenetics is the case of agouti mice, which carry the agouti gene. Mice that carry the agouti gene have yellow fur, are extremely obese, shaped much like a pincushion, and prone to diabetes and cancer. When agouti mice breed, most of the offspring are identical to the parents—yellow, obese, and susceptible to life shortening disease. However, a groundbreaking study showed that yellow agouti mice can produce offspring that look very different (Waterland & Jirtle, 2003). The mice in the photo above both carry the agouti gene, yet they look very different; the brown mouse is slender, lean, and has a low risk of developing diabetes and cancer, living well into old age.

Why are these mice so different? Epigenetics. The epigenome carries the instructions that determine what each cell in your body will become—a heart cell, muscle cell, or brain cell, for example. Those instructions are carried out by turning genes on and off.

In the case of the yellow and brown mice, the phenotype of the brown mice has been altered, but the DNA remains the same. Both carry the agouti gene, but in the yellow mouse the agouti gene is turned on all the time. In the brown mouse, it is turned off. In 2003, Waterland and Jirtle discovered that the agouti female's diet can determine her offspring's phenotype. In this study, female mice were fed foods containing chemicals that attach to a gene and turn it off. These chemical clusters are found in many foods such as onions, garlic, beets, soy, and the nutrients in prenatal vitamins. Yellow agouti mothers fed extra nutrients passed along the agouti gene to their offspring, but it was turned off. The mice looked radically different from them (brown) and were healthier (lean, not susceptible to disease) even though they carried the same genes.

Another example supports the finding that the prenatal environment can alter the epigenome and influence the lifelong characteristics of offspring. Pregnant mice were exposed to a chemical (bisphenol-A or BPA, found in certain plastics). When female mice were fed BPA two weeks prior to conception, the number of offspring with the yellow obese coat color signaling an activated agouti gene increased (Dolinoy, 2008). When the pregnant mice were exposed to BPA plus nutritional supplementation (folic acid and an ingredient found in soy products), the offspring tended to be slender and have brown coats, signaling that the agouti gene was turned off. These findings suggest that the prenatal environment can influence the epigenome—and thereby influence how genes are expressed—and that nutrition has the potential to buffer harm.

The most surprising finding emerging from studies of epigenetics, however, is that the epigenome can be influenced by the environment before birth and can be passed by males and females from one generation to the next without changing the DNA itself (Soubry, Hoyo, Jirtle, & Murphy, 2014; Szyf, 2015). This means that what you eat and do today could affect the epigenome—the development, characteristics, and health—of your children, grandchildren, and great grandchildren (Bale, 2015; Vanhees, Vonhögen, van Schooten, & Godschalk, 2014).

What Do You Think?

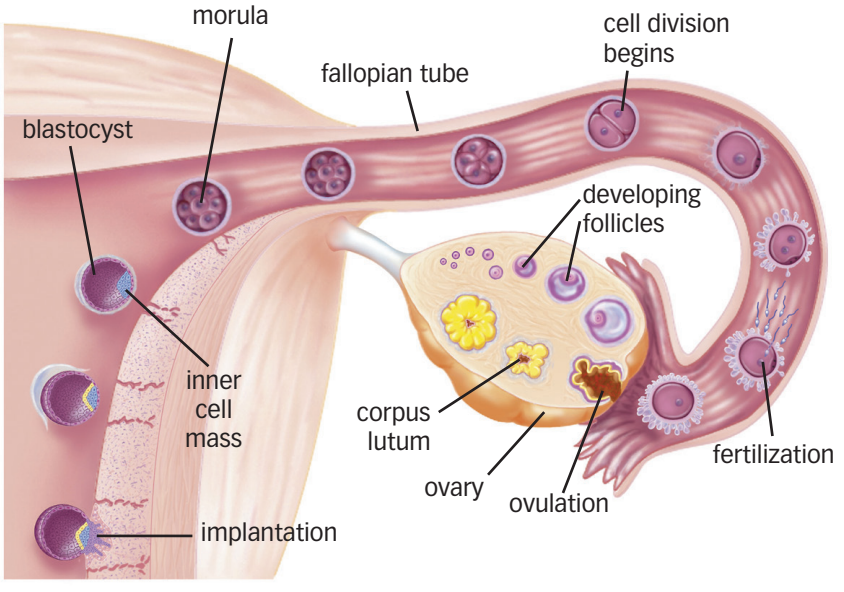
1. **Much of the research on epigenetics examines animals, but there is a growing body of work studying humans. In what ways, if any, might you expect research findings based on people to differ from the findings of animal research, described previously? Explain.**
2. **What might you do to "care for" your epigenome? Identify activities and behaviors that you think might affect the health of your genome.**

the transformative process in which the fertilized ovum, or zygote, progresses through several periods of development, finally emerging from the womb as a neonate. Prenatal development takes place over several stages representing shifts in developmental processes.

GERMINAL PERIOD (FIRST 2 WEEKS AFTER CONCEPTION)

During the **germinal period,** also known as the period of the zygote, the newly created zygote begins cell division as it travels down the fallopian tube, where fertilization took place, toward the uterus. About 30 hours after conception, the zygote then splits down

FIGURE 2.9: Germinal Period



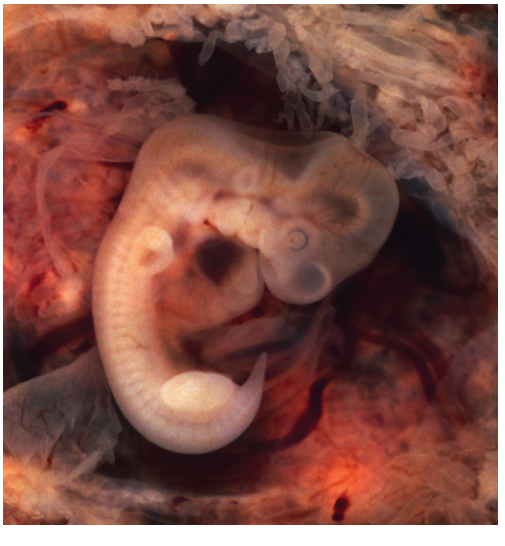
the middle, forming two identical cells (Moore & Persaud, 2016; Sadler, 2015). As shown in Figure 2.9, the two cells each split to form four cells, then eight, and so on. This process of cell division continues at a rapid pace. Any of these cells may become a person (or two, in the case of monozygotic or identical twins).

Cell differentiation begins roughly 72 hours after fertilization when the organism consists of about 16 to 32 cells. Differentiation means that the cells begin to specialize and are no longer identical. At 4 days, the organism consists of about 60 to 70 cells formed into a hollow ball called a **blastocyst**, a fluid-filled sphere with cells forming a protective circle around an inner cluster of cells from which the **embryo** will develop.

Implantation, in which the blastocyst burrows into the wall of the uterus, begins at about day 6 and is complete by about day 11 (Moore & Persaud, 2016; Sadler, 2015). By the end of the second week, when fully implanted into the uterine wall, the outer layer of the blastocyst begins to develop into part of the **placenta,** the principal organ of exchange between the mother and developing organism. The placenta will enable the exchange of nutrients, oxygen, and wastes via the umbilical cord. Also during this stage, the developing organism is encased in amniotic fluid, providing temperature regulation, cushioning, and protection from shocks.

EMBRYONIC PERIOD (3 TO 8 WEEKS AFTER CONCEPTION)

By the third week after conception, the developing organism—now called an embryo—begins a period of structural development during which the most rapid developments of the prenatal period take place. All of the organs and major body systems form during



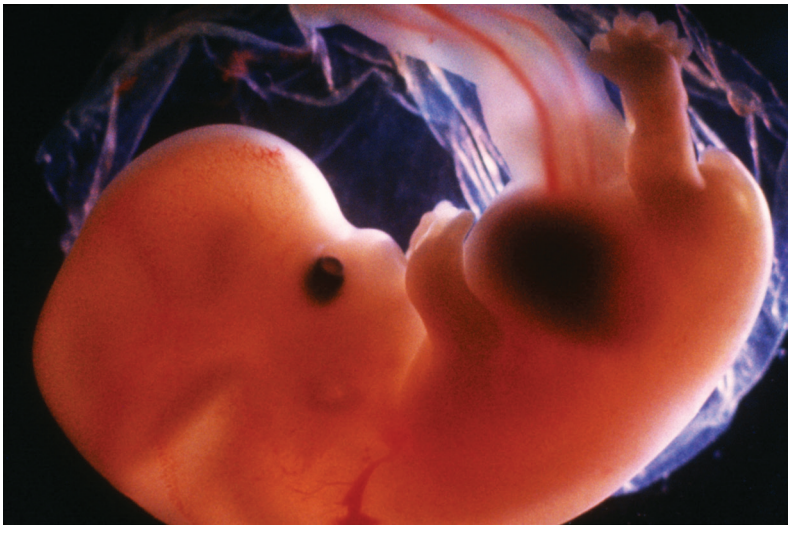


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PHOTO 2.9A & 2.9B: Embryonic Period (Third to Eighth Week After Conception)

Development proceeds very quickly during the embryonic period. Note the dramatic changes from the fifth week (left) to the seventh week (right) of prenatal development.

this **embryonic period**. The mass of cells composing the embryonic disk develops into two layers: the *ectoderm*, the upper layer, will become skin, nails, hair, teeth, sensory organs, and the nervous system; and the *endoderm*, the lower layer, will become the digestive system, liver, lungs, pancreas, salivary glands, and respiratory system. The middle layer, the *mesoderm*, forms later and will become muscles, skeleton, circulatory system and internal organs.

During the third week, at about 22 days after conception, the endoderm folds to form the **neural tube**, which will develop into the central nervous system (brain and spinal cord; Moore & Persaud, 2016; Stiles & Jernigan, 2010). Now the head can be distinguished. A blood vessel that will become the heart begins to pulse and blood begins to circulate throughout the body (Dye, 2000; Larsen, 2001). During days 26 and 27 arm buds appear, followed by leg buds on days 28 through 30 (Moore & Persaud, 2016; Sadler, 2015). The brain develops rapidly and the head grows faster than the other parts of the body during the fifth week of development. The eyes, ears, nose, and mouth begin to form during the sixth week. Upper arms, forearms, palms, legs, and feet appear. The embryo shows reflex responses to touch.

During the seventh week, webbed fingers and toes are apparent; they separate completely by the end of the eighth week. A ridge called the **indifferent gonad** appears; it will develop into the male or female genitals, depending on the fetus's sex chromosomes (Moore & Persaud, 2016). The Y chromosome of the male embryo instructs it to secrete testosterone, causing the indifferent gonad to create testes. In female embryos, no testosterone is released, and the indifferent gonad produces ovaries. The sex organs take several weeks to develop. The external genital organs are not apparent until about 12 weeks.

At the end of the embryonic period, 8 weeks after conception, the embryo weighs about one-seventh of an ounce and is one inch long. All of the basic organs and body parts have formed in a very rudimentary way. The embryo displays spontaneous reflexive movements, but it is still too small for the movements to be felt by the mother (Hepper, 2015). Serious defects that emerge during the embryonic period often cause a miscarriage, or *spontaneous abortion* (loss of the fetus); indeed, most miscarriages are the result of chromosomal abnormalities (Bainbridge, 2003; Suzumori & Sugiura-Ogasawara, 2010). The most severely defective organisms do not survive beyond the first trimester, or third month of pregnancy. It is estimated that up to 45% of all conceptions abort spontaneously, and most occur before the pregnancy is detected (Larsen, 2001; Moore & Persaud, 2016).

Lives in Context Video 2.1
4D Ultrasound

FETAL PERIOD (9 WEEKS TO BIRTH)

The fetal period is marked by the appearance of bone—at about the end of the eighth week. From 9 weeks until birth, the fetus grows rapidly, and its organs become more complex and begin to function. The end of the third month marks the close of the first trimester, at which time all parts of the fetus's body can move spontaneously, the legs kick, and the fetus can suck its thumb (an involuntary reflex). By the end of the 12th week, the upper limbs have almost reached their final relative lengths, but the lower limbs are slightly shorter than their final relative lengths (Sadler, 2015).

Second Trimester (14 to 26 Weeks)

By the 14th week, at the start of the second trimester, limb movements are coordinated, but they will be too slight to be felt by the mother until about 17 to 20 weeks. The heartbeat gets stronger. Eyelids, eyebrows, fingernails, toenails, and tooth buds form. The first hair to appear is **lanugo**, a fine down-like hair that covers the fetus's body; it is gradually replaced by human hair (Dye, 2000). The skin is covered with a greasy material called the **vernix caseosa**, which protects the fetal skin from abrasions, chapping, and hardening that can

occur with exposure to amniotic fluid (Moore & Persaud, 2016). At 21 weeks, rapid eye movements begin, signifying an important time of growth and development for the fetal brain. The brain begins to become more responsive. For example, startle responses have been reported at 22 to 23 weeks in response to sudden vibrations and noises (Hepper, 2015; Sadler, 2015). During weeks 21 to 25, the fetus gains substantial weight, and its body proportions become more like those of a newborn infant. Growth of the fetal body begins to catch up to the head, yet the head remains disproportionately larger than the body at birth.

Third Trimester (27 to 40 Weeks)

During the last 3 months of pregnancy, the fetal body grows substantially in weight and length; specifically, it typically gains over 5 pounds and grows 7 inches. At about 28 weeks after conception brain development grows in leaps and bounds. The cerebral cortex develops convolutions and furrows, taking on the brain's characteristic wrinkly appearance (Dye, 2000). The fetal brain wave pattern shifts to include occasional bursts of activity, similar to the sleep-wake cycles of newborns. By 30 weeks, the pupils of the eyes dilate in response to light. At 35 weeks, the fetus has a firm hand grasp and spontaneously orients itself toward light.

During the third trimester, pregnant women and their caregivers are mindful that the baby may be born prematurely. Although the expected date of delivery is 166 days or 38 weeks from conception (40 weeks from the mother's last menstrual period), about one in every eight American births is premature (Centers for Disease Control, 2014). The age of viability—the age at which advanced medical care permits a preterm newborn to survive outside the womb—begins at about 22 weeks after conception (Sadler, 2015). Infants born before 22 weeks rarely survive more than a few days because their brain and lungs have not begun to function. Although a 22- to 25-week fetus born prematurely may survive in intensive care, it is still at risk because its immature respiratory system may lead to death in early infancy. At about 26 weeks, the lungs become capable of breathing air and the premature infant stands a better chance of surviving if given intensive care. About 80% of infants born at 26 weeks survive and 87% of those born at 27 weeks (Stoll, Hansen, Bell, & Shankaran, 2010; Tucker & McGuire, 2004). Ninety-eight percent of 32-week premature infants survive.

At about the 166th day after conception, the placenta releases a hormone that triggers the onset of labor (Bainbridge, 2003). Hormones cause the mother's uterus to contract and relax at regular intervals, aiding delivery.

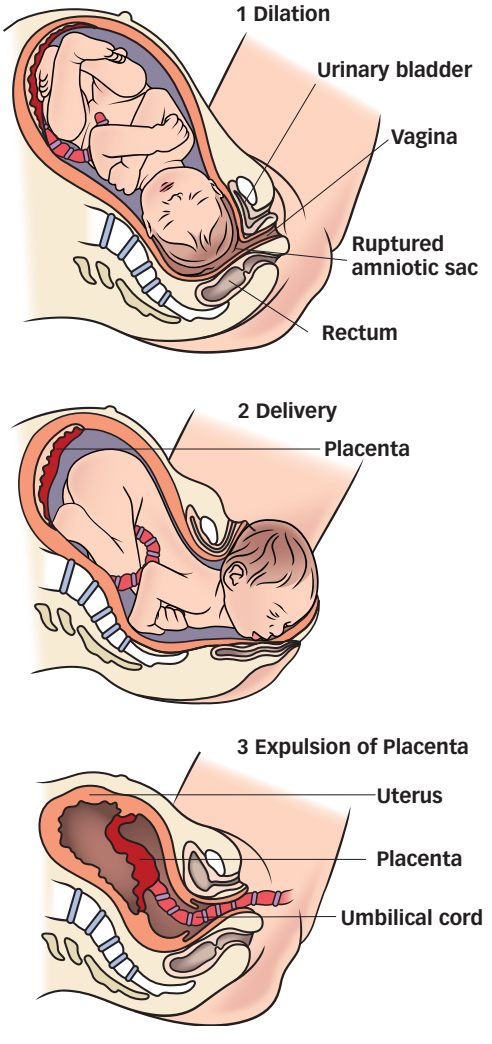
Lives in Context Video 2.2
The Process of Childbirth

CHILDBIRTH

Childbirth, also known as labor, progresses in three stages, as shown in Figure 2.10.

Sometimes a vaginal birth is not possible because of concerns for the health or safety reasons of the mother or fetus. A **cesarean section**, or C-section, is a surgical procedure that removes the fetus from the uterus through the abdomen. About 33% of all singleton births are cesarean deliveries (Hamilton, Martin, Osterman, Curtin, & Mathews, 2015). Cesarean sections are performed when labor progresses too slowly, the fetus is in breech position (feet first) or transverse position (crosswise in the uterus), the head is too large to pass through the pelvis, or the fetus or mother is in danger (Jha, Baliga, Kumar, Rangnekar, & Baliga, 2015; Visscher & Narendran, 2014). Babies delivered by cesarean are exposed to more maternal medication and secrete lower levels of the stress hormones that occur with vaginal birth that are needed to facilitate respiration, enhance circulation of blood to the brain, and help the infant adapt to the world outside of the womb. Interactions between mothers and infants, however, are similar for infants

FIGURE 2.10: Stages of Labor



STAGE	DETAILS	DURATION
Stage 1: Dilation	Labor begins when the mother experiences regular uterine contractions spaced at 10- to 15-minute intervals. The amniotic sac ("water") may rupture at any time during this stage. The contractions, which gradually become stronger and closer together, cause the cervix to dilate so that the fetus's head can pass through.	8 to 14 hours for a woman having her first child; for later-born children, the average is 3 to 8 hours.
Stage 2: Delivery	Begins when the cervix is fully dilated to 10 centimeters and the fetus's head is positioned at the opening of the cervix—known as "crowning." It ends when the baby emerges completely from the mother's body.	30 minutes to an hour and a half.
Stage 3: Delivery of the placenta	The placenta separates from the uterine wall and is expelled by uterine contractions.	Typically happens about 5 to 15 minutes after the baby has emerged, and the process can take up to a half hour.

delivered vaginally and by cesarean section (Durik, Hyde, & Clark, 2000). The Cultural Influences on Development feature examines some cultural differences in childbirth.

The average newborn is about 20 inches long and weighs about 7½ pounds. Boys tend to be slightly longer and heavier than girls. Newborns have distinctive features, including a large head (about ¼ of body length) that is often long and misshapen from passing through the birth canal. The newborn's skull bones are not yet fused—and will not be until about 18 months of age—permitting the bones to move and the head to mold to the birth canal, easing its passage. A healthy newborn is red-skinned and wrinkly at birth; skin that is bluish in color indicates that the newborn has experienced oxygen deprivation. Some babies emerge covered with lanugo, the fuzzy hair that protects the skin in the womb; other babies lose the lanugo prior to birth. The newborn's body is covered with vernix caseosa, a waxy substance that protects against infection; this dries up within the first few days. Although many hospital staff wash the vernix caseosa away, research suggests that it is a naturally occurring barrier to infection and should be retained at birth (Jha et al., 2015).

CULTURAL INFLUENCES ON DEVELOPMENT

Cultural Differences in Childbirth

PHOTO 2.10: Cultural Influences on Development: Cultural Differences in Childbirth

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Societies vary in their customs and perceptions of childbirth, including the privacy afforded to giving birth and how newborns are integrated into the community. In the United States, birth is a private event that usually occurs in a hospital, attended by medical personnel and one or two family members. In most cases, the first-time mother has never witnessed a birth but is well educated and may have well-informed expectations. After birth, the mother and infant are often visited by family during designated hospital visiting hours; the newborn usually rooms with the mother all or part of the day.

In a small village in southern Italy, birth is a community event. It usually takes place in a hospital, attended by a midwife (Fogel, 2007; Schreiber, 1977). Just after birth, the midwife brings the mother's entire family (immediate and extended) to the mother's room and they take turns congratulating the mother and baby, kissing them. The family provides a party including pastry and liqueurs. During labor and afterward the mother is supported and visited by many of her friends and relatives, to recognize the contribution that the mother has made to the community. The mother-in-law is an example of the social support system in place because from a few days before until about 1 month after the birth, she brings and feeds the mother ritual foods of broth, marsala, and fresh cheeses (Fogel, 2007; Schreiber, 1977).

In other cultures, birth is an even more public process. The Jahara of South America give birth under a shelter in full view of everyone in the village (Fogel, 2007). On the Indonesian island of Bali it is assumed that the husband, children, and other family will want to be present. The birth occurs in the home with the aid of a midwife and female relatives. As a result, Balinese women know what to expect in giving birth to their first child because they have been present at many births (Diener, 2000). The baby is immediately integrated into the family and community as he or she is considered a reincarnated soul of an ancestor. Many kin are present to support the mother and baby because the child is considered to be related to many more people than its parents.

Childbirth is tied to social status in the Brong-Ahafo region in Ghana: After a delivery, women achieve a higher social position and can then give advice to other women (Jansen, 2006). Home deliveries are highly valued. The more difficult the delivery and the less skilled assistance she receives, the more respect a woman attains, the higher her position will be, and the more influence she has on the childbirth decisions of other women, such as whether to give birth at home or in a medical setting and how to combine traditional and modern practices (Bazzano, Kirkwood, Tawiah-Agyemang, Owusu-Agyei, & Adongo, 2008).

Many cultures conduct rites that they believe protect newborns from evil spirits. Among the Maya of the Yucatan region of Mexico, there are few changes in the expectant mother's surroundings; the Mayan woman lies in the same hammock in which she sleeps each night. The father-to-be is expected to be present during labor and birth to take an active role but also to witness the suffering that accompanies labor. If the father is not present and the child is stillborn, it is blamed on the father's absence. The pregnant woman's mother is present, often in the company of other females including sisters, sisters-in-law, mothers-in-law, godmothers, and sometimes neighbors and close friends. The mother and child must remain inside the house for one week before returning to normal activity after birth because it is believed that the mother and newborn are susceptible to the influence of evil spirits from the bush (Gardiner & Kosmitzki, 2018).

A neighboring ethnic group, the Zinacanteco, place their newborns naked before a fire. The midwife who assisted the mother says prayers asking the gods to look kindly upon the infant. The infant is dressed in a long skirt made of heavy fabric extending beyond the feet; this garment is to be worn throughout the first year. The newborn is then wrapped in several layers of blankets, even covering the face, to protect against losing parts of the soul. These traditional practices are believed to protect the infant from illnesses as well as evil spirits (Brazelton, 1977; Fogel, 2007).

What Do You Think?

1. **Which of these birthing customs most appeals to you? Why?**
2. **If you, a family member, or friend have given birth, describe the process. Where did the birth occur? Who witnessed it? What happened afterward? When did family and friends meet the baby?**

After birth, newborns are routinely screened with the **Apgar scale**, which provides a quick and easy overall assessment of the baby's immediate health. As shown in Table 2.8, the Apgar scale is composed of five subtests: appearance (color), pulse (heart rate), grimace (reflex irritability), activity (muscle tone), and respiration (breathing). The newborn is rated 0, 1, or 2 on each subscale for a maximum total score of 10. A score of 4 or lower means that the newborn is in serious condition and requires immediate medical attention. The rating is conducted twice, 1 minute after delivery and again 5 minutes after birth; this timing ensures that hospital staff will monitor the newborn over several minutes. More than 98% of all newborns in the United States achieve a 5-minute score of 7 to 10, indicating good health (Martin, Hamilton, Osterman, Curtin, & Mathews, 2013).

TABLE 2.8 • Apgar Scale

	RATING (ABSENCE-PRESENCE)		
INDICATOR	**0**	**1**	**2**
Appearance (Color)	Blue	Pink body, blue extremities	Pink
Pulse (Heart rate)	Absent	Slow (below 100)	Rapid (over 100)
Grimace (Reflex irritability)	No response	Grimace	Coughing, crying
Activity (Muscle tone)	Limp	Weak and inactive	Active and strong
Respiration (Breathing)	Absent	Irregular and slow	Crying, good

Source: Apgar (1953).

INFANTS AT RISK: LOW BIRTH WEIGHT AND SMALL-FOR-DATE BABIES

One of the leading causes of infant mortality is low birth weight, accounting for 35% of mortality cases in infancy (Mathews & MacDorman, 2013). There are two types of low birth weight infants: those who are **preterm,** or premature (born before their due date) and those who are **small for date**, who are full term but have experienced slow growth and are smaller than expected for their gestational age. Infants are classified as **low birth weight** when they weigh less than 2,500 grams (5 ½ pounds) at birth; "very low" birth weight refers to a weight less than 1,500 grams (3 ½ pounds), and "extremely low" birth weight refers to a weight less than 750 grams (1 lb. 10 oz.; Alexander & Slay, 2002). Infants who are extremely low birth weight are most at risk for developmental challenges, handicaps, and difficulty surviving (under 1,000 grams; Fogel, 2007).

Low birth weight infants are at a disadvantage when it comes to adapting to the world outside the womb. At birth, they often experience difficulty breathing and are likely to suffer from respiratory distress syndrome, in which the newborn breathes irregularly and, at times, may stop breathing. Their survival depends on care in neonatal hospital units, where they are confined in isolettes that separate them from the world, regulating their body temperature, aiding their breathing with the use of respirators, and protecting them from infection. Many low birth weight infants cannot yet suck from a bottle, so they are fed intravenously.

The deficits that low birth weight infants endure range from mild to severe and correspond closely to the infant's birth weight, with extremely low birth weight infants suffering the greatest deficits (Hutchinson et al., 2013). Low birth weight infants are at higher risk for poor growth, cerebral palsy, seizure disorders, neurological difficulties, respiratory problems, and illness (Adams-Chapman et al., 2013; Agustines et al., 2000; Aylward, 2005; McGowan, Alderdice, Holmes, & Johnston, 2011; J. E. Miller et al., 2016). Higher rates of sensory, motor, and cognitive problems mean that low birth weight children are more likely to require special education and display poor academic achievement in childhood, adolescence, and even adulthood (Aarnoudse-Moens, Weisglas-Kuperus, van Goudoever, & Oosterlaan, 2009; Eichenwald & Stark, 2009; Hutchinson et al., 2013; MacKay, Smith, Dobbie, & Pell, 2010). Low birth weight children often experience

PHOTO 2.11: Infants at Risk: Low-Birthweight and Small-for-Date Babies

Low birthweight infants require extensive care. They are at risk for poor developmental outcomes and even death.

PORT-AU-PRINCE, HAITI - FEBRUARY 20: A premature baby, named Fred Jr., is carried to a scale to be weighed at the Project MediShare field hospital on the grounds of the Toussaint Louverture International Airport February 20, 2010 in Port-au-Prince, Haiti. More than a month after a 7.0 earthquake devistated Haiti, Project MediShare is coordinating hundreds of doctors, nurses and other medical professionals to aid the vicitims. (Photo by Chip Somodevilla/Getty Images

difficulty in self-regulation, poor social competence, and poor peer relationships, including peer rejection and victimization in adolescence (Georgsdottir, Haraldsson, & Dagbjartsson, 2013; Ritchie, Bora, & Woodward, 2015; Yau et al., 2013). As adults, low birth weight individuals tend to be less socially engaged, show poor communication skills, and may score high on measures of anxiety (Eryigit Madzwamuse, Baumann, Jaekel, Bartmann, & Wolke, 2015).

Parenting a low birth weight infant is stressful even in the best of circumstances (Howe, Sheu, Wang, & Hsu, 2014). Such infants tend to be easily overwhelmed by stimulation and difficult to soothe; they smile less and fuss more than their normal-weight counterparts, making caregivers feel unrewarded for their efforts. Often these infants are slow to initiate social interactions and do not attend to caregivers, looking away or otherwise resisting attempts to attract their attention (Eckerman, Hsu, Molitor, Leung, & Goldstein, 1999). Because low birth weight infants often do not respond to attempts to solicit interaction, they can be frustrating to interact with, can be difficult to soothe, and are at risk for less secure attachment to their parents (Jean & Stack, 2012; Mangelsdorf et al., 1996; Wolke, Eryigit Madzwamuse, & Gutbrod, 2014). Research also indicates that they may experience higher rates of child abuse (Bugental & Happaney, 2004; Klein et al., 1971).

Parental responses to having a low birth weight infant influence the child's long-term health outcomes, independently of perinatal risk, suggesting that the parenting context is an important influence on infant health (Pierrehumbert, Nicole, Muller-Nix, Forcada-Guex, & Ansermet, 2003). When mothers have knowledge about child development and how to foster healthy development, are involved with their children, and create a stimulating home environment, low birth weight infants tend to have good long-term outcomes (Benasich & Brooks-Gunn, 1996; Jones, Rowe, & Becker, 2009). For example, one study of low birth weight children showed that those who experienced sensitive parenting showed faster improvements in executive function and were indistinguishable from their normal-weight peers by age 5; however, those who experienced below-average levels of sensitive parenting showed lasting deficits (Camerota, Willoughby, Cox, Greenberg, & Investigators, 2015). Likewise, exposure to sensitive, positive, parenting predicted low birth weight children's catching up to their normal birth weight peers at age 8 in academic achievement, but exposure to insensitive parenting predicted much poorer functioning (Jaekel, Pluess, Belsky, & Wolke, 2015). Longitudinal research has found that low birth weight children raised in unstable, economically disadvantaged families tend to remain smaller in stature, experience more emotional problems, and show more long-term deficits in intelligence and academic performance than do those raised in more advantaged homes (Taylor, Klein, Minich, & Hack, 2001).

Interventions to promote the development of low birth weight children often emphasize helping parents learn coping strategies for interacting with their infants and managing stress (Chang et al., 2015; Lau & Morse, 2003). Interventions focused on teaching parents how to massage and touch their infants in therapeutic ways as well as increase skin-to-skin contact with their infants are associated with better cognitive and neurodevelopmental outcomes at age 2 (Procianoy, Mendes, & Silveira, 2010). One intervention common in developing countries where mothers may not have access to hospitals is **kangaroo care**, in which the infant is placed vertically against the parent's chest, under the shirt, providing skin-to-skin contact (Charpak et al., 2005). As the parent goes about

daily activities, the infant remains warm and close, hears the voice and heartbeat, smells the body, and feels constant skin-to-skin contact. Kangaroo care is so effective that the majority of hospitals in the U.S. offer kangaroo care to preterm infants. Babies who receive early and consistent kangaroo care grow more quickly, sleep better, score higher on measures of health, and show more cognitive gains throughout the first year of life (Boundy et al., 2015; Jefferies, 2012).

Thinking in Context 2.4

1. Petra noticed that her abdomen has not grown much since she became pregnant 3 months ago. She concluded that the fetus must not undergo significant development early in pregnancy. How would you respond to Petra?
2. Parents' decisions about childbirth reflect their knowledge about birth options as well as cultural values. Referring to Bronfenbrenner's bioecological model (see Chapter 1), identify factors at each bioecological level that may influence childbirth. For example, how might neighborhood factors influence birth options? Culture?
3. Thinking of how society and medical science have changed in recent decades, in what ways might recent cohorts of parents differ from prior cohorts? What implications might these differences hold for prenatal development and childbirth?

ENVIRONMENTAL INFLUENCES ON PRENATAL DEVELOPMENT

LO 2.5 Identify the principles of teratology, types of teratogens, and ways that teratogens can be used to predict prenatal outcomes.

The vast majority of infants are born healthy, but some are exposed before birth to environmental obstacles that hinder their development. A **teratogen** is an agent that causes damage to prenatal development, such as a disease, drug, or other environmental factor, producing a birth defect. The field of *teratology* attempts to find the causes of birth defects so that they may be avoided. Health care providers help pregnant women and those who intend to become pregnant to be aware of teratogens and avoid them, as much as possible, to maximize the likelihood of having a healthy baby.

PRINCIPLES OF TERATOLOGY

There are many ways in which teratogens may affect prenatal development, but it is not always easy to predict the harm caused by teratogens. Generally, the effects of exposure to teratogens on prenatal development vary depending on the following principles (Collins, 2006; Moore & Persaud, 2016; Sadler, 2015).

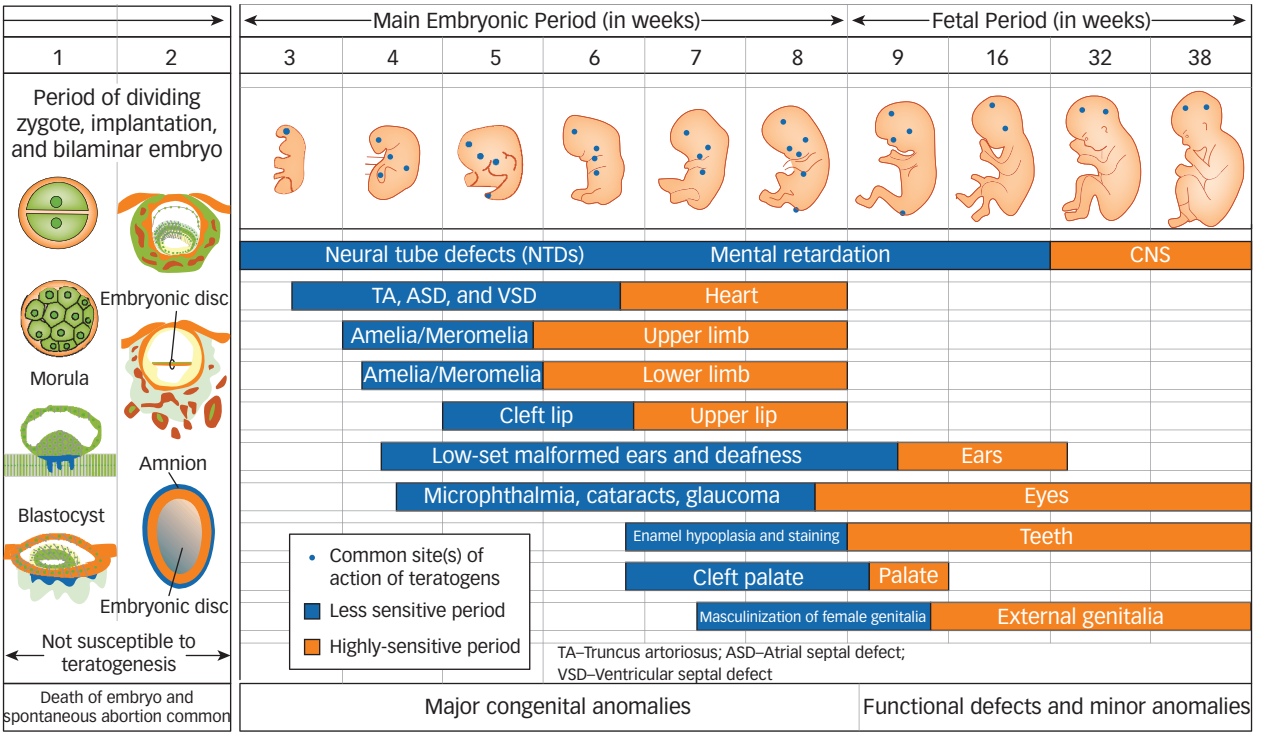
- *Critical Periods.* There are critical periods during prenatal development in which an embryo is more susceptible to damage from exposure to teratogens. The extent to which exposure to a teratogen disrupts prenatal development depends on the stage of prenatal development when exposure occurs. Generally, sensitivity to teratogens begins at about 3 weeks after conception (Sadler, 2015). Structural defects occur when the embryo is exposed to teratogen while that part of the body is developing. As shown in Figure 2.11, each organ of the body has a sensitive period in development during which it is most susceptible to damage from teratogens. Once a body part is fully formed, it is less likely to be harmed by exposure to teratogens; however, some body parts, like the brain, remain vulnerable throughout pregnancy.

- *Dose.* The amount of exposure (i.e., dosage) to a teratogen influences its effects. Generally, the greater the dose, the more damage to development; however, teratogens also differ in their strength. Some teratogens, like alcohol, display a powerful dose–response relationship so that larger doses—heavier and more frequent drinking—result in greater damage.
- *Individual differences.* Individuals vary in their susceptibility to particular teratogens based on the genetic makeup of both the organism and mother, as well as the quality of the prenatal environment.
- *Teratogens show complicated effects on development.* Different teratogens can cause the same birth defect, and a variety of birth defects can result from the same teratogen. Also, some teratogens have subtle effects that result in developmental delays that are not obvious at birth. For example, infants exposed prenatally to as little as an ounce of alcohol a day usually display no obvious physical deformities, but later, as children, they may demonstrate cognitive delays (Jacobson & Jacobson, 1996). Other teratogens display sleeper effects—effects that are not visible until many years later. For example, infants born to women who consumed diethylstilbestrol (DES), a hormone that was widely prescribed between 1945 and 1970 to prevent miscarriages, were born healthy, but as adults they were more likely to experience problems with their reproductive systems. Daughters born to mothers who took DES were more likely to develop a rare form of cervical cancer, have miscarriages, and give birth to infants who were premature or low birth weight (Barnes et al., 1980; Schrager & Potter, 2004).

TYPES OF TERATOGENS

Prenatal development can be influenced by many contextual factors, including maternal consumption of over-the-counter (OTC), prescription, and recreational drugs;

FIGURE 2.11: Sensitive Periods in Prenatal Development



Source: Levine and Munsch (2010, p. 113).

illness; environmental factors; and more, as shown in Table 2.9. Although the developing organism is vulnerable to many teratogens, the mother's body is designed to protect the growing fetus.

Some teratogens can be avoided by choice; for example, a woman can choose not to drink alcohol or smoke cigarettes during pregnancy. Others, however, may be involuntary, as in the case of maternal illness. Sometimes a pregnant woman and her doctor may have to make a difficult choice between forgoing a needed prescription drug and putting the fetus at risk. And, in any case, a woman may not know she is pregnant until after the first few weeks of the embryonic stage are already past. Thus, in the real world, almost no pregnancy can be entirely free of exposure to teratogens. However, each year about 97% of infants are born without defects (Centers for Disease Control, 2014).

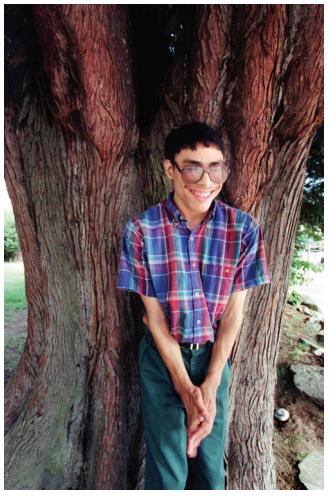
Prescription and Nonprescription Drugs

More than 90% of pregnant women take prescription or over-the-counter (OTC) medications (Servey & Chang, 2014). Prescription drugs that can act as teratogens include antibiotics, certain hormones, anticoagulants, anticonvulsants, and some acne drugs (Collins, 2006; Moore & Persaud, 2016; Sadler, 2015). In several cases physicians have unwittingly prescribed drugs to ease pregnant women's discomfort that caused harm to the fetus. For example, in the late 1950s and early 1960s many pregnant women were

TABLE 2.9 • Hazards to Prenatal Development

DRUGS	
Alcohol	Fetal alcohol syndrome, mental retardation; retarded fetal growth; joint abnormalities; ocular abnormalities
Amphetamines	Premature delivery; stillbirth; irritability and poor feeding among newborns
Antibiotics (Tetracycline, Streptomycin, Terramycin)	Premature delivery; restricted skeletal growth; cataracts
Barbiturates	Lethargy in the fetus; large doses cause anoxia (oxygen starvation), restricts fetal growth
Cocaine	Retarded fetal growth; prematurity, microcephaly; neurobehavioral disturbances; genital abnormalities
Heroin	Retarded fetal growth; premature labor; newborns suffer withdrawal
Lithium	Heart and blood vessel abnormalities
Marijuana	Smoked retards fetal growth
Tobacco	Retarded fetal growth; miscarriage, still birth; infant mortality
MATERNAL ILLNESS	
HIV/AIDS	Retarded growth; microcephaly; mental retardation; mother-to-child transmission
Rubella	During embryonic period, causes blindness and deafness; in first and second trimesters, brain damage
ENVIRONMENTAL POLLUTANTS	
Lead and mercury	Spontaneous miscarriage; preterm labor; brain damage
Radiation	Retarded growth; microcephaly; mental retardation; skeletal anomalies; cataracts

Sources: Moore & Persaud (2016); Sadler (2015); Weinhold (2009).



Betty Udesen/KRT/Newscom

PHOTO 2.12: Prescription and Nonprescription Drugs

Fetal alcohol syndrome is associated with distinct facial characteristics, growth deficiencies, and deficits in intellectual development, language, motor coordination, and the combined abilities to plan, focus attention, problem solve, and use goal directed behavior that persist throughout childhood and into adulthood.

prescribed thalidomide to prevent morning sickness. However, it was found that taking thalidomide 4 to 6 weeks after conception (in some cases, even just one dose) caused deformities of the child's arms and legs, and, less frequently, damage to the ears, heart, kidneys, and genitals (Laughton, Cornell, Boivin, & Van Rie, 2012; Vargesson, 2009). Nonprescription drugs, such as diet pills and cold medicine, can also cause harm, but research on OTC drugs lags far behind research on prescription drugs, and we know little about the teratogenic effect of many OTC drugs (Cabbage & Neal, 2011).

Alcohol

An estimated 14% to nearly 30% of pregnant women report consuming alcohol during their pregnancies (Arria et al., 2006; Meschke, Holl, & Messelt, 2013; Zhao et al., 2012). Indeed, alcohol abuse during pregnancy has been identified as the leading cause of developmental disabilities (O'Leary et al., 2013; Warren, Hewitt, & Thomas, 2011). **Fetal alcohol spectrum disorders** refer to the continuum of effects of exposure to alcohol, which vary with the timing and amount of exposure (Riley, Infante, & Warren, 2011). At the extreme end of the spectrum is **fetal alcohol syndrome (FAS)**, a cluster of defects appearing after heavy prenatal exposure to alcohol that is detected in 2 to 7 infants per 1,000 births (May et al., 2014; Thomas, Warren, & Hewitt, 2010). FAS is associated with a distinct pattern of facial characteristics (such as small head circumference, short nose, small eye opening, and small midface), pre- and postnatal growth deficiencies, and deficits in motor coordination, language, and cognitive development, including the combined abilities to plan, focus attention, problem solve, and use goal-directed behavior (Jirikowic, Gelo, & Astley, 2010; Mattson, Crocker, & Nguyen, 2011; Thomas et al., 2010). The effects of exposure to alcohol within the womb persist throughout childhood and have been found to be associated with deficits in learning and memory in early adulthood (Coles et al., 2011; McLachlan, Roesch, Viljoen, & Douglas, 2014; Wheeler, Kenney, & Temple, 2013).

Even moderate drinking is harmful as children may be born displaying some, but not all, of the problems of FAS, or *fetal alcohol effects* (Thomas et al., 2010). Consuming 7 to 14 drinks per week during pregnancy is associated with lower birth size; growth deficits through adolescence; and deficits in attention, memory, and cognitive development (Alati et al., 2013; J.-H. Chen, 2012; Lundsberg, Illuzzi, Belanger, Triche, & Bracken, 2015; O'Leary & Bower, 2012). Even less than one drink per day has been associated with negative effects on fetal growth (Day et al., 2002; Day & Richardson, 2004; Mariscal et al., 2006) and with deficits in cognition at 1 year of age (Lu, 2005; Testa, Quigley, & Das Eiden, 2003) and behavior problems through 5 years of age (Flak et al., 2014). Scientists have yet to determine whether there is a safe level of drinking, but the only way to be certain of preventing alcohol-related risks is to avoid alcohol during pregnancy altogether.

Cigarette Smoking

Every package of cigarettes sold in the United States includes a warning about the dangers of smoking while pregnant. Fetal deaths, premature births, and low birth weight are up to twice as frequent in mothers who are smokers than in those who do not smoke (Juárez & Merlo, 2013). Infants exposed to smoke while in the womb are prone to congenital heart defects, respiratory problems, and sudden infant death syndrome and, as children, show more behavior problems and attention difficulties and score lower on intelligence and achievement tests (Kiechl-Kohlendorfer et al., 2010; Lee & Lupo, 2013). Moreover, maternal smoking during pregnancy shows epigenetic effects on offspring, influencing how genetic processes and pathways of growth and development unfold in childhood into late adolescence and likely beyond (Han et al., 2015; Knopik, Maccani, Francazio, & McGeary, 2012; Richmond et al., 2015). There is no safe level of smoking during pregnancy.

Marijuana

The effects of marijuana on prenatal development are not well understood. Marijuana use during early pregnancy negatively affects fetal length and birth weight (A. C. Huizink, 2013; Hurd et al., 2005; Moore & Persaud, 2016). Although some studies suggest few consistent findings from infancy through adolescence (Huizink, 2013), others link prenatal exposure to marijuana to impairments in attention, memory and cognitive skills, as well as impulsivity at ages 4 and 10 and poor achievement in adolescence (Goldschmidt, Richardson, Willford, Severtson, & Day, 2012; Gray, Day, Leech, & Richardson, 2005; A. Huizink & Mulder, 2006; Wu, Jew, & Lu, 2011). Some researchers has found that once the effects of exposure to other teratogens is controlled, marijuana does not show a teratogenic effect (Nordstrom-Klee, Delaney-Black, Covington, Ager, & Sokol, 2002; van Gelder et al., 2010). Regardless, the safest course is for pregnant women to avoid marijuana.

Cocaine and Heroin

Infants exposed to cocaine and heroin face special challenges, such as signs of addiction and withdrawal symptoms including tremors, irritability, abnormal crying, disturbed sleep, and impaired motor control. Prenatal exposure to cocaine and heroin is associated with reduced birthweight, shorter length, smaller head circumference, and impaired motor performance at birth (Frank, Augustyn, Knight, Pell, & Zuckerman, 2001). Exposure to these drugs during prenatal development influences brain development, particularly the regions associated with attention, arousal, and regulation (Behnke & Smith, 2013; Coyle, 2013; Lebel et al., 2013; Roussotte et al., 2011). At one month after birth, babies who were exposed to cocaine had difficulty regulating their arousal states and showed poor movement skills, poor reflexes, and greater excitability (Fallone et al., 2014).

Though it was once believed that cocaine- and heroin-exposed infants would suffer life-long cognitive deficits, research suggests more mixed and subtle effects (Bandstra, Morrow, Mansoor, & Accornero, 2010; Behnke & Smith, 2013; Lambert & Bauer, 2012). Prenatal cocaine exposure has a small but lasting effect on attention and behavioral control and language skills through late childhood (Lewis et al., 2011, 2013; Singer, Minnes, Min, Lewis, & Short, 2015), but it is not linked with impairments in overall development, IQ, or school readiness in toddlers, elementary school–aged children, or middle school–aged children (Accornero et al., 2011; Behnke & Smith, 2013; Goldschmidt et al., 2012; Min, Minnes, Yoon, Short, & Singer, 2014). Moreover, quality care can lessen the long-term impact of prenatal exposure to substances (Behnke & Smith, 2013; Lewis et al., 2011).

The challenge of determining the effects of prenatal exposure to drugs is that most infants exposed to illicit drugs, such as cocaine and heroin, are also exposed to other substances, including tobacco, alcohol, and marijuana (Jones, 2006; Passey, Sanson-Fisher, D'Este, & Stirling, 2014), making it difficult to isolate the effect of each drug on prenatal development. We must be cautious in interpreting findings about illicit drug use and the effects on prenatal development because there are many other contextual factors that often co-occur with substance use and also pose risks for development—including poverty, malnutrition, social isolation, stress, and diminished parental responsiveness (Bandstra et al., 2010; Bendersky & Lewis, 1999; Frank et al., 2001). For example, parents who a buse drugs tend to provide poorer quality care, a home environment less conducive to cognitive development, and parent–child interaction that is less sensitive and positive than the environments provided by other parents (Hans, 2002). Children raised by substance-abusing parents are at risk for being subjected to overly harsh discipline and lack of supervision (Burlew et al., 2012) as well as disruptions in care due to factors such as parental incarceration, inability to care for a child, and even death (e.g., from a drug overdose or drug gang violence). Disentangling the long-term effects of prenatal exposure to substances, subsequent parenting, and contextual factors is challenging. Researchers and health care providers who construct interventions must address the contextual and parenting-related risk factors to improve the developmental outlook for children exposed

to drugs prenatally (Butz et al., 2001; Calhoun, Conner, Miller, & Messina, 2015; Kilbride, Castor, Hoffman, & Fuger, 2000).

Maternal Illness

Depending on the type and when it occurs, an illness experienced by the mother during pregnancy can have devastating consequences for the developing fetus. For example, rubella (German measles) prior to the 11th week of pregnancy can cause a variety of defects including blindness, deafness, heart defects, and brain damage, but after the first trimester, adverse consequences become less likely (Santis, Cavaliere, Straface, & Caruso, 2006). Some sexually transmitted diseases, such as syphilis, can be transmitted to the fetus during pregnancy (Gomez et al., 2013; Sánchez & Wendel, 1997). Others, such as gonorrhea, genital herpes, and HIV, can be transmitted as the child passes through the birth canal during birth or though bodily fluids after birth (see the Lives in Context feature). Because some diseases, such as rubella, can be prevented with vaccinations, it is important for women who are considering becoming pregnant to discuss their immunization status with their health care provider.

Some illnesses with teratogenic effects, such as the Zika virus, are not well understood. Children born to women infected with the Zika virus are at greater risk of microencephaly (reduced head size). They may also show a pattern of defects now known as *congenital Zika syndrome*, which includes severe microcephaly characterized by partial skull collapse, damage to the back of the eye, and body deformities including joints and muscles with restricted range of motion (Centers for Disease Control and Prevention, 2017).

Environmental Hazards

Prenatal exposure to chemicals, radiation, air pollution and extremes of heat and humidity can impair development. Infants prenatally exposed to heavy metals, such as lead and mercury, whether through ingestion or inhalation, score lower on tests of cognitive ability and intelligence and have higher rates of childhood illness (Sadler, 2015; Vigeh, Yokoyama, Matsukawa, Shinohara, & Ohtani, 2014; Xie et al., 2013). Exposure to radiation can cause genetic mutations. Infants born to mothers pregnant during the atomic bomb explosions in Hiroshima and Nagasaki and after the nuclear power accident at Chernobyl displayed many physical deformities, mutations, and intellectual deficits. Prenatal exposure to radiation is associated with Down syndrome, reduced head circumference, intellectual disability, reduced intelligence scores and school performance, and heightened risk of cancer (Chang, Lasley, Das, Mendonca, & Dynlacht, 2014). About 85% of the world's birth defects occur in developing countries, supporting the role of context in influencing prenatal development directly via environmental hazards and also indirectly through the lack of opportunities and resources for education, health, and financial support (Weinhold, 2009).

MATERNAL CHARACTERISTICS AND BEHAVIORS

Teratogens—and the avoidance of them—are, of course, not the only determinants of how healthy a baby will be. A pregnant woman's characteristics, such as her age, and her behaviors during pregnancy, including nutrition and emotional well-being, also influence prenatal outcomes.

Maternal Age

U.S. women are becoming pregnant later in life than ever before. Between 1990 and 2010, the pregnancy rate for women aged 35 to 39 increased from 119 per 1,000 women to 137 per 1,000 women. During the same 20-year period, the rate for women aged 40 to

LIVES IN CONTEXT

HIV Infection in Newborns

PHOTO 2.13: Lives In Context: HIV Infection in Newborns
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The rate of mother-to-child transmission of HIV has dropped in recent years as scientists have learned more about HIV. The use of cesarean delivery as well as prescribing anti-HIV drugs to the mother during the second and third trimesters of pregnancy, and to the infant for the first six weeks of life, has reduced mother-to-child HIV transmission from more than 20% to less than 2% in the United States and Europe (Rudin, 2004; Torpey, Kabaso, et al., 2010; Torpey, Kasonde, et al., 2010).

Aggressive treatment may further reduce the transmission of HIV to newborns, and research suggests that it may even induce remission (Rainwater-Lovett, Luzuriaga, & Persaud, 2015; Pollack & McNeil, 2013; National Institute of Allergy and infectious Diseases, 2014). However, in developing countries such interventions are widely unavailable. Worldwide, mother-to-child HIV transmission remains a serious issue. For example, in Zambia, 40,000 infants acquire HIV each year (Torpey, Kasonde, et al., 2010). Treating newborns is critical, though not always possible. Worldwide, 20% to 30% of neonates with HIV develop AIDS during the first year of life and most die in infancy (United Nations Children's Fund, 2013).

Globally, breast feeding accounts for 30% to 50% of HIV transmission in newborns (Sullivan, 2003; World Health Organization, 2011). The World Health Organization (2010) recommends providing women who test positive for HIV with information about how HIV may be transmitted to their infants and counseling them not to breast feed. Yet cultural, economic, and hygienic reasons often prevent mothers in developing nations from seeking alternatives to breast feeding. For example, the widespread lack of clean water in some countries makes the use of powdered formulas dangerous. Also, in some cultures, women who do not breast feed may be ostracized from the community (Sullivan, 2003). Balancing cultural values with medical needs is a challenge.

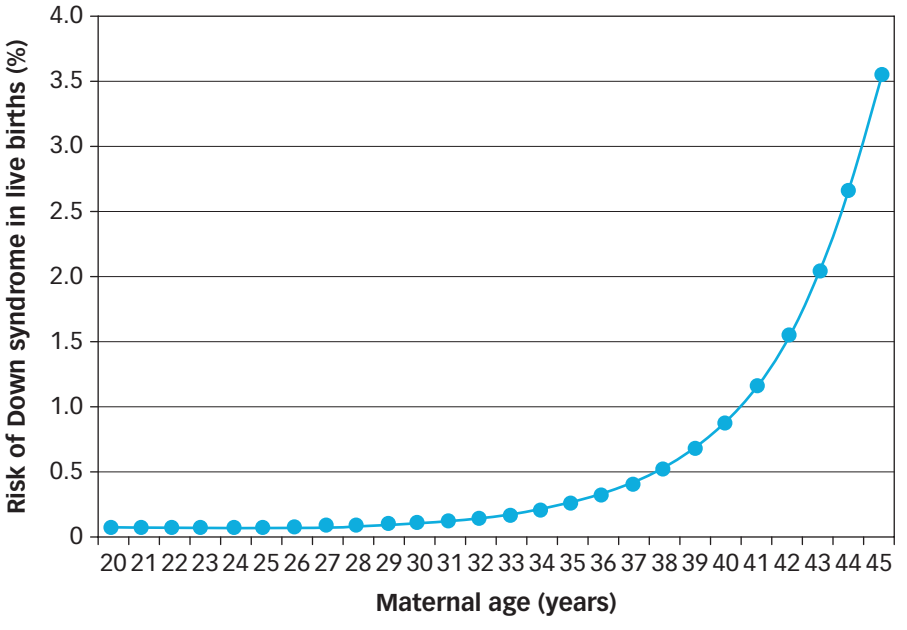
Children with HIV are at high risk for a range of illnesses and health conditions, including chronic bacterial infections; disorders of the central nervous system, heart, gastrointestinal tract, lungs, kidneys, and skin; growth stunting; neurodevelopmental delays, including brain atrophy, which contribute to cognitive and motor impairment; and delays in reaching developmental milestones (Blanchette, Smith, Fernandes-Penney, King, & Read, 2001; Laughton, Cornell, Boivin, & Van Rie, 2013; Sherr, Mueller, & Varrall, 2009; Palmer, 2003; Venkatesh et al., 2010).

What Do You Think?

1. **Imagine that you work as an HIV educator with women in an underdeveloped country. What challenges might you face in encouraging women to take steps to reduce the potential for HIV transmission to their infants? How might you help them?**

44 increased from 11 to 19 per 1,000 (Curtin, Abma, & Kost, 2015). Women who give birth past the age of 35, and especially past 40, are at greater risk for pregnancy and birth complications, including miscarriage and stillbirth, than are younger women. They are more vulnerable to pregnancy-related illnesses such as hypertension and diabetes, and their pregnancies involve increased risks to the newborn, including low birth weight, preterm birth, respiratory problems, and related conditions requiring intensive neonatal care (Grotegut et al., 2014; Kenny et al., 2013; Khalil, Syngelaki, Maiz, Zinevich, & Nicolaides, 2013). The risk of having a child with Down syndrome also increases sharply with maternal age, especially after age 40 (Hazlett et al., 2011; see Figure 2.12).

FIGURE 2.12: Maternal Age and Risk of Down Syndrome



Although the risk for Down Syndrome increases dramatically with maternal age, most infants are born healthy, regardless of maternal age.

Sources: Data from Cuckle, Wald, and Thompson (1987); figure from Newberger (2000).

Although risks for complications rise linearly with each year (Salem Yaniv et al., 2011), it is important to realize that the majority of women older than 35 give birth to healthy infants. Differences in context and behavior may compensate for some of the risks of advanced maternal age. For example, longer use of oral contraceptives is associated with a lower risk of giving birth to a child with Down syndrome (Nagy, Győrffy, Nagy, & Rigó, 2013). Older mothers tend to be healthier and show lower rates of alcohol consumption and cigarette smoking than do younger mothers (Salihu, Shumpert, Slay, Kirby, & Alexander, 2003).

Nutrition

The quality of the father's and mother's diets influences the health of the sperm and egg (Sinclair & Watkins, 2013). Most women need to consume 2,200 to 2,900 calories per day to sustain a pregnancy (Kaiser, Allen, & American Dietetic Association, 2008; Simkin, Whalley, & Keppler, 2001), but over 1 billion people in the world are chronically hungry (Food and Agriculture Organization of the United Nations, 2009) and even more are food insecure. Dietary supplements can reduce many of the problems caused by maternal malnourishment, but adequate caloric intake is crucial for healthy prenatal development (Ortolano, Mahmud, Iqbal Kabir, & Levinson, 2003).

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PHOTO 2.14: Maternal Age

Mothers who consume nutritious diets tend to have fewer complications during pregnancy and give birth to healthier babies

Some deficits resulting from an inadequate diet cannot be remedied. For example, inadequate consumption of folic acid (a B vitamin) very early in pregnancy can result in the formation of neural tube defects stemming from the failure of the neural tube to close. **Spina bifida** occurs when the lower part of the neural tube fails to close and spinal nerves begin to grow outside of the vertebrae, often resulting in paralysis. Surgery must be performed before or shortly after birth, but lost capacities cannot be restored (Scott Adzick, 2013). Another neural tube defect, **anencephaly**, occurs when the top part of the neural tube fails to close and all or part of the brain fails to develop, resulting in death shortly after birth. As researchers have learned and disseminated the knowledge that folic acid helps prevent these defects, the frequency of neural tube defects has declined to about 1 in 1,000 births (Cordero et al., 2010; Williams et al., 2015). However, in a national study of U.S. mothers, only 24% consumed the recommended dose of folic acid during pregnancy (Tinker, Cogswell, Devine, & Berry, 2010).

Emotional Well-Being

Although stress is inherently part of almost everyone's life, exposure to chronic and severe stress during pregnancy poses risks including low birth weight, premature birth, and a longer postpartum hospital stay (Dunkel, Schetter, & Tanner, 2012; Field, 2011). Maternal stress influences prenatal development because stress hormones cross the placenta, raising the fetus's heart rate and activity level. Long-term exposure to stress hormones in utero is associated with higher levels of stress hormones in newborns (Kapoor, Lubach, Ziegler, & Coe, 2016). As a result, the newborn may be more irritable and active than a

LIFESPAN DEVELOPMENT AND THE BRAIN

Pregnancy and the Maternal Brain

How does pregnancy influence mothers? The developing embryo and fetus receive a great deal of research attention, but what does pregnancy mean for mothers' development? Women's bodies undergo a radical transformation during pregnancy. For example, the hormone progesterone increases up to 15 fold and is accompanied by a flood of estrogen that is greater than the lifelong exposure prior to pregnancy. Research has shown that hormonal shifts are associated with brain changes during puberty as well as later in life. Do the hormonal changes with pregnancy influence women's brain structure? Animal research suggests that pregnancy is accompanied by neurological changes, including changes in neural receptors, neuron generation, and gene expression, that are long-lasting (Kinsley & Amory-Meyer, 2011). It is likely that pregnancy is also associated with neural changes in humans, but there is little research to date (Hillerer, Jacobs, Fischer, & Aigner, 2014).

In a recent groundbreaking study Elseine Hoekzema (2017) and colleagues conducted brain scans of women who were attempting to become pregnancy for the first time as well as their partners. Women who became pregnancy were scanned again after giving birth and again, at least two years later. The fathers and women who not become pregnant were also assessed. The new mothers experienced reductions in the brain's gray matter, signifying increased neural efficiency in regions of the brain involved in social cognition, specifically, theory of mind, which enables us to sense another person's emotions and perspective (Schurz, Radua, Aichhorn, Richlan, & Perner, 2014). Theory of mind underlies a mother's ability to interpret her infant's mental states and is important for secure parent–infant attachment and for the development of the child's own social cognitive functions (Meins, Fernyhough, Fradley, & Tuckey, 2001). The changes in gray matter volume predicted mothers' attachment to their infants in the postpartum period, as indicated by mothers' increased neural activity in response to viewing photos of their infant as compared with other infants. Other research suggests that pregnancy is associated with the enhanced ability to recognize faces, especially those displaying emotions (Pearson, Lightman, & Evans, 2009). Gestational alterations in the brain structures that are implicated in social processes may offer an adaptive advantage to mothers by facilitating her ability to recognize the needs of her child and to promote mother infant bonding. Moreover, similar to finding with animals (Kinsley & Amory-Meyer, 2011), the neural changes that accompanied pregnancy were long lasting, persistent two years after birth.

The pregnancy-related neurological changes were so marked and predictable that all of the women could be classified as having undergone pregnancy or not on the basis of the volume changes in gray matter. Notably, fathers did show a change in grey matter volume, suggesting that the neural effects of pregnancy are biological in nature rather than associated with the contextual changes that occur with the transition to parenthood.

What Do You Think?

What adaptive purpose might pregnancy-related neurological changes serve?

low-stress infant and may have difficulties in sleep, digestion, and self-regulation (Davis, Glynn, Waffarn, & Sandman, 2011; Kingston, Tough, & Whitfield, 2012). Later in childhood, he or she may have symptoms of anxiety, attention-deficit/hyperactivity disorder, and aggression (Glover, 2011). Stress in the home may make it difficult for parents to respond with warmth and sensitivity to an irritable infant (Brockington, 1996; Sameroff & Chandler, 1975). Social support can mitigate the effects of stress on pregnancy and infant care (Feldman, Dunkel-Schetter, Sandman, & Wadhwa, 2000; Ghosh, Wilhelm, Dunkel-Schetter, Lombardi, & Ritz, 2010).

Prenatal Care

Prenatal care, a set of services provided to improve pregnancy outcomes and engage the expectant mother, family members, and friends in health care decisions, is critical for the health of both mother and infant. About 26% of pregnant women in the U.S. do not seek

prenatal care until after the first trimester; 6% seek prenatal care at the end of pregnancy or not at all (U.S. Department of Health and Human Services, 2014). Inadequate prenatal care is a risk factor for low birth weight and preterm births as well as infant mortality during the first year (Partridge, Balayla, Holcroft, & Abenhaim, 2012). In addition, use of prenatal care predicts pediatric care throughout childhood, which serves as a foundation for health and development throughout the lifespan (Handler et al., 2003).

Why do women delay or avoid seeking prenatal care? A common reason is the lack of health insurance (Maupin et al., 2004). Although government-sponsored health care is available for the poorest mothers, many low-income mothers do not qualify for care, or lack information on how to take advantage of care that may be available. Other barriers to seeking prenatal care include difficulty in finding a doctor, lack of transportation, demands of caring for young children, ambivalence about the pregnancy, depression, lack of education about the importance of prenatal care, lack of social support, and poor prior experiences in the health care system, and family crises (Daniels, Noe, & Mayberry, 2006; Heaman et al., 2015; Mazul, Salm Ward, & Ngui, 2016).

Moreover, there are significant ethnic and socioeconomic disparities in prenatal care. Inadequate prenatal care is most likely among Native American women (23%), followed by African American (19%), Latino (17%), Asian American (14%), and white American women (13%; U.S. Department of Health and Human Services, 2013). African American women, in particular, are far more likely than all other groups to give birth to low birth weight or preterm infants (U.S. Department of Health and Human Services, 2014). Ethnic differences are thought to be largely influenced by socioeconomic factors, as the ethnic groups least likely to seek early prenatal care are also the most economically disadvantaged members of society.

Although prenatal care predicts better birth outcomes, cultural factors also appear to protect some women and infants from the negative consequences of inadequate prenatal care. In a phenomenon termed the *Latino paradox,* Latino mothers, despite low rates of prenatal care, tend to experience low birth weight and mortality rates below national averages. These favorable birth outcomes are striking because of the strong and consistent association between socioeconomic status and birth outcomes, and because Latinos as a group are among the most socioeconomically disadvantaged ethnic populations in the United States (McGlade, Saha, & Dahlstrom, 2004; Ruiz, Hamann, Mehl, & O'Connor, 2016).

Several factors are thought to account for the Latino paradox, including strong cultural support for maternity, healthy traditional dietary practices, and the norm of selfless devotion to the maternal role (*marianismo;* Fracasso & Busch-Rossnagel, 1992; McGlade et al., 2004). These protective cultural factors interact with strong social support networks and informal systems of health care among Latino women, in which women tend to take responsibility for the health needs of those beyond their nuclear households. Mothers benefit from the support of other family members such as sisters, aunts, and other extended family. In this way, knowledge about health is passed down from generation to generation. There is a strong tradition of women helping other women in the community and warm interpersonal relationships, known as *personalismo,* are highly valued (Fracasso & Busch-Rossnagel, 1992; McGlade et al., 2004).

Although these cultural factors are thought to underlie the positive birth outcomes seen in Latino women, they appear to erode as Latino women acculturate to American society: The birth advantage has been found to decline in subsequent U.S.-born generations. Recent findings have called the existence of the Latino paradox into question, as some samples have illustrated that socioeconomic disadvantage cannot be easily ameliorated by cultural supports (Hoggatt, Flores, Solorio, Wilhelm, & Ritz, 2012; Sanchez-Vaznaugh et al., 2016).

Thinking in Context 2.5

1. Referring to Bronfenbrenner's bioecological model (see Chapter 1), identify factors at each bioecological level that may influence development in the womb.
2. Imagine that you are a health care provider conferring with a woman who is contemplating becoming pregnant. Give some examples of specific advice you would offer to help her promote a healthy pregnancy and baby.

Apply Your Knowledge

Dr. Preemie is conducting a research study of the prevalence and correlates of drug use in college students. Because of the sensitive nature of the research topic, Dr. Preemie promises her participants confidentiality. Each college student who participates completes a set of surveys and an interview about his or her lifestyle and drug use habits. One participant, Carrie, reveals that she engages in moderate to heavy drug use (i.e., drinks two to four alcoholic beverages each day, and smokes marijuana several times per week). During the interview, Carrie mentions that she's feeling nauseous. Concerned, Dr. Preemie asks, "Do you want to stop the interview and go to the campus medical center?" "No," Carrie replies, "It's just morning sickness. I'm pregnant." "Oh," says Dr. Preemie, who nods, and continues with the interview.

Afterward, in her office, Dr. Preemie is torn and wonders to herself, "I'm worried about Carrie. Drugs and alcohol disrupt prenatal development, but I promised confidentiality. I can't tell anyone about this! Should I say something to Carrie? I'm supposed to be nonjudgmental! Intervening might keep other students from participating in my research, for fear that I'd break my promises. I don't know what to do."

1. What are the effects of teratogens, like drugs and alcohol, on prenatal development?
2. Describe the course of prenatal development. How do the effects of exposure to teratogens change during prenatal development?
3. Consider Dr. Preemie's dueling obligations. As a researcher, is she is responsible to Carrie as a participant in her study? Is Dr. Preemie responsible to the developing fetus? Her institution? Do Dr. Preemie's actions have any ramifications for the other participants in her study? How might these responsibilities conflict?
4. What should Dr. Preemie do?
5. How might your response change if Carrie were smoking cigarettes rather than using alcohol and drugs? What are the effects of smoking on prenatal development?

CHAPTER 2

2.1 Describe the process of cell reproduction and patterns of genetic inheritance.

SUMMARY

Most cells in the human body reproduce through mitosis, but sex cells reproduce by meiosis, creating gametes with 23 single, unpaired chromosomes. Some genes are passed through dominant–recessive inheritance, in which some genes are dominant and will always be expressed regardless of the gene it is paired with. Other genes are recessive and will only be expressed if paired with another recessive gene. When a person is heterozygous for a particular trait, the dominant gene is expressed and the person remains a carrier of the recessive gene. Incomplete dominance is a genetic inheritance pattern in which both genes influence the characteristic. Polygenic traits are the result of interactions among many genes. Some traits are determined by genomic imprinting, determined by whether it is inherited by the mother or the father.

KEY WORDS

chromosomes
deoxyribonucleic acid (DNA)
mitosis
meiosis
zygote
dizygotic (DZ) twins
Monozygotic (MZ) twins
homozygous
heterozygous
dominant–recessive inheritance
Incomplete dominance
sickle cell trait
polygenic inheritance
genomic imprinting

THINKING IN CONTEXT

1. Why do twins occur? From an evolutionary developmental perspective, does twinning serve an adaptive purpose for our species? Why or why not?
2. Consider your own physical characteristics, such as hair and eye color. Are they indicative of recessive traits or dominant ones?
3. Do you think that you might be a carrier of recessive traits? Why or why not?

2.2 Define and provide examples of genetic disorders and chromosomal abnormalities.

SUMMARY

PKU is a recessive disorder that occurs when both parents carry the allele. Disorders carried by dominant alleles, such as Huntington's disease, are expressed when the individual has a single allele. Some recessive genetic disorders, like the gene for hemophilia are carried on the X chromosome. Males are more likely to be affected by X-linked genetic disorders, such as hemophilia. Fragile X syndrome is an example of a dominant recessive disorder carried on the X ch romosome. Because the gene is dominant, it must appear on only one X chromosome to be displayed. Klinefelter syndrome occurs in males born with an extra X chromosome (XXY) and Jacob's syndrome occurs when males have an extra Y chromosome (XYY). Females are diagnosed with triple X syndrome when they are three X chromosomes and Turner syndrome when they are born with only one X chromosome. The most common chromosome disorder is trisomy 21, known as Down syndrome.

KEY WORDS

Phenylketonuria (PKU)
fragile X syndrome
Down syndrome
mutations

THINKING IN CONTEXT

1. Discuss how PKU illustrates the following two themes in human development: (1) the role of nature and nurture in development and (2) interactions among domains of development.
2. Identify risk factors for genetic and chromosomal disorders. What can prospective parents do to minimize the risks? What specific advice do you give?
3. Suppose you are a 36-year-old woman pregnant with your first child. What would be the advantages and disadvantages of the four types of prenatal diagnostic testing described in Table 2.6? What information would your health care provider need in order to recommend testing appropriate for your particular case?

2.3 Explain how the dynamic interactions of heredity and environment influence development.

SUMMARY

Behavioral genetics is the field of study that examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors. Heritability research examines the contributions of the genotype in determining phenotypes but also provides information on the role of experience through three types of studies: selective breeding studies, family studies, and adoption studies. Genetics contributes to many traits, such as intellectual ability, sociability, anxiety, agreeableness, activity level, obesity, and susceptibility to various illnesses.

Passive, evocative, and active gene–environment correlations illustrate how traits often are supported by both our genes and environment. Gene-environment interactions illustrate the ways that heredity and environment influence each other. Reaction range refers to the idea that there is a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints. Some traits illustrate canalization and require extreme changes in the environment to alter their course. The epigenetic framework is a model for understanding the dynamic ongoing interactions between heredity and environment whereby the epigenome's instructions to turn genes on and off throughout development are influenced by the environment.

KEY WORDS

genotype
phenotype
Behavioral genetics
Heritability
Gene–environment interactions
range of reaction
canalization
Gene–environment correlation
niche-picking
epigenetic framework

THINKING IN CONTEXT

To answer the following questions, begin by thinking about how your own development reflects interactions among your genes and sociocultural context. Then, describe a skill, ability, or hobby in which you excel.

1. How might a passive gene–environment correlation account for this ability? For example, in what ways has the context in which you were raised shaped this ability?
2. In what ways might this ability be influenced by an evocative-genetic-environment correlation?
3. Provide an example of how this ability might reflect an active gene–environment correlation.
4. Which genetic-environment correlation do you think most accurately accounts for your skill, ability, or hobby?
5. How might you apply the epigenetic framework to account for your ability?

2.4 Discuss the stages of prenatal development, stages of childbirth, and challenges for infants at risk.

SUMMARY

The germinal period is a time of rapid cell division. The embryonic period, from weeks 2 to 8, is a period of rapid cell differentiation. From 9 weeks until birth, the fetus grows rapidly, and the organs become more complex and begin to function. At about the 166th day after conception, the placenta releases a hormone that triggers the onset of labor. The first stage of labor begins when the mother experiences regular uterine contractions that cause the cervix to dilate so that the fetus's head can pass through. Delivery occurs during the second stage, and the placenta is expelled during the third stage. At birth, low birth weight infants often experience difficulty breathing and are at high risk for mortality. Low birth weight infants experience higher rates of sensory, motor, and language problems, learning disabilities, behavior problems and deficits in social skills into adolescence. The long-term outcomes of low birth weight vary considerably and depend on the environment in which the children are raised.

KEY WORDS

ovum
germinal period
Cell differentiation
blastocyst
embryo
Implantation
placenta
embryonic period
neural tube
indifferent gonad
lanugo
vernix caseosa
cesarean section
Apgar scale
preterm
small for date
low birth weight
kangaroo care

Test your understanding of the content. Review the flashcards and quizzes at **edge.sagepub.com/kuthercontext**

THINKING IN CONTEXT

1. Petra noticed that her abdomen has not grown much since she became pregnant 3 months ago. She concluded that the fetus must not undergo significant development early in pregnancy. How would you respond to Petra?
2. Parents' decisions about childbirth reflect their knowledge about birth options as well as cultural values. Referring to Bronfenbrenner's bioecological model (see Chapter 1), identify factors at each bioecological level that may influence childbirth. For example, how might neighborhood factors influence birth options? Culture?
3. Thinking of how society and medical science have changed in recent decades, in what ways might recent cohorts of parents differ from prior cohorts? What implications might these differences hold for prenatal development and childbirth?

2.5 Identify the principles of teratology, types of teratogens, and ways that teratogens can be used to predict prenatal outcomes.

SUMMARY

Teratogens include diseases, drugs, and other agents that influence the prenatal environment to disrupt development. Generally, the effects of exposure to teratogens on prenatal development vary depending on the stage of prenatal development and dose. There are individual differences in effects, different teratogens can cause the same birth defect, a variety of birth defects can result from the same teratogen, and some teratogens have subtle effects that result in developmental delays that are not obvious at birth or not visible until many years later. Prescription and nonprescription drugs, maternal substance use, material illness, and environmental factors can potentially harm the developing fetus.

KEY WORDS

teratogen
Fetal alcohol spectrum disorders
fetal alcohol syndrome (FAS)
Spina bifida
anencephaly

THINKING IN CONTEXT

1. Referring to Bronfenbrenner's bioecological model (see Chapter 1), identify factors at each bioecological level that may influence development in the womb.
2. Imagine that you are a health care provider conferring with a woman who is contemplating becoming pregnant. Give some examples of specific advice you would offer to help her promote a healthy pregnancy and baby.